Casos Clínicos

001 - PREGNANCY AND LACTATION-ASSOCIATED OSTEOPOROSIS: AN OFTEN OVERLOOKED CONDITION

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Background: Pregnancy and lactation-associated osteoporosis (PLO) is a rare condition, characterized by atraumatic fractures, mainly vertebral, occurring during the third trimester of pregnancy or the early postpartum period. Its pathophysiology remains unclear. We present two cases of patients with PLO.
Cases report: The first case involves a previously healthy 38-year-old woman who developed sudden-onset, non-traumatic dorsolumbar pain three months postpartum after her first pregnancy. Two months later, she was diagnosed with multiple fragility fractures of the dorsal and lumbar vertebrae. Vertebroplasty was performed at D11 and L1, and she was managed with analgesia. The bone densitometry revealed a L1-L4 lumbar Z-score of -2.7. Secondary causes of osteoporosis were excluded, including primary hyperparathyroidism, Cushing's syndrome, thyroid disorders, anorexia, malabsorption syndromes, and medications. She was advised to stop breastfeeding and started on teriparatide 30 µg/day, along with calcium and vitamin D supplementation.

The second case involves a previously healthy 21-year-old woman who had persistent atraumatic dorsolumbar pain after her first pregnancy. Eight years later, four months after her second childbirth, she experienced worsening dorsal pain. A CT scan revealed multiple fragility vertebral fractures, including four old dorsal fractures and a new fracture at the ninth dorsal vertebra, with significant dorsal kyphosis. The bone densitometry revealed a L1-L4 lumbar Z-score of -3.6. Besides vitamin D deficiency, no secondary causes of osteoporosis were identified. She was advised to stop breastfeeding and was treated with alendronate 70mg/week, alongside calcium and vitamin D supplementation, which she took irregularly. As a result, eighteen months later, she sustained a new fragility lumbar fracture, and a switch to zoledronic acid 5 mg/year was made.
Conclusions: Increased awareness of PLO is crucial for timely diagnosis and management, while randomized controlled trials are needed to further understanding and treatment strategies.

018 - ENTRE ARTICULAÇÕES E PERICÁRDIO: O PAPEL DO ANAKINRA NA ARTRITE REUMATOIDE

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Introdução: A pericardite é a manifestação cardíaca mais comum da artrite reumatoide (AR), presente em 30 a 50% dos casos. Embora geralmente assintomática e autolimitada, pode evoluir para formas incessantes ou recorrentes, refratárias ao tratamento convencional.
Descrição de caso: Mulher de 50 anos, fumadora, sem antecedentes relevantes, com diagnóstico recente de AR seropositiva (FR 65U/mL e anti-CCP 221UI/mL) não erosiva, com ANA positivos (1/160 homogéneo) e anti-ENA negativos, sob prednisolona (PDN) 7.5mg/d em redução gradual e metotrexato 10mg/semana.

Cinco meses após o diagnóstico, recorreu ao serviço de urgência por dor torácica pleurítica, apresentando desnivelamento do segmento PR, derrame pericárdico ligeiro, PCR 4.98mg/dL e marcadores miocárdicos normais. Diagnosticada pericardite aguda, tendo iniciado colchicina 0.5 mg 2x/dia e ácido acetilsalicílico (AAS) 750 mg 3x/dia com redução gradual. Após melhoria inicial, apresentou recorrência da dor ao fim de 10 dias, tendo sido excluídas complicações e ajustada dose do AAS. Cerca de um mês depois, novo recrudescimento da dor torácica após suspensão do AAS, com derrame pericárdico mínimo e perturbação da motilidade septal. Face a pericardite incessante, reiniciou colchicina e AAS em fulll dose (1000mg 3x/dia), mantendo dor torácica residual. Angio-TC coronária excluiu doença aterosclerótica significativa. Ecocardiogramas subsequentes não evidenciaram derrame ou sinais de constrição, com função biventricular preservada. Simultaneamente, observou-se agravamento da atividade articular com redução da corticoterapia, com necessidade de anti-inflamatório e aumento da PDN para 10 mg/dia. O metotrexato foi titulado até 25mg/semana, com resposta parcial. Dada a pericardite incessante e refratária a tratamento convencional em doente com AR, optou-se por iniciar anakinra 100mg/dia subcutâneo, após discussão multidisciplinar. Observou-se resposta clínica significativa, permitindo suspensão do AAS e redução da PDN. A doente cessou tabagismo e mantém

estabilidade clínica, com plano de suspensão da colchicina após seis meses de controlo sustentado.

Discussão/conclusão: A pericardite na AR pode evoluir para formas incessantes (≥4-6 semanas e <3 meses) ou recorrentes em até 50% dos casos, com risco aumentado de constrição pericárdica e tamponamento.

A terapêutica inicial inclui AAS ou AINEs associados a colchicina. Corticoides são segunda linha, devendo ser introduzidos com cautela devido ao risco dose-dependente de recorrência; doses baixas (PDN 0.2-0.5mg/kg/dia) são consideradas seguras e indicadas em doenças imunomediadas. Em casos refratários, imunossupressores como imunoglobulinas endovenosas, azatioprina e agentes inibidores da IL1 podem ser considerados. O anakinra, antagonista do recetor de IL-1, demonstrou eficácia na pericardite incessante/recorrente, com rápido alívio sintomático, suspensão da corticoterapia, redução das recorrências e, em alguns casos, reversão da constrição pericárdica. Apesar da eficácia modesta na AR face a outros biológicos, é útil em casos refratários ou com envolvimento relevante da via IL-1. Neste caso, permitiu controlar simultaneamente a pericardite e a atividade articular, reforçando a importância de uma abordagem multidisciplinar e individualizada na gestão das manifestações sistémicas da AR.

020 - TOCILIZUMAB AS A TREATMENT OPTION IN SJOGREN SYNDROME WITH ASSOCIATED LYMPHOCYTIC INTERSTITIAL PNEUMONIA

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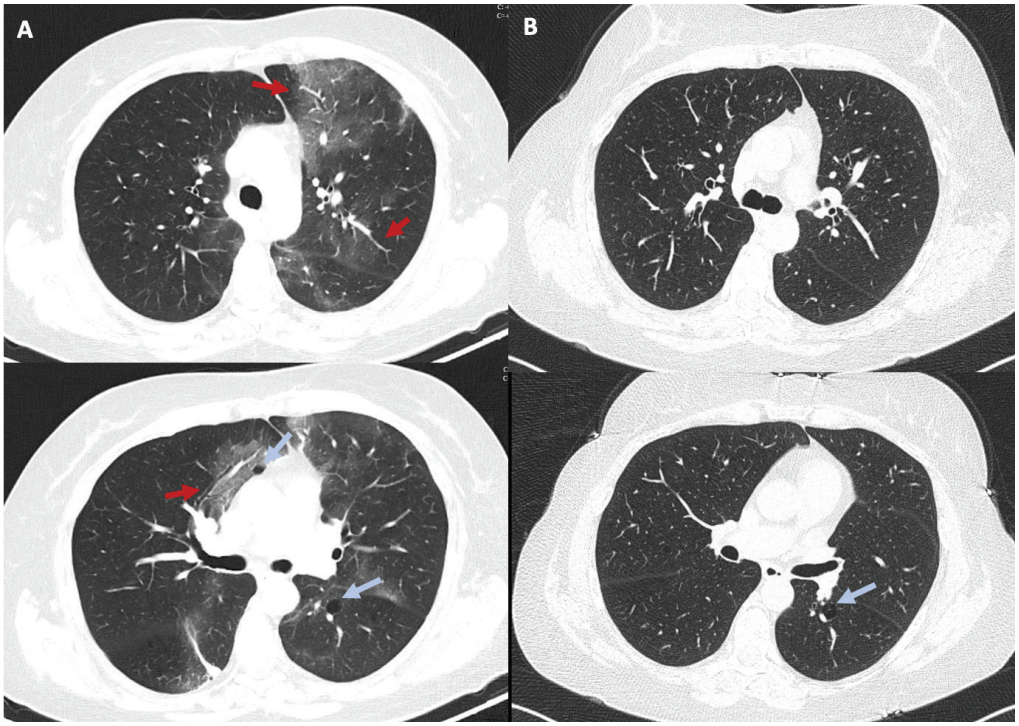
Presentation: A 64-year-old woman with rheumatoid arthritis and Sjogren Syndrome (SS) was followed in a rheumatology clinic for 15 years, treated with rituximab and methotrexate.

She was admitted in the rheumatology department with a history of daily fever and productive cough for over 2 months. She denied skin rashes, myalgia, genitourinary or gastrointestinal symptoms. Blood tests revealed anemia, lymphopenia, and elevated c-reactive protein (135mg/L), erythrocyte sedimentation rate (108mm/h) and ferritin levels (1054ng/mL), with normal kidney and liver function. Rituximab



020 – Figure 1. A - Computed tomography of the chest at the beginning of follow-up showing multilobar ground-glass opacities (red arrow) with lobular septa thickening and localized crazy paving. Scattered cysts are visible (blue arrow). B - Chest tomography of the chest after 3 months of tocilizumab showing complete resolution of the ground-glass opacifications. Some cysts are still present (blue arrow).

was stopped due to a suspected infection, but extensive tests excluded viral, bacterial, mycobacterial, and fungal infections. A chest computed tomography (CT) revealed multilobar ground-glass opacifications with lobular septa thickening and small cysts (figure 1, A). The bronchoalveolar lavage (BAL) had intense lymphocytosis (58%) and no evidence of infection. She was started on 40mg of prednisolone with a transitory improvement but with recrudescence of fever after 4 days. A lung biopsy was performed and showed septal thickening with lymphoid proliferation, consistent with lymphocytic interstitial pneumonia (LIP). Given the minimal response to a high dose of prednisolone and the evident inefficacy of rituximab, tocilizumab (TCZ) 162mg/weekly was started. Three months later, there was a major improvement of the clinical and biochemical features and chest CT showed a complete resolution of the previous abnormalities (figure 1, B).

Discussion: LIP is a rare, benign lymphoproliferative disease, often linked to SS. Clinical manifestations are unspecific and chest CT frequently shows cysts and ground-glass opacities, but definitive diagnosis requires a lung biopsy, particularly in immunosuppressed patients, where other differential diagnosis must be excluded. There are no trials in treatment of LIP, and corticosteroids and other immunosuppressors are commonly used but have a variable response. This is the first reported case of a successful treatment with TCZ, positioning it as a valuable treatment option in LIP.

022 - PLEUROPERICARDIAL SEROSITIS AS A MANIFESTATION OF RHEUMATOID ARTHRITIS REACTIVATION

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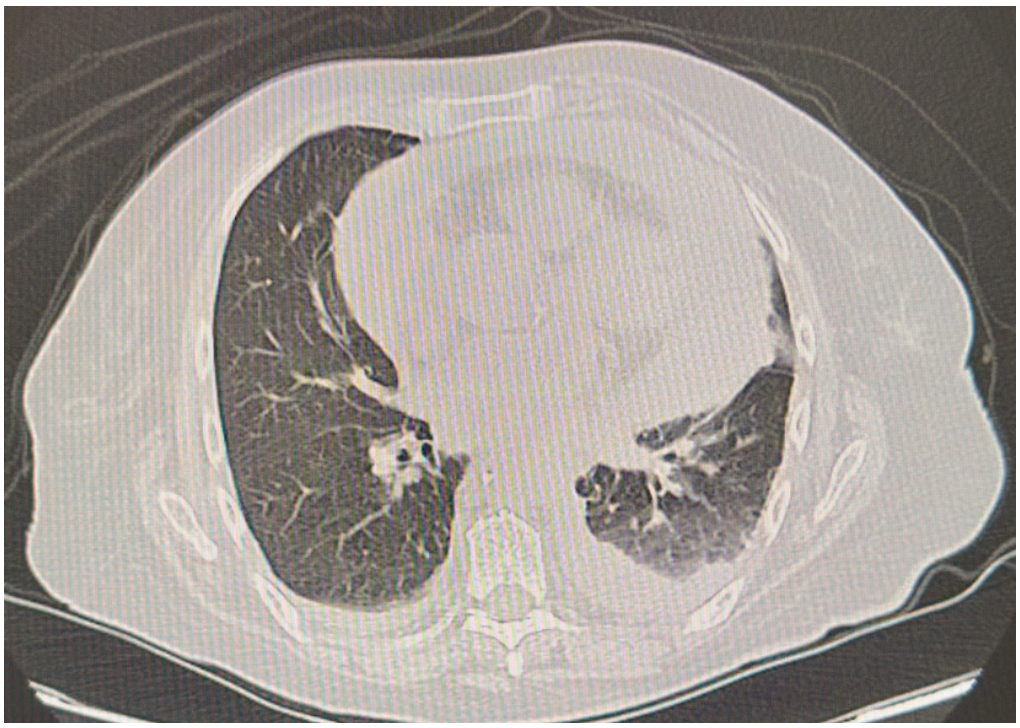
Introduction: Rheumatoid arthritis (RA) is a chronic autoimmune disease primarily affecting synovial joints, but it may also manifest with systemic, potentially severe extra-articular involvement. Among these, pleural and pericardial involvement is uncommon and typically occurs in patients with long-standing disease. In the absence of active joint activity, the diagnosis of RA in a patient presenting with serositis can be challenging.

Case description: A 75-year-old Caucasian woman presented with a 3-week history of progressive fatigue and pleuritic chest pain. The patient denied dyspnea or syncope. Her medical history included a seropositive, nonerosive RA diagnosed in 2007, for which she had discontinued follow-up and immunosuppressive therapy approximately one year earlier.

On examination, she appeared fatigued, with re-



022 – Figure 1. Axial thoracic CT showing bilateral pleural effusion and circumferential pericardial effusion

duced breath sounds at both lung bases. Musculoskeletal examination revealed no joint swelling, tenderness, or limited range of motion. A single fever peak of 38.1ºC was noted during hospitalization. Laboratory tests showed normocytic anemia (Hb 10.1 g/dL), leukocytosis (14,500/µL), thrombocytosis (572,000/µL), and markedly elevated CRP (131 mg/L). Cardiac biomarkers were mildly elevated: high-sensitivity troponin T at 0.026 ng/mL and BNP at 1292 pg/mL. Complement levels were normal. Immunological testing revealed persistently positive rheumatoid factor (341 IU/mL) and ACPA (183–253 U/mL), both at high titers. ANA, ENA, and ANCA were negative. Extensive infectious serologies and malignancy screening were unremarkable. Thoracic CT angiography (Figure 1) revealed moderate bilateral pleural effusion with partial lower lobe atelectasis and a circumferential pericardial effusion with pericardial thickening. There were no signs of tamponade on examination or echocardiography.

The overall presentation and imaging findings were consistent with pleuropericardial serositis in the context of RA reactivation. The patient was started on prednisolone 40 mg/day, with rapid symptomatic improvement. Methotrexate was reintroduced as maintenance therapy. Follow-up imaging showed complete resolution of the effusion.

Conclusion: Serosal surface involvement is a recognized but uncommon extra-articular feature of RA, particularly when occurring in isolation. In this case, the absence of joint activity contributed to diagnostic delay. Although the therapeutic response was nonspecific, the diagnosis was supported by high-titer seropositivity, a compatible clinical history, and thorough exclusion of alternative etiologies. This case highlights the importance of considering RA in the differential of polyserositis, especially in seropositive patients recently withdrawn from immunosuppressive therapy.

027 - ANTI-ELASTASE ANCA-ASSOCIATED VASCULITIS AS GRANULOMATOSIS WITH POLYANGIITIS

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Background: Granulomatosis with polyangiitis (GPA) is an antineutrophil cytoplasmic antibody (ANCA)-associated vasculitis (AAV) that primarily affects small to medium-sized vessels and is commonly associated with the presence of anti-protease 3 (anti-PR3) antibodies. However, other recognized antigen targeted by ANCA, anti-elastase, has been reported in patients with AAVs but its clinical significance remains unclear.

Case Presentation: We report the case of a 38-year-old male patient who had been suffering from yellowish nasal discharge and persistent nasal congestion for 3 years, as well as intermittent epistaxis, with no seasonal variation. He had been treated with short cycles of topical and oral corticosteroids, with partial improvement of his symptoms. He also had been treated with several oral antibiotics but with scarce effect. There were no system-specific complaints. His past medical history included an idiopathic pericarditis 10 years before. He was not taking any chronic medication. There was no history for drug abuse, especially cocaine intake.

He was first referred to our university hospital for an ENT appointment, in which clinical examination revealed thick and dry crusts in the nasal cavities and a sinus CT scan identified a septal perforation. He was submitted to surgical cleaning of these obstructive crusts. In the procedure there was extensive anterior septal perforation and mucopurulent secretions. Biopsies were performed on nasal septum and right inferior nasal turbinate.

Histological analysis revealed ulcerated mucosal flaps with necrosis, neovascularization with eosinophilic predominant infiltrate, and an arterial vessel with parietal thickening, focal fibrinoid necrosis, luminal occlusion and focal inflammatory permeation with histiocytic cells.

Blood tests are described in table 1. Serological testing was positive for p-ANCA, with negative anti-PR3 and anti-MPO, but a positive anti-elastase.

He was diagnosed with GPA, considering the septal perforation and absence of an allergic background, especially asthma. There was also no evidence of systemic manifestations of AAV. We started induction of remission with Rituximab and oral corticosteroid. There was a great improvement of his nasal symptoms, including resolution of nasal discharge and crusts. Nasal cavity patency was restored.

Discussion: This case highlights a rare presentation of GPA confined to the upper airways, in which histopathological findings were essential to establish the diagnosis. Despite the presence of p-ANCA and anti-elastase antibodies, the combination of clinical, radiological, and histological features strongly supported this diagnosis. Anti-elastase antibodies, although previously reported in AAV, lack disease specificity and may occur in other inflammatory or infectious contexts, underscoring the importance of cautious interpretation. Notably, the patient met the 2022 ACR/EULAR classification criteria for eosinophilic granulomatosis with polyangiitis (EGPA) but not for GPA. How-

027 – TABLE 1. Laboratorial findings for diagnostic workup. ANCA were identified by imunofluoresecence. anti-PR3 and anti-MPO were determined by FEIA. Anti-elastase was determined by ELISA, although the specific unit of measurement was not provided in the laboratory report.

	Diagnosis	Follow-up	Reference Range
Creatinine	0.84	1.00	0.72 – 1.18 mg/dL
Urinalysis	No active sediment	No active sediment	-
Urinary Protein/Creatinine Ratio	41	45	< 200 mg/g
B-Type Natriuretic Peptide	5.2	-	< 100 pg/mL
CRP	0.17	0.08	< 0.50 mg/dL
ESR	6	7	1 – 20 mm/h
Leucocytes	8.8	8.1	3.9 – 10.2 G/L
Neutrophils	3.7	4.5	1.5 – 7.7 G/L
Eosinophils	1.16	0.38	0.02 – 0.50 G/L
Platelets	394	317	150 – 450 G/L
p-ANCA	Positive	-	-
c-ANCA	Negative	-	-
Anti-PR3	< 0.2	-	< 2.0 IU/mL
Anti-MPO	0.2	-	< 3.5 IU/mL
Anti-Elastase	Positive	-	-
Antinuclear antibodies	Negative	-	-
Rheumatoid Factor	< 9	-	< 20 IU/mL
IgG	12.03	9.23	5.40 – 18.22 g/L
IgA	2.49	1.71	0.63 – 4.84 g/L
IgM	0.56	0.31	0.22 – 2.40 g/L
IgE	< 16	-	< 100 IU/mL
HIV (serology)	Negative	-	-
HCV (serology)	Negative	-	-
HBV (serology)	Immune	-	-
IGRA	Negative	-	-
Parasitological faecal culture	Negative	-	-
Nasal mucosa culture	S. aureus*	-	-

c-ANCA: cytoplasmic antineutrophil cytoplasmic antibody; anti-PR3: anti-protease 3; anti-MPO: Anti-myeloperoxidase; IGRA: interferon-gamma release assay; HIV: human immunodeficiency virus; HBV: hepatitis B virus; HCV: hepatitis C virus; Ig, immunoglobulin. * S. aureus was interpreted as colonization

ever, the absence of asthma or peripheral eosinophilia and the presence of nasal septum perforation, which is exceedingly rare in EGPA, favored the diagnosis of GPA. This case reinforces the need for an integrated diagnostic approach in limited AAV forms, particularly when serological findings are atypical.

028 - NECROSE AVASCULAR DA CABEÇA DO FÉMUR – DIAGNÓSTICO PRECOCE ... E DEPOIS?

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Introdução: A necrose avascular da cabeça do fémur (NACF) deve-se à disrupção da perfusão sanguínea proximal do fémur, podendo resultar em afundamento da cabeça do fémur e limitação funcional severa. Nenhum tratamento médico é 100% eficaz, sendo por vezes necessária cirurgia, nomeadamente colocação de prótese total da anca (PTA). O diagnóstico precoce permite tratamento médico atempado, podendo evitar a necessidade de PTA. O diagnóstico, no entanto, exige elevada suspeição clínica, visto a dor na bacia ser muitos vezes desvalorizada e/ou assumida como

em relação com o excesso ponderal, atividade laboral, etc. É mais frequente em doentes medicados com corticoides, mesmo em doses baixas, e em doentes com lúpus eritematoso sistémico (LES) e síndroma dos anticorpos anti fosfolipídicos (SAAF).

Casos clínicos: C1: Mulher de 40 anos, educadora de infância, com LES e SAAF 2rio, sob prednisolona (PDN) 2.5mg id, que iniciou coxalgia bilateral de predomínio direito, de caráter inflamatório, que atribuiu à profissão. A RM confirmou NACF bilateral. Suspendeu a PDN e iniciou perda ponderal, estatina e bloqueador de canais do cálcio (BCC), com remissão clínica e estabilidade imagiológica. C2: Mulher de 48 anos, cozinheira, com LES e SAAF 2rio, com episódios prévios de TVP e TEP, sob PDN 5mg id e Edoxabano. Iniciou coxalgia bilateral de caráter inflamatório, que associou à profissão. A RM confirmou NACF. Suspendeu a PDN e iniciou perda ponderal, estatina e BCC, com remissão clínica. C3: Homem de 45 anos, administrativo, com LES e SAAF 2rio, com TVP prévia, e válvula aórtica mecânica, após endocardite, sob PDN 5mg id e Varfarina. Iniciou coxalgia direita de caráter inflamatório, confirmando a RM, NACF. Não foi possível suspender a PDN, iniciando estatina, BCC e fisioterapia, que incluiu ondas de choque extracorpórea. Ponderado bifosfonato, mas dada a sua interação com a Varfarina, optou-se pelo Denosumab. Apesar destas medidas, progrediu para afundamento da cabeça do fémur e necessitou de PTA.

Discussão: Os 3 casos apresentados relembram a necessidade de não desvalorizar a coxalgia no jovem, sobretudo se com medicação e/ou patologias que predisponham à NACF. A suspeição diagnóstica pode resultar em diagnóstico precoce e este pode conduzir a medidas médicas que, em conjunto, podem impedir a progressão da NACF. Realçam também a inexistência de tratamento médicos inteiramente eficazes e a importância dos corticoides e o benefício da sua suspensão precoce, quando possível.

029 - SARCOIDOSE, UM DESAFIO DIAGNÓSTICO (A PROPÓSITO DE 3 CASOS CLÍNICOS)

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Introdução: A sarcoidose é uma doença granulomatosa multissistémica, com o envolvimento intratorácico a ser o mais frequente, caracterizado por adenopatias hilares e padrão pulmonar micronodular difuso. Pode, no entanto, envolver qualquer órgão, podendo ser "silenciosa" ou ter manifestações clínicas referentes aos órgãos afetados, sendo nestes casos as manifestações comuns à de muitas outras patologias. É, assim, "uma grande imitadora". O curso da doença pode ser autolimitado, com remissão espontânea, mas também pode assumir um curso crónico. Pelo exposto, o seu diagnóstico é frequentemente um desafio, exigindo um elevado grau de suspeição.

Casos clínicos: (1) Mulher, 71 anos, avaliada por quadro consumptivo e hepatite citocolestática. Apuradas xeroftalmia, xerostomia e xerose cutânea marcadas, adenopatias mediastino-hilares, pulmão com padrão reticulo-intersticial bilateral, ECA, ANA, anti-Ro e anti-La muito elevados. Ponderado S. Sjögren complicada de doença linfoproliferativa versus sarcoidose, preenchendo critérios de S. Sjögren. Enquanto era avaliada, ocorreu remissão espontânea do quadro clínico, persistindo apenas as xeroses, estudo imune positivo e ECA elevado por períodos. Embora se suspeitasse de sarcoidose, o seu diagnóstico apenas foi obtido 10 anos depois, após repetição da biopsia hepática. (2) Homem, 57 anos, com insuficiência cardíaca inaugural. Diagnosticadas miocardite, polisserosite e adenomegálias predominantemente mediastínicas. Na altura, com estudo imune negativo e ECA sérico normal. Posteriormente, com aumento do ECA e cintigrafia com Gálio67 compatível com sarcoidose. Ocorreu remissão espontânea do quadro enquanto era investigado. (3) Mulher, que aos 25 anos iniciou episódios autolimitados de dor e relevo das tatuagens. Aos 37 anos, estes episódios passaram a acompanhar-se de artralgias de caráter inflamatório das articulações satélite às tatuagens. Ponderada sarcoidose, tinha ECA sérico normal. 2 anos depois iniciou uveíte e, nesta altura, com ECA aumentado e cintigrafia com Gálio67 compatível com sarcoidose.

Discussão: Os 3 casos apresentados ilustram a variedade de apresentações clínicas de sarcoidose, que podem ser sistémicas, cardiopulmonares, cutâneas, articulares, etc., e realçam a importância da suspeição diagnóstica. Em todos eles, a hipótese de sarcoidose foi colocada ad inicium, mas apenas foi comprovada meses ou anos depois. Por fim, referir que a remissão espontânea do quadro, não sendo usual noutras patologias, reforça a suspeita diagnóstica.

036 - OSTEOPOROSE SECUNDÁRIA A MASTOCITOSE SISTÉMICA: UM CASO CLÍNICO

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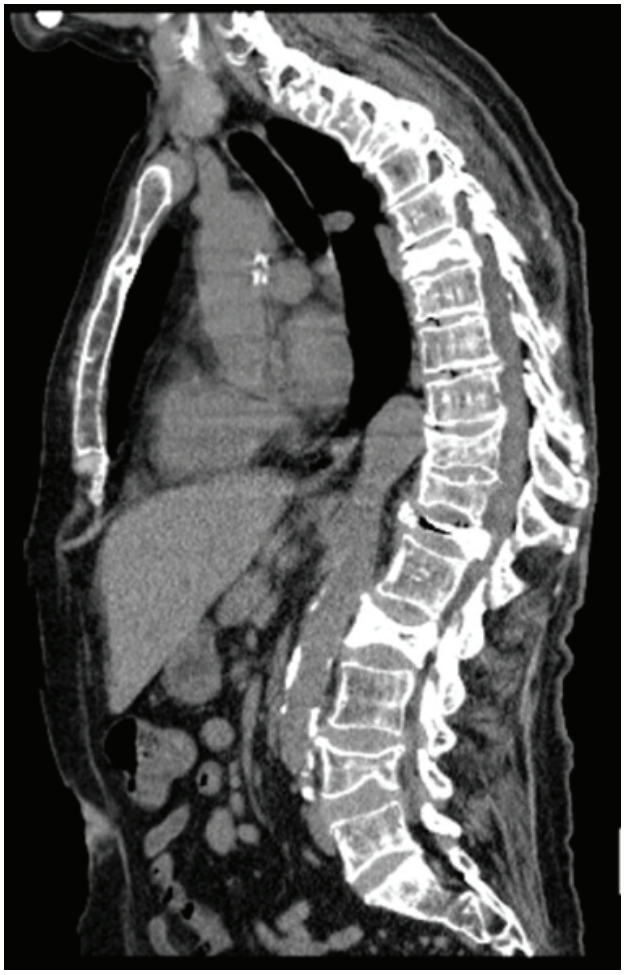

036 – Figure 1. Tomografia computarizada da coluna

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Introdução: A mastocitose sistémica (MS) é uma doença rara caracterizada pela proliferação anormal de mastócitos. A osteoporose (OP) constitui uma complicação frequente desta condição, sobretudo nas formas sistémicas, com predileção pelo osso trabecular e associação a fraturas de fragilidade óssea, constituindo um importante marcador de gravidade da doença (1, 2).
Caso Clínico: Relata-se o caso de uma mulher de 59 anos, fumadora, com diagnóstico de MS, confirmado por biópsia da medula óssea, evidenciando infiltração extensa por mastócitos, expressão do antigénio CD25, presença da mutação D816V no gene c-KIT e triptase sérica superior a 200 µg/L. Entre os sinais de gravidade, destacava-se a dor óssea e o envolvimento ósseo com múltiplas fraturas de baixo impacto, incluindo a cabeça umeral bilateral, ramos isquiopúbicos e várias vértebras (D3, D6, D10, D11, D12, L2, L4, L5) (Figura 1). Clinicamente, apresentava perda de estatura de 4 cm, hipercifose dorsal e dor à palpação das apófises espinhosas na transição dorsolombar. A densitometria óssea revelou valores compatíveis com osteopenia (T-score -2,3 e -2,1 na coluna lombar e colo do fémur, respetivamente) e os marcadores do metabolismo fosfocálcico (cálcio, fósforo, 25(OH) vitamina D, paratormona, fosfatase alcalina) encontravam-se dentro dos limites normais. Foi iniciada terapêutica com zoledronato 5 mg endovenoso mensal. Contudo, apesar da adesão terapêutica, ocorreu uma nova fratura de fragilidade óssea, pelo que se propôs a alteração do esquema terapêutico para denosumab 60 mg semestral e avapritinib 200mg/dia.
Discussão: Este caso evidencia a gravidade do envolvimento ósseo com fraturas patológicas na MS, condição que afeta até 30% dos doentes (1, 3). O tratamento precoce da OP é crucial para aumentar a densidade mineral óssea e reduzir o risco de fraturas, melhorando significativamente a qualidade de vida do doente (1, 2). Fármacos antirreabsortivos como os bifosfonatos são a primeira linha de tratamento, embora com dados de eficácia limitados (2, 4). O uso de denosumab e, mais recentemente, de inibidores da tirosina-quinase, que visam o tratamento específico da MS, nomeadamente o avapritinib, têm-se mostrado promissores, ainda que careçam de validação clínica robusta (1).
Conclusão: A OP, enquanto complicação grave da MS, exige uma vigilância clínica contínua e estratégias terapêuticas individualizadas, sobretudo perante a evidência de fraturas recorrentes, apesar do tratamento instituído.

045 - SYPHILIS AND AUTOIMMUNITY: CASE REPORT WITH POSITIVITY FOR LUPUS-SPECIFIC AUTOANTIBODY (ANTI-SM)

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Introduction: Syphilis, a systemic infection caused by Treponema pallidum, is historically known as “the great imitator” due to its ability to mimic various diseases, including autoimmune disorders, with clinical and laboratory manifestations similar to collagenoses. Systemic lupus erythematosus (SLE), in particular, can be mimicked both clinically and serologically, potentially leading to significant diagnostic and therapeutic errors. A thorough assessment of the clinical and epidemiological context is essential to avoid inappropriate

045 – TABLE 1. Comparison: Systemic Lupus Erythematosus (SLE) vs Secondary Syphilis

Category	Systemic Lupus Erythematosus (SLE)	Syphilis (emphasis on secondary form)
Etiologic Agent	Systemic autoimmune disease with multifactorial etiology.	Treponema pallidum, a spirochete bacterium.
Pathogenesis	Autoantibodies and immune complexes causing multisystem inflammation.	Chronic infection with distinct clinical stages.
Epidemiology	Young women, African descendants, Hispanics.	Both sexes, increasing incidence in risk groups.
Cutaneous Manifestations	Malar rash, discoid lupus, photosensitivity, livedo reticularis.	Palmar-plantar exanthem, "moth-eaten" alopecia.
Musculoskeletal	Symmetric, non-deforming arthritis.	Arthralgia, usually without true arthritis.
Neurological	Seizures, psychosis, peripheral neuropathy, transverse myelitis.	Neurosyphilis: meningitis, Argyll Robertson pupils, tabes dorsalis.
Cardiovascular	Pericarditis, myocarditis, Libman-Sacks endocarditis.	Syphilitic aortitis, aortic aneurysm.
Renal	Lupus nephritis (various classes), hematuria, proteinuria, nephrotic or nephritic syndrome.	Rarely directly affects the kidneys.
Hematological	Hemolytic anemia, leukopenia, lymphopenia, thrombocytopenia.	Anemia, lymphadenopathy, leukocytosis or leukopenia.
Gastrointestinal	Serositis (abdominal pain), hepatomegaly, lupus pancreatitis.	Syphilitic hepatitis, hepatic gummas, syphilitic gastritis.
Immunological Factors	Positive ANA (usually high titers), anti-dsDNA, anti-Sm, hypocomplementemia.	May have low-titer positive ANA; false-positive VDRL.
Serologies	Anti-dsDNA, anti-Sm, anti-Ro/SSA, anti-La/SSB, anti-histone, antiphospholipid antibodies.	VDRL/RPR (non-treponemal), FTA-ABS, TPPA (treponemal).
False Positives	VDRL may be false-positive in SLE due to anticardiolipin antibodies.	ANA may be false-positive in secondary syphilis.
Differential Diagnosis	Includes syphilis, HIV, viral hepatitis, rheumatologic diseases.	SLE is one of the main differentials in secondary syphilis.
Confirmatory Diagnosis	ACR/EULAR 2019 criteria (clinical + immunological), biopsies, autoantibodies.	Treponemal (FTA-ABS, TPPA) + non-treponemal (VDRL, RPR) tests, PCR, dark-field microscopy.
Treatment	Immunosuppressants (corticosteroids, hydroxychloroquine, azathioprine, mycophenolate, cyclophosphamide, belimumab).	Benzathine penicillin (early forms), aqueous penicillin G (neurosyphilis), doxycycline if allergic.
Prognosis	Variable depending on severity and treatment adherence; risk of irreversible organ damage.	Excellent with early diagnosis; sequelae possible in late stages.
Clinical Mimicry	Syphilis can mimic SLE (rash, arthralgia, serologic changes), delaying correct diagnosis.	Secondary syphilis may present as "lupus-like" with similar systemic and immunological features.

management, especially in regions experiencing a resurgence of syphilis.

Case report: A 53-year-old previously healthy woman sought medical care with a three-week history of symmetric polyarthralgia affecting small joints, associated with a non-pruritic maculopapular rash on the limbs and trunk, as well as asthenia, myalgia, and intermittent low-grade fever. On physical examination, she presented with erythematous-brownish lesions on the extensor surfaces of the limbs and mild cervical lymphadenopathy. Laboratory tests revealed an ESR of 68 mm/h, CRP of 1.34 mg/dL, ANA with fine speckled pattern at 1:640, low C3 and C4 levels, polyclonal hypergammaglobulinemia on protein electrophoresis, and positivity for anti-RNP and anti-Sm antibodies (37 U/mL). The absence of symptoms such as photosensitivity, oral ulcers, renal or neurological involvement raised diagnostic doubts. Given the clinical context and a highly positive VDRL (1:64), indicative of active infection, treatment with benzathine penicillin (7,200,000 IU, in three weekly doses) was initiated. The patient showed progressive improvement of symptoms, resolution of cutaneous and articular manifestations, normalization of inflammatory markers, and seroconversion of autoantibodies within three months.

Discussion: Syphilis can induce nonspecific immune activation, immune complex formation, and transient production of autoantibodies, including markers considered highly specific, such as anti-Sm. This phenomenon, described in case reports, is attributed to immune tolerance breakdown triggered by molecular mechanisms still under investigation, such as antigenic mimicry and polyclonal hyperstimulation. According to Firestein et al. (2021), autoantibodies like ANA,

anti-RNP, and even anti-Sm can emerge in infectious conditions, although their presence must be interpreted cautiously and always correlated with clinical findings. This case highlights the critical role of the rheumatologist in identifying infectious differential diagnoses, even in the presence of highly suggestive autoantibodies.

Conclusion: Even highly specific autoantibodies for SLE can transiently appear in infections such as syphilis. Careful clinical management prevented potentially harmful immunosuppressive treatment, underscoring the importance of multidisciplinary clinical reasoning in rheumatology practice. Case reports like this reinforce the need for broad etiological investigation before establishing an autoimmune disease diagnosis.

047 - COLONIC VOLVULUS AND INTRAVENOUS IMMUNOGLOBULIN IN SYSTEMIC SCLEROSIS: A CASE REPORT

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Background: The gastrointestinal (GI) tract is virtually always affected in systemic sclerosis (SSc), making these manifestations the most common in this orphan disease. Inflammatory, vascular, and fibrosis events result in peristaltic dysfunction, ranging from gastroesophageal reflux to major complications like pseudo-obstruction. Symptoms are primarily managed with supportive treatments. A recent systematic review of 25 SSc patients showed that intravenous immunoglobulin (IVIG) therapy may improve GI symptoms, with a favorable safety profile1.

Case report: We describe a 64-year-old female presenting a one-year history of extreme nausea, vomiting, and diarrhea, resulting in a 30kg weight loss. She reported her skin feeling thicker but denied experiencing Raynaud's phenomenon. Physical examination revealed a mRss of 36. Immunology showed positivity at a 1/160 titer with antiPL-7. Capillaroscopy showed no specific pattern. Cancer screening, including PET scan, was negative. A presumptive diagnosis of diffuse cutaneous SSc with GI involvement was made.

During follow-up, the patient continued to experience GI complaints and general malaise. 5 months after diagnosis, she reported severe vomiting and constipation. Imaging revealed intestinal occlusion due to stenosis at the splenic flexure, with colonic and small intestinal dilation, and pneumatosis. Surgery confirmed torsion of the splenic flexure, with no palpable tumoral masses. A colonic distortion procedure was performed.

Despite symptoms initially improving, they recurred 2 weeks later, and a new CT scan showed similar colonic changes.

Given the severity of GI involvement, recurrent episodes of pseudo-obstruction, and severe malnutrition, off-label IVIG therapy (2g/kg/day for 5 days) was proposed. Improvement was reported after the 3rd infusion.

Alongside non-pharmacological measures, GI symptoms were controlled, and the patient regained weight. Due to the severity of GI complaints, rapidly progressive disease, and the absence of Raynaud's phenomenon, a skin biopsy was performed, which confirmed the diagnosis of SSc. An esophageal manometry revealed hypotonicity and absent contractions, findings consistent with SSc.

Conclusion: This case highlights the complexity and severity of GI involvement in SSc, with colonic volvulus as a rare but life-threatening complication. The observed sustained improvement of GI symptoms matches systematic reviews1 and case series2 reports, supporting its efficacy in symptom relief and improving patient outcomes, even in severe manifestations like intestinal pseudo-obstruction. Lastly, it serves as a reminder to open the rationale of differential diagnosis, as the absence of Raynaud's phenomenon may challenge the initial diagnosis of SSc, which was ultimately confirmed. As more robust data is needed, this case contributes to the limited literature on severe GI manifestations in SSc and the potential of IVIG in managing refractory symptoms.

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058 - BEYOND POLYMYALGIA: AN UNEXPECTED DIAGNOSIS IN AN ELDERLY MAN

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Background: Systemic lupus erythematosus (SLE) is a heterogeneous autoimmune disease with a broad

spectrum of clinical manifestations. Although typically diagnosed in women of childbearing age, it can also present in older individuals, often with atypical or insidious symptoms that may mimic other inflammatory or degenerative conditions. Recognition in this population can be challenging, especially when musculoskeletal complaints predominate, potentially delaying diagnosis and appropriate treatment. The following report outlines a case of late-onset SLE presenting with a polymyalgia rheumatica (PMR)-like phenotype.
Clinical Report: A 62-year-old male smoker presented to the emergency department with a 3-month history of inflammatory polyarthralgia affecting the shoulders, hips and hands, with significant functional impairment. He also reported unquantified weight loss and erythematous skin lesions on the upper limbs and face. He was started on prednisolone 20 mg daily with a tapering regimen, resulting in mild improvement, and was referred to the Rheumatology outpatient clinic, where he was seen one month later.

Further history revealed previous non-scarring alopecia with an occipital patch of hair loss in the previous year, xerostomia, and Raynaud's phenomenon. On physical examination, there was no evidence of peripheral arthritis or active cutaneous lesions, but he presented with proximal muscle weakness and marked stiffness involving both the pelvic and shoulder girdles.

Laboratory investigations showed an erythrocyte sedimentation rate (ESR) of 44 mm/hour, C-reactive protein (CRP) of 0.6 mg/dL, leukopenia with lymphopenia (500/µL), positive rheumatoid factor (64.7 IU/mL), negative anti-cyclic citrullinated peptide antibodies, and positive antinuclear antibodies (ANA) with a homogeneous nuclear pattern (AC-1). He also tested positive for anti-SSA, anti-SSB, anti-nucleosome, and anti-histone antibodies, as well as anti-dsDNA antibodies (43 U/mL). Serum electrophoresis revealed a polyclonal hypergammaglobulinemia. Creatine kinase (CK) levels were within the normal range. A thoracoabdominopelvic CT scan identified a pleural effusion, with no evidence of occult malignancy.

A diagnosis of SLE was established and no medication potentially associated with drug-induced lupus was identified. Treatment with subcutaneous methotrexate and hydroxychloroquine was initiated, leading to progressive clinical improvement.
Discussion: This case highlights an atypical presentation of SLE in an elderly male, initially manifesting with inflammatory polyarthralgia and prominent proximal stiffness, initially raising the differential diagnoses of PMR and seronegative rheumatoid arthritis, particularly given the age, proximal stiffness, and absence of overt arthritis. The presence of systemic features and the autoimmune serological profile — notably lymphopenia, positive ANA with multiple extractable nuclear antigens, and anti-dsDNA positivity — were key to establishing the diagnosis.

Musculoskeletal involvement is common in SLE, but the presentation in this case, mimicking a PMR-like phenotype, is uncommon and reinforces the need for a broad differential diagnosis in older patients with inflammatory symptoms. The normal CK level helped exclude inflammatory myopathy as the cause of proximal weakness.

This case illustrates the importance of maintaining a high index of suspicion for SLE in atypical demographic settings, as timely immunosuppressive treatment can lead to meaningful clinical improvement and potentially prevent irreversible organ damage.

062 - A UTILIZAÇÃO DE IMUNOGLOBULINAS INTRAVENOSAS NUM CASO REFRATÁRIO DE DERMATOMIOSITE

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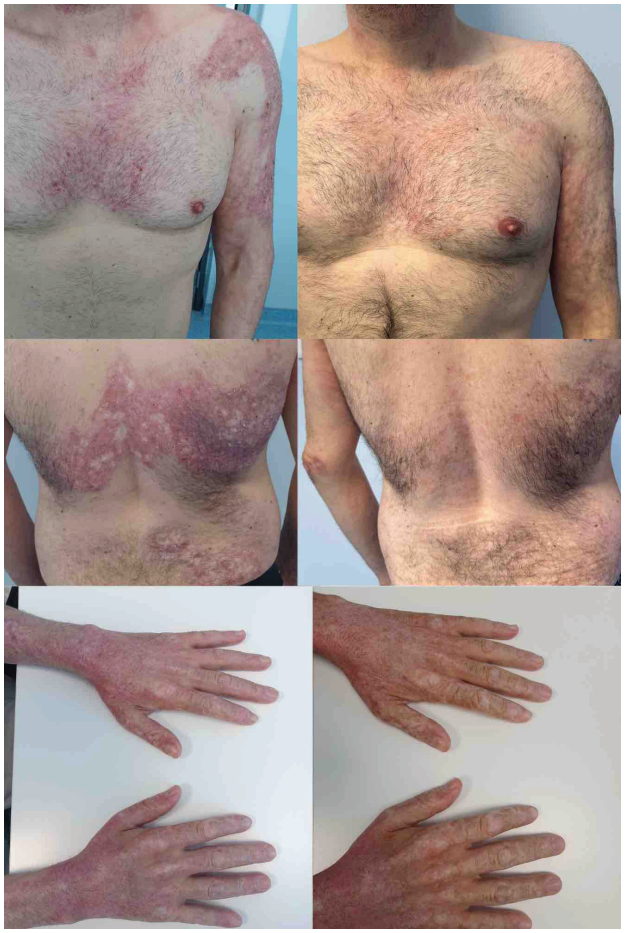
Introdução: A dermatomiosite é uma miopatia inflamatória que se caracteriza por défice muscular proximal e alterações cutâneas (nomeadamente pápulas de Gottron e rash heliotropo), com possível envolvimento de outros órgãos. Laboratorialmente é frequente o aumento das enzimas musculares e a associação a determinados autoanticorpos tem valor prognóstico.

Em 10-20% das miopatias inflamatórias em idade adulta há uma associação a neoplasia, sendo o risco maior na presença de autoanticorpos como anti-TIF1γ e anti-NXP2.
Caso clínico: Doente do sexo masculino, 42 anos, referenciado à consulta de Reumatologia em 2021 por quadro, com 5 anos de evolução, de lesões cutâneas maculopapulares fotossensíveis na face, tronco e membros superiores com extensão ao dorso das mãos e ulceração na face dorsal das articulações metacarpofalângicas, em associação a défice de força muscular proximal dos membros. Seguido previamente em consultas de Dermatologia e Medicina Interna e tinha sido medicado com tópicos, corticoterapia oral e metotrexato.

Na primeira consulta apresentava rash macular violáceo palpebral e eritematoso com áreas hipopigmentadas na região peitoral, dorso e membros superiores, incluindo na face dorsal das mãos e dos dedos, sem défice de força muscular objetivável.

Do estudo prévio tinha registo de uma biópsia cutânea de 2018 com descrição de "aspetos compatíveis com dermatomiosite". O estudo laboratorial mostrou:



062 – Figure 1. Lesões cutâneas de dermatomiosite ao diagnóstico (esquerda) e após 12 meses de tratamento com IgIV (à direita).

ligeira elevação da proteína C-reativa (5.7mg/L), enzimas musculares normais, anticorpos anti-nucleares positivos (1/100 padrão mosqueado) e painel de miopatias inflamatórias positivo para anticorpos anti-TIF1γ. A eletromiografia mostrou achados sugestivos de miopatia. O estudo paraneoplásico revelou um pico monoclonal na região das gamaglobulinas em eletroforese, tendo o doente sido referenciado a Hematologia com diagnóstico de gamapatia monoclonal de significado indeterminado (MGUS-gamapatia IgG/lambda), com indicação para vigilância.

Tinha sido medicado com metotrexato em 2019, noutro hospital, suspenso após infeção por herpes zoster. Em 2021, na consulta de Reumatologia, iniciou prednisolona 30mg id (0.5mg/kg, em esquema de desmame) e hidroxicloroquina 400mg id (suspensa por rash pruriginoso generalizado). Em agosto de 2021 iniciou rituximab (3 ciclos de 1g, 2 tomas semestrais intervaladas por 2 semanas) com resposta inicial parcial a nível cutâneo, mas com recrudescimento das queixas musculares após redução da dose de prednisolona (abaixo de 10-15mg/dia). Em março de 2023 apresentou agravamento do rash cutâneo com necessidade de corticoterapia em doses mais elevadas, tendo iniciado micofenolato de mofetil (MMF) em dose crescente até 3g/dia, sem efeito significativo a nível cutâneo. Em maio de 2024 foi decidido associar imunoglobulinas intravenosas (IgIV 2mg/kg, 5 dias consecutivos a cada 4 semanas) em associação ao MMF, e, aos 12 meses de tratamento, o doente apresentou melhoria marcada das lesões cutâneas (imagem 1), permitindo a suspensão da prednisolona e mantendo-se o doente com força muscular preservada.

Conclusão: As IgIV, através das suas ações anti-inflamatórias e imunomoduladoras, são uma alternativa terapêutica nos casos de miopatias refratárias à corticoterapia e a imunossupressores clássicos, nomeadamente no envolvimento cutâneo grave.

Apresentamos um caso desafiante de dermatomiosite com envolvimento cutâneo extenso e refratário aos glucocorticoides e imunossupressores clássicos, em associação a MGUS, que apresentou uma franca resposta às IgIV.

064 - CAN LONG-TERM OSIMERTINIB THERAPY CONTRIBUTE TO OSTEOPOROSIS?

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Introduction: Osimertinib is an epidermal growth factor receptor tyrosine kinase inhibitor (EGFR-TKI) used in the treatment of non-small-cell lung carcinoma. In-vivo mice studies have demonstrated that the EGFR signaling pathway plays an important role in bone regulation, mainly having an anabolic role in bone metabolism1. This raises the question: can EGFR inhibitors have a potential role in osteoporosis (OP) pathogenesis?
Case report: A 70-year-old female was referred from the oncological pulmonology department to our Osteoporosis clinic for evaluation of several vertebrae fractures, which carried visits to the Emergency Department. In this context, and before our first evaluation, CT-scans were performed, and over the last year, fractures in the 7th, 8th, and 12th dorsal vertebrae, as well as the 1st, 3rd, 4th, and 5th lumbar vertebrae were documented, with no lesions suggestive of bone metastasis.

In her first appointment, besides progressive height

loss, she reported several episodes of sudden-onset back pain within the past year. Prior trauma was denied. She had a medical history of lung adenocarcinoma, treated surgically and with chemotherapy. For the past six years, she had been on maintenance therapy with Osimertinib, which was effective in disease stability.

When assessing other OP potential risk factors, it was found that menopause occurred at the age of 40, for which she had taken hormone replacement therapy. Past treatments for OP were denied.

Blood workup was unremarkable. A Dual-energy X-ray absorptiometry showed severely low femoral and lumbar spine T-scores of -4,7SD and -5,3SD, respectively, and Z-score of -2,9SD and -2,6SD, respectively.

With these findings, an osteoporosis with a very high risk of new fractures diagnosis was assumed, and after discussing treatment options, the patient opted for denosumab therapy, along with calcium and vitamin D supplementation. Osimertinib therapy was maintained.

Despite treatment, during follow-up, the patient had a new fracture in the 2nd lumbar vertebrae.

Conclusion: This case highlights the potential role of Osimertinib as a secondary cause for osteoporosis, possibly being the first report to do so. While other causes in this patient cannot be excluded, preexisting studies showing changes in bone metabolism warrant attention among rheumatologists. Perhaps patients receiving long-term EGFR-TKI therapy face a higher risk for bone density loss, and preventive measures such as bone density monitoring and adequate supplementation should be recommended. More studies are needed to understand EGFR-TKI role in osteoporosis, especially as the population of long-term cancer patients treated with these kind of drugs is growing.

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065 - LYMPHOCYTIC INTERSTITIAL PNEUMONIA ASSOCIATED WITH SJÖGREN'S DISEASE:A RARE PULMONARY MANIFESTATION WITH COMPLETE RESOLUTION FOLLOWING IMMUNOSUPPRESSION

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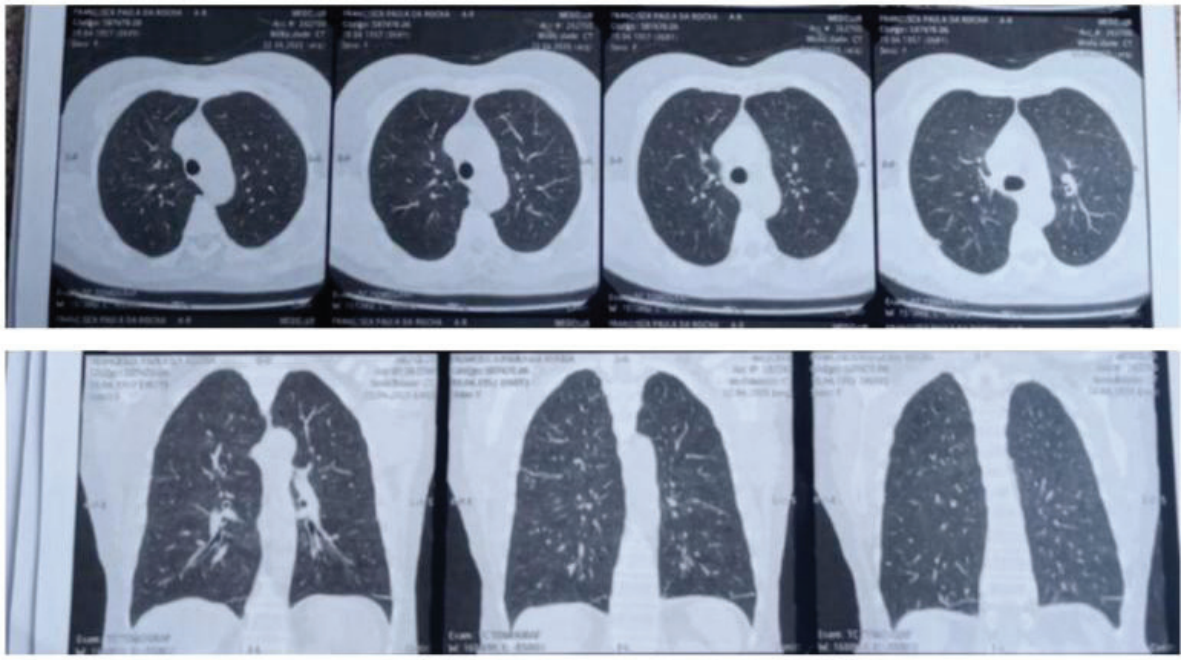
Introduction: Pulmonary involvement in Sjögren's disease represents one of the main causes of morbidity and mortality, including complications such as interstitial lung disease (ILD), which may present with various histological and radiological patterns. Lymphocytic interstitial pneumonia (LIP), a rare form of ILD, is characterized by diffuse infiltration of mature lymphocytes, and may be associated with pulmonary cystic changes, bronchiectasis, and centrilobular ground-glass opacities [1,2]. Early recognition is crucial to prevent progression to irreversible pulmonary fibrosis and functional respiratory decline [3].

Case Report: A 68-year-old woman with no prior comorbidities presented with symmetric peripheral polyarthritis involving both small and large joints (metacarpophalangeal, proximal interphalangeal,knees,and shoulders),with morning stiffness lasting over 60 minutes.Laboratory tests revealed elevated acute-phase reactants (CRP 1.71 and ESR 132), polyclonal hypergammaglobulinemia,homogeneous nuclear ANA (1:640),anti-Ro/SSA (155 U/mL), and anti-La/SSB (75 U/mL), with negative serologies for viral hepatitis and HIV. The Schirmer test showed tear production of 3 mm in 5 minutes, fulfilling the classification criteria for Sjögren's syndrome [4,5]. Hydroxychloroquine 400 mg/day and methotrexate 15 mg/week were initiated, but the latter was discontinued due to gastrointestinal intolerance.The patient subsequently reported progressive exertional dyspnea and dry cough, without fever. High-resolution computed tomography (HRCT) of the chest demonstrated findings consistent with LIP: mild emphysema, multiple sparsely distributed bilateral air cysts, central bronchiectasis, bronchial wall thickening, and centrilobular ground-glass opacities [6]. The diagnosis was supported by clinical, serological, and radiological findings, without need for lung biopsy. Systemic corticosteroid therapy with prednisone 1 mg/kg/day was initiated, combined with mycophenolate mofetil 2 g/day as a steroid-sparing agent. The patient showed complete remission of respiratory symptoms and significant radiological improvement after 3 months of treatment, with no signs of recurrence to date.

Conclusion: Although rare, LIP should be considered within the spectrum of systemic manifestations of Sjögren's disease, particularly in patients with positive anti-Ro/SSA and anti-La/SSB antibodies who develop insidious respiratory symptoms. This case highlights the importance of a multidisciplinary approach, with early diagnosis based on imaging and clinical immunology, avoiding invasive procedures and allowing effective therapeutic response. Mycophenolate mofetil proved to be a safe and effective immunosuppressive alternative, contributing to the reversal of inflammatory interstitial changes and improved pulmonary functional prognosis.



065 – Figure 1. Tomography image showing multiple bilaterally distributed air cysts, associated with bronchiectasis and centrilobular opacities

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076 - ESCLEROMIOSITE: UMA CONECTIVITE EM DISFARCE — O VALOR DO DIAGNÓSTICO INTEGRADO

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Introdução: A escleromiosite é uma entidade emergente no espetro das doenças do tecido conjuntivo, com características sobrepostas de esclerose sistémica (SSc) e miopatia inflamatória imunomediada (IIM). Embora os dados sejam heterogéneos, estima-se que o envolvimento muscular afete entre 10 a 90% dos doentes com SSc. Contudo, a escleromiosite distingue-se por apresentar um fenótipo clínico, imunológico e histopatológico específico.

Caso clínico: Homem de 28 anos, sem antecedentes relevantes além de rinite alérgica, internado em outubro de 2024 por quadro com 10 meses de evolução de edema facial, puffy hands, edema dos pés, mialgias, astenia, fenómeno de Raynaud e disfagia para sólidos, com agravamento progressivo. Ao exame objetivo, apresentava força muscular simétrica grau 4/5 nos grupos proximais e distais, sem atrofia e ausência de esclerose cutânea, úlceras digitais, telangiectasias, microstomia, distrofia ungueal ou outras lesões cutâneas.

Do estudo analítico efetuado, destacava-se, enzimas musculares elevadas (CK 1687 mg/dL, mioglobina 1083 mg/dL, aldolase 85 mg/dL), ANAs positivos (com título de 1/640, padrão nucleolar), bem como, anti-PM/Scl100, anti-PM/Scl75 e anti-Ro52 positivos. A videocapilaroscopia do leito ungueal revelou um padrão esclerodérmico em fase ativa. Pelos sintomas de astenia, realizou TC torácica que demonstrou achados enquadráveis em pneumonia intersticial não específica (NSIP), e estudo funcional respiratório compatível com padrão restritivo e capacidade de difusão do monóxido de carbono (DLCO) ligeiramente diminuída, indicativo de envolvimento pulmonar. O ecocardiograma apresentava dilatação ligeira das quatro câmaras, mas função sistólica biventricular preservada, sem sinais de hipertensão pulmonar. A RMN das coxas demonstrou

envolvimento difuso das fáscias e edema muscular ocasional e a eletromiografia dos membros inferiores foi compatível com padrão miopático inflamatório. A biópsia muscular revelou fibras em necrose e regeneração dispersas, presença de fibras basófilas e infiltrado inflamatório linfoplasmocitário no peri e epimísio/fáscia, sem características que permitissem uma classificação nosológica específica de miopatia imunomediada.

Dada a presença de miosite com características sobrepostas de esclerose sistémica, confirmou-se o diagnóstico de escleromiosite. Assim, e tendo em conta o envolvimento pulmonar, foi iniciado tratamento com prednisolona 15 mg/dia e micofenolato de mofetil até 3 g/dia. Por manter a astenia e elevação contínua de CK (7445 mg/dL), aldolase (104 mg/dL) e mioglobina (2350 mg/dL), decidiu-se pelo início de rituximab. Aguarda a realização de manometria esofágica.

Discussão: Este caso reflete, de forma didática, a apresentação típica da escleromiosite: sobreposição clínica e imunológica, envolvimento fascial predominante, autoanticorpos anti-PM/Scl e envolvimento pulmonar tipo NSIP. Dados recentes sugerem que certos autoanticorpos, como o anti-PM/Scl, estão associados a fenótipos específicos, incluindo maior risco de doença pulmonar intersticial (1). A ausência de esclerose cutânea reforça a importância de uma abordagem clínica abrangente que considere o estudo imagiológico, histológico e serológico.

Sublinha-se, portanto, a relevância da escleromiosite como entidade distinta, com implicações prognósticas e terapêuticas, e a necessidade do seu reconhecimento atempado. O diagnóstico precoce permitiu instituir terapêutica imunossupressora dirigida com o objetivo de prevenir complicações irreversíveis e o impacto deletério na funcionalidade do doente.

084 - PEMBROLIZUMAB-ASSOCIATED DIGITAL GANGRENE: A CASE REPORT

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Background: Immune checkpoint inhibitors (ICIs) have revolutionized the treatment of several cancers, including non-small cell lung carcinoma (NSCLC). However, they may trigger immune-related adverse events (irAEs), which can affect virtually any organ system. Vasculitic or vasculopathic irAEs, including digital ischemia and gangrene, are exceptionally rare but potentially severe.

Case Report: We report the case of a 61-year-old male, active smoker, with no history of autoimmune disease, diagnosed with bilateral segmental pulmonary embolism and stage IV lung adenocarcinoma with bilateral adrenal metastases. Tumor PD-L1 expression was 10%. He was started on first-line treatment with carboplatin, pemetrexed, and pembrolizumab. After 12 weeks of therapy, he developed painful acrocyanosis of the fingers—particularly the right index and middle fingers—associated with paresthesia and worsened by cold exposure. There was no history of trauma, similar prior symptoms or symptoms suggestive of systemic rheumatic disease

Initial management with topical nitrates, pentoxifylline, and subsequently amlodipine 5 mg (due to labile blood pressure) provided limited relief. Laboratory evaluation revealed a positive antinuclear antibody (ANA) at 1:1280 with a homogeneous pattern, elevated erythrocyte sedimentation rate (ESR 70 mm/h), and negative anti-extractable nuclear antigen (ENA) panel. Antineutrophil cytoplasmic antibodies (ANCA), cryoglobulins, and complement levels were normal. Screening for antiphospholipid antibodies showed negative anti-β2 glycoprotein I (IgM and IgG), negative anticardiolipin IgM, and equivocal anticardiolipin IgG. Lupus anticoagulant was not tested due to anticoagulation with rivaroxaban for the pulmonary embolism.

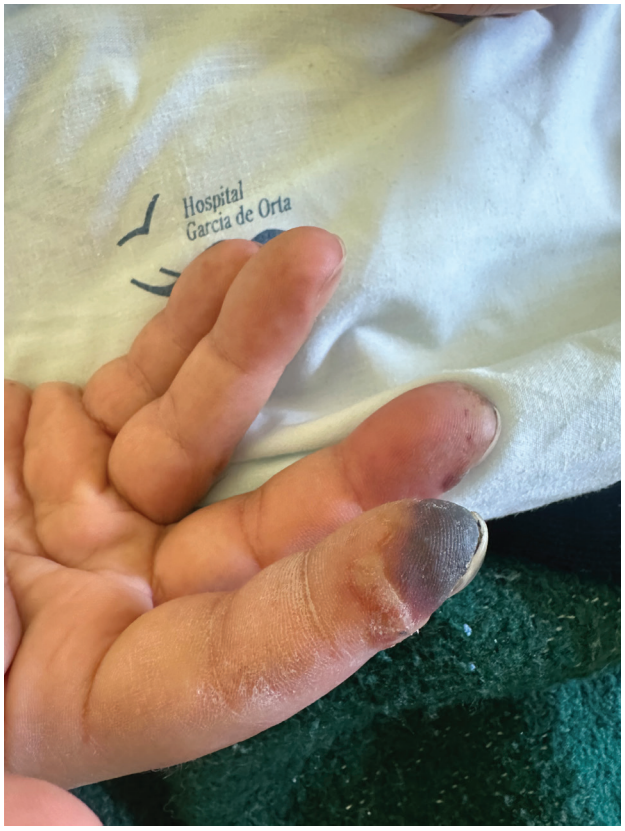
Doppler ultrasound and transthoracic echocardiography ruled out peripheral vascular obstruction and cardiac thromboembolic sources. Nailfold videocapillaroscopy revealed no pathological microvascular abnormalities.

Despite treatment, symptoms progressed to persistent dry gangrene of the digits.

Pembrolizumab was discontinued, and a course of oral prednisolone at 30 mg/day was initiated, with a taper over three weeks, but it was ineffective. The patient was hospitalized for a 10-day course of intravenous iloprost and high-dose corticosteroid therapy (1.5 mg/kg/day), consisting of two cycles of 5 consecutive days, given due to stabilization only after the first cycle. A secondary infection was suspected during admission and treated with flucloxacillin, with good clinical response.

Two months after discharge, the patient remained clinically stable, without further progression of digital necrosis. He is currently awaiting surgical amputation of one distal phalanx.

Discussion/Conclusion: This case highlights a rare but severe vasculopathic irAE associated with pembrolizumab, manifesting as digital ischemia and progressing to gangrene despite early intervention. Al-



084 – Figure 1. Digital ischemia and gangrene

though the clinical and immunological features strongly support an immune-mediated mechanism related to the immune checkpoint inhibitor, a potential contributory role of pemetrexed—rarely associated with digital ischemia in the literature—cannot be entirely excluded. Early recognition and multidisciplinary management are crucial to improving outcomes.
Images:

085 - A PEÇA DO PUZZLE QUE FALTAVA

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Introdução: As doenças reumáticas podem apresentar-se com manifestações clínicas diversas, por vezes semelhantes entre si, e levar tempo ou mesmo nunca vir a exibir o quadro clínico "completo" que as caracteriza, tornando o diagnóstico diferencial bastante desafiante.
Caso clínico: Homem de 63 anos, sem antecedentes de relevo, seguido em consulta de Reumatologia com o diagnóstico de Espondilartrite com envolvimento axial e periférico dada a presença de lombalgia crónica inflamatória com sacroileíte radiográfica de grau II, oligoartrite, episódio prévio de uveíte anterior do olho direito e positividade para HLA-B27, e medicado com salazopirina 3g/dia e etoricoxib 90 mg em SOS. Cerca de 8 anos após o diagnóstico, iniciou quadro de febre recorrente, tosse produtiva, dispneia e elevação dos parâmetros inflamatórios, associado ao aparecimento de consolidações pulmonares, sem isolamento microbiológico e sem resposta a diversos antibióticos, que motivou várias idas ao serviço de urgência e vários internamentos. Do extenso estudo realizado, foi detetado tromboembolismo pulmonar (TEP) e trombose venosa profunda (TVP) e positividade para o anticorpo anti-cardiolipina IgG (> 40 U em 2 doseamentos espaçados por 12 semanas) pelo que se assumiu o diagnóstico de Síndrome Antifosfolipidico e se iniciou varfarina. Após exclusão de etiologia infeciosa, e na suspeita de pneumonia organizativa concomitante, foi iniciada prednisolona na dose 50mg/dia (0.75mg/Kg/dia) com melhoria marcada clínica e analítica. Após desmame até à dose de 5 mg/dia, teve recidiva do quadro de febre, sintomas respiratórios e elevação dos parâmetros inflamatórios, novo episódio de TEP e de TVP, mesmo sob varfarina em níveis terapêuticos (INR 2-3), e uveíte aguda anterior bilateral. Em associação, o doente apresentou lesões cutâneas sugestivas de eritema nodoso dispersas pelos membros inferiores e de pseudofoliculite e reportou história de aftose oral com vários anos de evolução. A pesquisa de HLA-B51 foi negativa. Dada a refratariedade dos eventos trombóticos a varfarina, o quadro inflamatório associado com boa resposta a corticoterapia e os novos sintomas mucocutâneos, foi feita uma revisão diagnóstica concluindo tratar-se de Síndrome de Behçet. Foi retomada prednisolona na dose de 20 mg id com posterior desmame, suspensa salazopirina e iniciada azatioprina com titulação progressiva até 150 mg/dia, em associação a varfarina, com resolução da clínica, normalização dos parâmetros inflamatórios e sem recidiva de novos eventos trombóticos e de uveíte até à data.
Discussão: Contrariamente ao que acontece no Síndrome Antifosfolipídico, o TEP é uma manifestação rara do Síndrome de Behçet, em que, por se tratar de uma vasculite sistémica, os trombos tendem a aderir fortemente à parede do vaso inflamado. Está descrita a presença de anticorpos anti-fosfolipídicos em doentes com Behçet, a qual poderá potenciar o seu risco trombótico. Após exclusão de aneurismas pulmonares, a utilização de varfarina aliada à imunossupressão poderá ser uma estratégia terapêutica adequada na prevenção secundária de eventos trombóticos em doentes com Behçet e anticorpos antifosfolipidicos positivos.

087 - A RARE DIAGNOSIS OF KIKUCHI-FUJIMOTO DISEASE: THE RELEVANCE OF SUSTAINED CLINICAL FOLLOW-UP

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Introduction: Kikuchi-Fujimoto disease (KFD), also known as histiocytic necrotizing lymphadenitis, is a rare, benign, often self-limiting condition of unknown etiology. It predominantly affects women between the second and fourth decades of life and is mainly characterized by fever, constitutional symptoms and lymphadenopathy, typically cervical. Definitive diagnosis is based on histological evaluation of an excisional lymph node biopsy, which allows for the exclusion of the main differential diagnosis: Lymphoma, systemic lupus erythematosus (SLE), and infectious lymphadenitis. There are no established therapeutic guidelines. Management is based on nonsteroidal anti-inflammatory drugs (NSAIDs), corticosteroids, hydroxychloroquine, or intravenous immunoglobulin, depending on severity and recurrence. Patients with KFD should undergo regular clinical follow-up due to the potential for recurrence and association with SLE.

Here, we report a case of KFD and review its etiopathogenesis, clinical features, diagnosis, and treatment.

Case report: We describe a female patient with a past medical history notable for Hashimoto's thyroiditis, stable until age 36, when she developed polyarthralgia involving both proximal and distal joints symmetrically, associated with morning stiffness lasting one hour and excessive night sweats and fatigue. The patient experienced gradual improvement until age 45, when she developed painful axillary lymphadenopathy and worsening fatigue. Mammography and breast ultrasound revealed a large right axillary lymph node (25×10 mm), and thyroid ultrasound identified cervical lymph nodes that could be consistent with thyroiditis, though other diagnoses could not be excluded. An excisional biopsy of the axillary lymph node was performed for histological characterization, confirming proliferative-phase KFD.

The patient was referred to Rheumatology due to this diagnosis, prior history of arthralgias, and concurrent presentation of a cutaneous rash (urticarial-like lesions) in sun-exposed areas and peri-oral edema. She denied Raynaud's phenomenon, weight loss, other skin, respiratory or gastrointestinal symptoms, recent travel or animal exposure.

Laboratory evaluation revealed elevated C-reactive protein (CRP) of 7.6 mg/dL (reference <0.5 mg/dL). Antinuclear antibodies (ANA) were positive at a titer of 1:160 with a fine speckled pattern; anti-dsDNA were negative. Complement levels were within normal limits. Cryoglobulins were absent. Serum angiotensin-converting enzyme (ACE) was 44.4 U/L (reference <52 U/L). Serological tests for Epstein-Barr virus (EBV IgM), hepatitis C virus (anti-HCV), and HIV-1/2 were non-reactive. Huddleston, Rose Bengal, and Wright tests were negative. Serum and urine immunofixation revealed no monoclonal protein.

Given the persistence of the clinical picture for over 12 months, treatment was initiated with prednisolone 20 mg/day, followed by hydroxychloroquine 200 mg/day, leading to reduction in lymph node size and improvement of articular and cutaneous symptoms. Corticosteroids were tapered, with only occasional need due to lichen planus. Currently, at 52 years of age, patient remains on hydroxychloroquine 200 mg/day, with sustained remission of symptoms.

Conclusion: This case aims to highlight Kikuchi-Fujimoto disease and the importance of ongoing clinical follow-up given its potential association with SLE. We further review the epidemiology, pathophysiology, clinical manifestations, diagnostic workup, and management of this rare and challenging condition.

088 - MYOCARDITIS IN MIXED CONNECTIVE TISSUE DISORDER – A CASE REPORT

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Background: Mixed connective tissue disease (MCTD) is a rare autoimmune disorder characterised by the presence of symptoms of several connective diseases, such as systemic lupus erythematosus, systemic sclerosis, inflammatory myopathies, and rheumatoid arthritis, along with positivity for anti-ribonucleoprotein antibody (anti-RNP). Myocarditis is a rare presentation of MCTD and is associated with a worse prognosis.

Case report: We present the case of a 62 year-old woman with a history of diabetes and hypertension,

referred to our clinic due to progressive proximal muscle weakness for 18 months. She also reported loss of 14kg (20% of body weight), shortness of breath, hand joint pain, dry mouth and a facial rash. On examination, there was proximal muscle weakness affecting also the neck flexors (MMT8 score 59/80), alongside hand polyarthritis (MCP and PIP joints), Raynaud's phenomenon and rash of the frontal area and cheekbones.

Initial workup showed mildly elevated CRP (0.95mg/dL) and ESR (23mm/h), positive ANA (1/640), anti-RNP, RF (162.0UI/mL) and anti-CCP (701.3UQ), as well as elevated creatine kinase (1354U/L), myoglobin (357ng/mL) and troponin T (460ng/L). A pelvic MRI showed extensive areas of muscle oedema in the proximal muscles of the thighs with muscle atrophy and lipomatous infiltration, and the muscle biopsy was compatible with polymyositis. Due to the elevated troponin T levels, despite a normal electrocardiogram and echocardiogram, the patient underwent a cardiac MRI which showed signs of active myocarditis. She was then diagnosed with MCTD with articular, muscular, cutaneous, vascular and cardiac involvement and was started on IV pulses (methylprednisolone 1000mg/day for 3 days), followed by oral prednisolone (PDN) 40mg/day (~1mg/kg/day) and subcutaneous methotrexate (titrated to 25mg/week). She was also started on ramipril 2.5mg/day and bisoprolol 2.5mg/day and began physical therapy.

In the following 6 months, she reported improvement in these symptoms and the blood work showed a decrease in the CRP and ESR (0.77mg/dL and 7mm/h) and muscle enzymes levels (creatine kinase 43U/L, myoglobin <21ng/mL and troponin T 79.4ng/L). The follow-up cardiac MRI showed no signs of myocarditis. While the initial response to treatment was favourable, the disease would flare up with PDN dose lower than <15mg/day. With such high doses of corticosteroids, her diabetes and hypertension worsened and required further medication. As such, it was decided to escalate the immunosuppression to MMF and reduce the PDN dose.

Discussion: Cardiovascular (CV) involvement in MCTD appears to be frequent, with an estimated prevalence of 24-63%, most commonly presenting as acute pericarditis, pericardial effusion and conduction disturbances. Myocarditis is reported in only around 1.8% of cases and is thought to be part of the myositis-spectrum of the disease. While the overall prognosis of MCTD seems to be good, CV involvement is associated with worse prognosis – not only due to the severity of the organ involvement, but also because of the treatments used - high doses of corticosteroids, cyclophosphamide and mycophenolate mofetil (MMF).

In this case, the patient presented with exertional dyspnoea and marked elevation of troponin T which raised the suspicion of myocarditis, despite the absence of chest pain and normal routine cardiac exams. Only after a cardiac MRI was performed the diagnosis was confirmed. This case report aims to increase awareness for subclinical myocarditis in the setting of MCTD in order to adequately treat these patients and improve outcomes.

102 - INTERSTITIAL GRANULOMATOUS DERMATITIS: A RARE AND UNDERRECOGNIZED MANIFESTATION OF SYSTEMIC LUPUS ERYTHEMATOSUS

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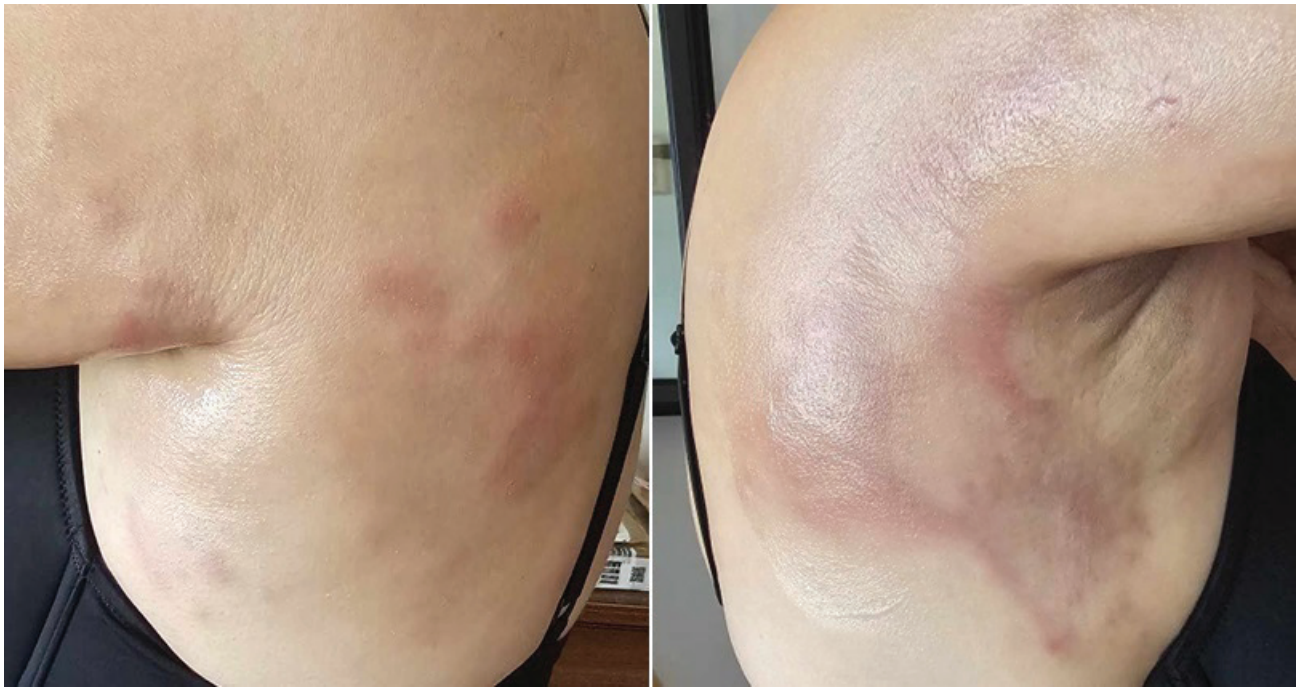
Background: Interstitial granulomatous dermatitis (IGD) is a rare idiopathic dermatosis with an unclear etiology, often associated with underlying conditions, such as autoimmune diseases, inflammatory disorders, and medications. Early recognition is crucial, as IGD can mimic other cutaneous manifestations and impact therapeutic decisions. It is characterised by a distinctive histopathological pattern and is frequently linked to arthritis, particularly in the context of Ackerman syndrome.

Case presentation: We report the case of a 37-year-old woman diagnosed with Systemic Lupus Erythematosus (SLE) at the age of 26, with cutaneous (malar rash), hematologic (leukopenia), and articular involvement. Her immunological profile revealed a positive antinuclear antibody (ANA) titer of 1/1280 and positivity for anti-double-stranded DNA, SSA, SSB, and RNP antibodies. She presented with well-demarcated, painful, erythematous-violaceous papules and linear lesions bilaterally in the axillary regions (Figure 1). Her medical history was remarkable for hydroxychloroquine (HCQ) discontinuation 5 months prior due to retinal toxicity. Current medication included methotrexate, leflunomide and low-dose corticosteroids owing to difficult-to-control arthritis. A skin biopsy revealed a dense interstitial infiltrate in the reticular dermis, featuring lymphocytes and histiocytes surrounded by collagen fibres exhibiting a globular morphology with



105 – Figure 1. Bilateral axillary rash illustrating the "rope sign"

a floating appearance - findings consistent with IGD. The patient responded favourably to topical tacrolimus 0.1%, showing significant clinical improvement within weeks.

Conclusion: The association between IGD and SLE is seldom reported, with only a limited number of cases described in the literature. This case expands the recognised cutaneous spectrum of SLE. Recognition of the "rope sign" and prompt histopathological evaluation are essential to ensure accurate diagnosis and guide targeted management.

104 - EFEITO ADVERSO RARO E GRAVE DA IMUNOTERAPIA: MIOSITE, MIOCARDITE E MIASTENIA EM DOENTE SOB PEMBROLIZUMAB

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Os inibidores do checkpoint imune (ICI) são utilizados no tratamento de neoplasias avançadas com sobre-expressão de PD-L1, por coestimulação entre células B e células T, com potencial de efeitos adversos imunomediados. O caso reportado insere-se nesse contexto.

Homem, 78 anos, com adenocarcinoma do cólon com metastização peritoneal e sobre-expressão de PD-L1, sob pembrolizumab (ICI) com intuito paliativo. 3 dias após a 2ª toma, desenvolveu défice de força generalizado, disfagia, diplopia binocular, visão turva, dispneia e edema dos membros inferiores.

Ao exame físico, encontrava-se apirético, normotenso, normocárdico, sem exantema cutâneo. A auscultação pulmonar revelou crepitações bibasais. A avaliação da força muscular mostrava défice na flexão da coxa (G2); abdução e flexão dos braços (G3); flexão e extensão de antebraços e pernas (G4). Destacava-se edema dos membros inferiores até ao joelho com sinal Godet.

Analiticamente: elevação de marcadores de citólose hepática - AST 451 U/L (N 12-40); ALT 183 U/L (N 7-40); rabdomiólise - CK 4542 U/L (N 46-17), Mioglobina 5000 ng/mL (N < 110); Troponina I 3,042 ng/mL (N < 0,045); e de parâmetros inflamatórios- Proteína C reativa 14 mg/L (N < 5,0). Os anticorpos ANA; anti-AchR eram negativos. O eletrocardiograma e o ecocardiograma não tinham alterações de relevo.

Observado por Reumatologia, Neurologia e Cardiologia, diagnosticada miopatia induzida por ICI, levantada a possível coexistência de miastenia com a mesma etiologia e considerado provável o atingimento do músculo cardíaco.

Pela gravidade do quadro clínico - miopatia induzida por ICI com acometimento possível do músculo

cardíaco, músculos da deglutição e músculos extrínsecos do olho - iniciada corticoterapia (pulsos de 500mg metilprednisolona 3 dias, após os quais iniciou prednisolona na dose de 0,5 mg/Kg/dia). Acordou-se com Oncologia a suspensão definitiva de ICI. Associadamente, pela diabetes mellitus mal controlada, introduziu-se tocilizumab endovenoso como poupador de corticosteróides.

Uma semana após início da terapêutica, o doente apresentou evolução clínica favorável, melhoria da força muscular proximal (G4 a nível dos grupos musculares proximais dos membros superiores e inferiores), conseguindo deambular como anteriormente. Também houve evolução analítica favorável - AST 113 U/L; ALT 178 U/L; CK 707 U/L; Troponina I,154 ng/mL, Mioglobina 1618 ng/mL.

O doente foi readmitido 1 mês após a alta hospitalar por pneumonia nosocomial, iniciando antibioterapia de largo espectro. Durante o internamento, não houve agravamento do défice de força muscular periférico e, analiticamente, houve resolução da rabdomiólise/lesão miocárdica (CK 39 U/L; Mioglobina 104 U/L; Troponina I 0,052 ng/mL). Apesar da resolução do quadro infeccioso, persistiu insuficiência respiratória e houve agravamento da disfagia. A avaliação por Neurologia conduziu à conclusão de que se justificava por provável crise miasténica, optando-se pela realização de imunoglobulinas (30 g durante 5 dias). Não houve benefício significativo. Optou-se, após discussão multidisciplinar com Doenças Infecciosas e Oncologia Médica, retomar tocilizumab, também sem benefício no tratamento do quadro miasténico.

O caso clínico sublinha a importância da vigilância apertada a doentes sob ICI. A miosite associada a ICI com atingimento do músculo cardíaco requer diagnóstico e tratamento precoces. Mostra também a importância da abordagem multidisciplinar destes doentes. O tratamento com tocilizumab revelou-se eficaz na miosite e miocardite associadas a ICI; contudo, não melhorou o quadro de miastenia.

108 - EOSINOPHILIC GRANULOMATOSIS WITH POLYANGIITIS WITH A POLYMYALGIA RHEUMATICA-LIKE MUSCULOSKELETAL FLARE: A DIAGNOSTIC AND THERAPEUTIC CHALLENGE

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Introduction: Eosinophilic Granulomatosis with Polyangiitis (EGPA) is a rare ANCA-associated vasculitis (AAV) marked by adult-onset asthma, peripheral eosinophilia, and systemic small-vessel inflammation with multi-organ involvement.[1] While arthralgia and myalgia are common, atypical manifestations can emerge, complicating disease monitoring and management.[1]

Case report: A 56-year-old male with a history of chronic rhinosinusitis and late-onset asthma presented to the emergency department with a 4-day history of left lower limb paresthesias and a painful, pruritic rash on both legs, which had progressed to a painless ulcer. He also reported asthenia, anorexia, a 5 kg weight loss over 3 months, migratory arthralgia of large joints, and a year-long persistent dry cough. On examination, he was hypertensive (164/98 mmHg), with purpuric lesions and a painless ulcer over the medial aspect of the left ankle. Laboratory workup revealed leukocytosis with marked eosinophilia (4.99 G/L, 29%), elevated CRP (10.1 mg/dL), and ESR (30 mm/h). During hospitalization, he developed proteinuria (0.5 g/24h), hematuria (>20 RBCs/field), and leukocyturia with preserved renal function. Chest CT angiography revealed peribronchovascular ground-glass opacities in both upper lobes. Electromyography confirmed isolated sensory mononeuropathy of the left superficial peroneal nerve. Skin biopsy showed leukocytoclastic vasculitis. ANCA testing revealed strong p-ANCA positivity (>600 U/mL) and anti-MPO 80 UI/mL.

EGPA was diagnosed, with cutaneous, renal, neurologic, pulmonary and otorhinolaryngologic involvement. Remission induction included 3 pulses of 1g intravenous methylprednisolone, followed by oral prednisolone (PDN) (1 mg/kg/day), cyclophosphamide and rituximab (RTX), with good response. RTX was continued for maintenance.

One year later, the patient developed new-onset arthralgias affecting the shoulders, hips, and wrists, with marked morning stiffness. Inflammatory markers (CRP and ESR) were normal, as were ANCAs and anti-MPO antibodies. Shoulder musculoskeletal (MSK) ultrasound revealed bilateral biceps tenosynovitis, moderate glenohumeral synovitis, and wrist synovitis with Doppler signal, suggesting a flare manifesting as polymyalgic syndrome. There were no findings suggestive of large vessel vasculitis, namely giant cell arteritis, or other signs of active EGPA disease. Methotrexate (MTX) was initiated, along with a short course of PDN (15 mg/day). Clinical improvement within 48 hours supported the diagnosis. He remains in remission on RTX (1 g every 6 months) and MTX (17.5 mg/week), with PDN (5 mg/day) being tapered.

Discussion: While MSK symptoms are common, a

PMR-like pattern is rare and may reflect relapse or concurrent rheumatic condition. In this patient, already on maximal standard therapy with RTX, the emergence of new joint symptoms prompted further evaluation. Given the lack of clear guidelines for EGPA flares under RTX, adding rather than switching to MTX was preferred. Rapid improvement supported this treatment choice in selected cases.

Conclusion: This case underscores the diagnostic and therapeutic complexity of EGPA, particularly when atypical MSK symptoms emerge despite appropriate therapy. Early recognition and tailored treatment are essential to avoid misdiagnosis and ensure optimal disease control. Further data is needed to guide management in cases where standard care proves insufficient.

110 - OSTEOMALÁCIA: UM RETRATO CLÍNICO DE UMA CAUSA ESQUECIDA DE FRATURAS DE FRAGILIDADE

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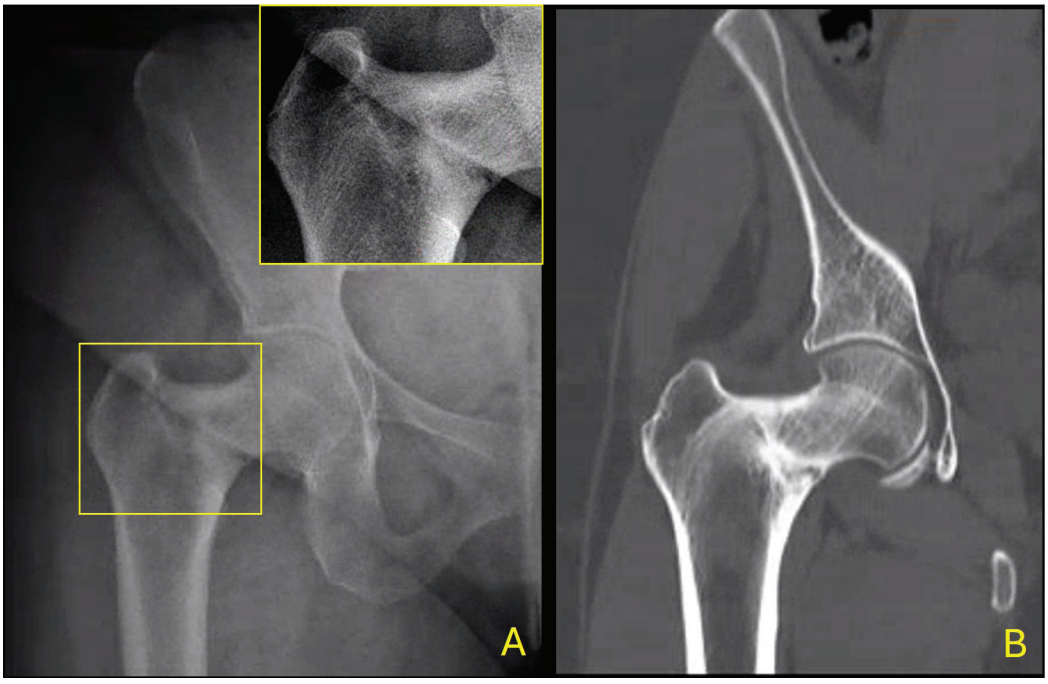
Introdução: As fraturas de fragilidade são aquelas que resultam de trauma de baixo impacto ou que ocorrem mesmo na ausência de trauma apurável. Estas estão relacionadas com alterações na biomecânica do osso, quer por menor densidade óssea, quer por uma geometria ou mineralização anormal. A osteomalácia resulta precisamente de um defeito na mineralização óssea, tendo como principal causa nos adultos o défice de vitamina D, podendo manifestar-se através de dor óssea e articular ou manter-se subclínica até resultar em fratura.

Caso clínico: Apresentamos o caso de uma doente do sexo feminino de 38 anos, leucodérmica, operária fabril com antecedentes pessoais conhecidos de obesidade grau II e rinite alérgica, que em novembro de 2011 recorreu aos Cuidados de Saúde Primários por uma coxalgia direita de ritmo mecânico, com alguns dias de evolução, de início espontâneo, sem trauma associado. Negava dor articular, muscular ou óssea prévia, alterações da marcha e fraturas prévias. Neste contexto, realizou uma radiografia da bacia, seguida de tomografia computorizada da articulação coxofemoral direita que revelaram uma fratura incompleta do colo femoral direito, sugestiva de pseudofratura de Looser-Milkman (imagem 1), tendo a doente sido encaminhada para as consulta de Ortopedia e Reumatologia.

À avaliação na consulta de Reumatologia foi con-



110 – Figure 1. Imagem 1(A,B): Representação radiográfica (A) e em Tomografia Computorizada (B) de fratura incompleta do colo femoral direito.

statada discreta coxa vara bilateralmente e dor à palpação trocantérica à direita, além de obesidade, sendo o restante exame físico normal. Analiticamente, a destacar um défice de vitamina D (25-OH-vitamina D) de 12 ng/mL (défice<20) e uma hormona paratiroideia intacta (PTH-I) próxima do limite superior da normalidade - 62.9 pg/mL (15.0-65.0) – estando a calcemia, fosforemia, fosfatase alcalina, função tiroideia e renal e parâmetros hepatobiliares dentro da normalidade. A avaliação densitométrica revelou uma densidade mineral óssea (DMO) normal ao nível do colo femoral (CF) (0.897g/cm2; Z-score +0.7) e osteopénia na coluna lombar (CL) total (0.765g/cm2; Z-score -2.3).

Foi assumido o diagnóstico de osteomalácia secundária ao défice de vitamina D e a doente iniciou tratamento com suplementação com calcifediol e carbonato de cálcio.

Do ponto de vista ortopédico, a fratura foi abordada de forma conservadora, com vigilância semestral nos primeiros dois anos após fratura.

Para exclusão de etiologias secundárias de hipovitaminose D, foi revista a história pregressa, antecedentes pessoais (biológicos e farmacológicos) e familiares que não revelaram outras etiologias identificáveis.

À data atual - catorze anos depois - a doente mantém suplementação com vitamina D e cálcio, encontrando-se sem limitação funcional ou novas fraturas. A reavaliação osteodensitométrica – 7 anos após a primeira - revelou uma sobreponibilidade da DMO (CF: 0.879g/cm2; Z-score +0.9; CL: 0.729g/cm2; Z-score -2.3).

Este caso realça a importância do estudo etiológico de fraturas de fragilidade e sublinha o papel da correção do défice de vitamina D na prevenção de fraturas e consequente morbilidade. A esta relevância acresce o facto de frequentemente - como no presente caso - a hipovitaminose D afetar indivíduos mais jovens, com maior potencial para anos perdidos por incapacidade, na ausência de um diagnóstico e tratamento atempados.

112 - SIMULTANEOUS BILATERAL ATYPICAL FEMORAL FRACTURE WITH DENOSUMAB

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Introduction: Antiresorptive therapies such as denosumab or bisphosphonates (BPs) reduce the risk of vertebral and non-vertebral and hip fractures, which are major causes of mortality, disability and economic burden.

However, concerns have been raised about rare but potentially severe adverse events such as atypical femoral fractures (AFFs). Some authors report an estimated incidence of 0.8 per 10,000 patient-years of AFFs in patients under denosumab.

Presentation of case: We present the case of a 67-year-old woman with medical history left hip replacement surgery due to osteoarthritis, dyslipidemia, and nephrolithiasis. She had a relevant family history of osteoporosis in both her parents and her son was diagnosed with mild form of osteogenesis imperfecta.

She was referred to our osteoporosis clinic after 5-year course of intravenous zoledronic acid (5mg/year) due to vertebral fragility fractures (L1 and L2). Despite bone mineral density (BMD) improvement and no recurrence of fractures, she maintained vertebral osteoporosis and has hence started on denosumab 60mg every 6 months in 2022.

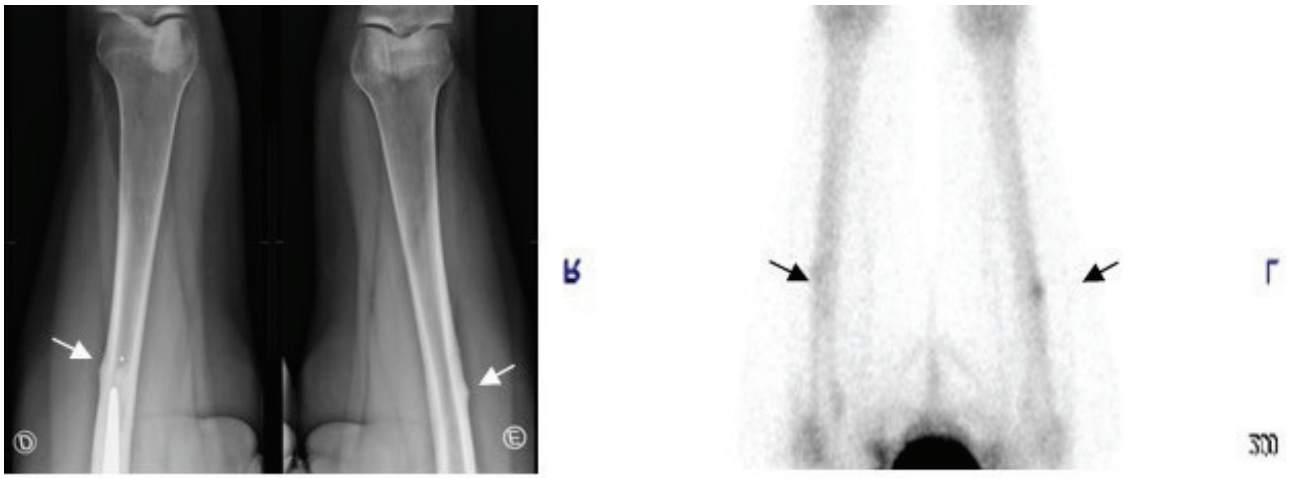
In January 2025 she was assessed due to gradual onset of an ill-defined bilateral thigh, without any reported trauma, with progressive functional impairment requiring the use of a walking aid.

Bilateral hip radiographs, computed tomography, and bone scintigraphy were performed, confirming bilateral AFFs (Figure 1). Current BMD assessment by DEXA showed a L1-L4 T-score worsening (from -2.6 to -3.0) and femoral neck improvement (from -1.5 to -0.9). Denosumab was discontinued and treatment with teriparatide was initiated.

Discussion: To the best of our knowledge, this is the third reported case of simultaneous bilateral AFF in a patient treated with denosumab. The patient fullfills all criteria (4 major and 6 minor) for AFF stablished by the 2010 ASBMR Task Force.

The clinical features of the patient described are highly consistent with those reported in the literature. In the two previously reported cases of AFFs, both patients were women of similar age. One had undergone long-term bisphosphonate therapy, such as our patient. The second case involved a patient with a medical history of breast cancer with bone metastases and a prior pathological hip fracture following a fall, who had only been treated with denosumab. The remaining reported cases of unilateral AFFs also share key features with our case, including sex, advanced age, long-standing osteoporosis and previous bisphosphonate use.

While the occurrence of this rare type of fracture does not provide conclusive evidence of a causal relationship between denosumab and AFFs. It seems to be clear that the risk of incidence of AFF seems to be higher in patients with prior bisphosphonates use. We need more research for this rare condition to clarify



129 – Figure 1. A – right femoral radiograph; B – left femoral radiograph; C - bone scintigraphy

and refine the treatment protocols.

Conclusions: AFFs is a rare adverse effect and only a few cases have been reported with the use of denosumab. However, we must keep in mind that prescribing denosumab to a patient previously treated with bisphosphonates increases such risk.

We emphasize previous guidance which states that the screening of radiographs of femurs should be considered in such cases where the patient has received treatment with multiple antiresorptive drugs.

115 - WHEN PLEURITIC PAIN HIDES A SHRINKING LUNG

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Introduction: Systemic lupus erythematosus (SLE) is a complex disease with multiorgan involvement. Pulmonary manifestations are frequent, occurring in 60–80% of cases. Shrinking lung syndrome (SLS), one of the rarest pulmonary manifestations, has a prevalence of <1%. Clinically, it presents with dyspnea, cough, and pleuritic chest pain. No diagnostic criteria exist for SLS; it relies on a combination of clinical presentation, elevated hemidiaphragm, reduced lung volumes with a restrictive pattern on pulmonary function tests, and exclusion of other causes. Some reported cases show improvement with corticosteroids and immunosuppressive therapy, including azathioprine, cyclophosphamide, methotrexate, mycophenolate mofetil, and rituximab. Theophylline and beta-agonists may provide symptomatic relief for diaphragmatic weakness. However, clinical guidelines for managing this rare but important manifestation remain unestablished.

Case: We present the case of a 26-year-old woman referred to Rheumatology for inflammatory polyarthralgia, malar rash, photosensitivity, and left-sided pleuritic chest pain, which worsened at night and when lying on her side. She denied dyspnea, cough, palpitations, syncope, fatigue, fever, or night sweats. She was taking prednisolone (PDN) 10 mg/day and had a history of deep vein thrombosis. Physical examination was unremarkable except for a malar rash. Laboratory tests showed elevated inflammatory markers, complement consumption, high-titer ANA (>1:640, homogeneous pattern), high-titer anti-dsDNA (800 IU/mL), and positivity for anti-nucleosome, anti-ribosomal P protein, antiphospholipid antibodies, and lupus anticoagulant. A diagnosis of SLE and secondary antiphospholipid syndrome was established. Treatment with hydroxychloroquine 400 mg/day and warfarin was initiated.

Echocardiography was normal, and the electrocardiogram showed sinus tachycardia. A ventilation-perfusion scan showed no evidence of pulmonary thromboembolism. Chest CT revealed subsegmental atelectasis of the lingula, resulting in elevation of the left hemidiaphragm. Pulmonary function tests demonstrated reduced lung volumes and a restrictive pattern. Electromyography of the phrenic nerves showed amplitudes at the lower limit of normal. A diagnosis of shrinking lung syndrome was made. Azathioprine was initiated and titrated to 175 mg/day. The patient showed clinical improvement, with resolution of pleuritic pain and decrease in anti-dsDNA levels, allowing tapering of PDN to 5 mg/day.

Discussion: SLS is rare; its pathophysiology, treatment responses, and long-term outcomes remain poorly understood. An extensive study is necessary to exclude other causes. Corticosteroids and immunosuppressive agents may lead to clinical improvement, although no established guidelines currently exist for the management of SLS.

118 - OSTEOPOROSIS ASSOCIATED WITH A PLS3 GENE MUTATION: A CASE REPORT

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Introduction: Osteoporosis (OP) is the most common metabolic bone disease, mainly affecting postmenopausal women, but also occurring in younger individuals and men. Secondary OP may arise due to chronic conditions, medications, or alcohol use. In young adults with severe OP, multiple fractures, a family history of OP, and no clear secondary cause, monogenic bone diseases should be investigated. The prevalence of OP in young patients is underestimated, and management strategies remain unclear.
Case Report: We report the case of a 42-year-old man with multiple fragility fractures since age 30. After a femoral neck and olecranon low-energy fracture, he initiated alendronate 70 mg/week in 2013 for six years. Despite good treatment compliance, he sustained several additional fragility fractures: the right 5th metacarpal (2014), bilateral wrists (2015), and vertebral bodies T3 and T4 (2018). At this point, the patient was referred to Rheumatology. He showed no stigma of osteogenesis imperfecta, and possible secondary OP causes were ruled out. A detailed family history uncovered a maternal uncle with multiple fragility fractures from a young age. Therapeutic failure was assumed, and therapy was switched to teriparatide 20 mcg/day (2019–2021). Despite no significant improvement in bone density, the patient did not suffer further fractures. Zoledronate was used as sequential therapy. He was also referred to a clinical geneticist. A hemizygous c.342C>G (p.Ser114Arg) variant in the PLS3 gene was identified by genomic test.
Discussion: The X-linked PLS3 gene, primarily affecting hemizygous males, encodes Plastin-3, a protein with bone-regulatory properties. The exact mechanism by which PLS3 mutations cause OP and fractures is unknown. Patients with PLS3 mutations typically develop early-onset OP, making sequential treatment essential. However, current therapies are based on postmenopausal OP studies, highlighting the urgent need for research into the efficacy and safety of treatment options in this population.

120 - NOT ALWAYS POLYMYALGIA RHEUMATICA: UNMASKING CYCLOSPORINE-INDUCED MYOPATHY

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Introduction: Cyclosporine A (CsA) is an immunosuppressant widely used in transplant recipients to prevent organ rejection, also being employed in the treatment of various autoimmune diseases. It is associated with several well-documented adverse effects, including nephrotoxicity, gastrointestinal disturbances, and gingival hyperplasia. CsA-induced myopathy is a rare adverse event, with only a limited number of cases reported in the literature.
Case Presentation: We report the case of a 74-year-old man with a history of kidney transplantation in 2009, maintaining immunosuppressive therapy with CsA and mycophenolate mofetil since then. In 2019, he presented pain and stiffness in the pelvic girdle, which led to difficulty ambulating, rising from a chair, and maintaining an upright posture. There was no involvement of the shoulder girdle or peripheral joints, fever, or other symptoms. On physical examination, shoulder mobility was preserved; but there was significant pain with hip movement and marked limitation in all ranges of motion. Laboratory tests revealed elevated inflammatory markers (CRP 177mg/L and ESR 93 mm/h) with normal muscle enzyme levels. A PET scan showed an intensely FDG-avid focus in the anterior pelvic region, predominantly involving soft tissues, but MRI of the thighs only described a bilateral partial tear of the adductor muscles. A presumptive diagnosis of polymyalgia rheumatica (PMR) was made, and treatment with prednisolone (PDN) 15 mg/day was initiated, resulting in an initial significant clinical and laboratory response. However, tapering PDN was difficult, and the patient continued to experience fluctuating episodes of worsening pain, particularly on the right thigh.

In 2022, due to worsening anterior thigh pain, particularly with movement and palpation, and associated limb weakness, a repeat MRI of the thighs was performed. This revealed findings consistent with myopathy in the right adductor and hamstring compartments, with atrophy and diffuse fatty infiltration, as well as areas of hyperintensity on fluid-sensitive sequences. Electromyography demonstrated a myopathic pattern. Muscle enzymes and a comprehensive immunologic work-up, including viral serologies, were negative. Muscle biopsy showed no inflammatory infiltrates but did reveal necrotic fibers, MHC I expression, and fibers with altered morphology, suggestive of a myopathic process. Retrospective review of imaging revealed that the abnormalities identified in the 2022 MRI were already present in 2019.

Since PDN had only been introduced after the ini-

tial MRI, steroid-induced myopathy was considered unlikely. Also, faced that the patient developed a myopathic condition under immunosuppression, CsA-induced myopathy was suspected. In collaboration with nephrology, CsA was discontinued in 2023 and replaced with everolimus. Following CsA discontinuation, the patient experienced significant clinical improvement, with resolution of pain and restoration of muscle strength. In 2025, follow-up MRI demonstrated mild improvement in muscular interstitial oedema particularly in the short heads of the biceps femoris.

Discussion: This case highlights that myopathy can occur as an adverse effect of CsA, sometimes with an atypical presentation. Its pathogenesis remains poorly understood and muscle biopsy findings are highly variable. Unlike PMR, CsA-induced myopathy typically does not cause elevated inflammatory parameters, which made this case even more challenging. The most important clue is that symptoms typically improve or completely resolve following CsA discontinuation.

123 - A UNIQUE CASE OF COEXISTENCE OF HAJDU-CHENEY SYNDROME AND A MILD VASCULAR EHLERS-DANLOS PHENOTYPE: CLINICAL AND GENETIC INSIGHTS

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Introduction: Hajdu-Cheney syndrome (HCS) is a rare autosomal dominant skeletal dysplasia (prevalence <1/1,000,000) characterized by acroosteolysis, osteoporosis, craniofacial dysmorphism, and short stature. Vascular Ehlers-Danlos syndrome (vEDS), due to pathogenic COL3A1 variants, is another rare disorder (prevalence 1:50,000–1:200,000). To our knowledge, no association between these two entities has been previously described.

Case Presentation: A 62-year-old woman with a long-standing HCS diagnosis presented with progressive joint deformities. Since age 50, she had classic HCS features: acroosteolysis, craniofacial dysmorphism (persistent cranial sutures, micrognathia, thin upper lip), and severe osteoporosis (T-score –3.6 lumbar spine), treated sequentially with teriparatide, strontium ranelate, and now zoledronic acid. Examination showed acroosteolysis of the 3rd and 4th right-hand fingers, reversible flexion deformities in multiple fingers of both hands, and clawed toes.

Genetic testing, performed 12 years after the initial diagnosis, identified a NOTCH2 c.415+26G>C variant of uncertain significance, not previously described but located in a region previously implicated in HCS. Exome sequencing revealed a novel heterozygous COL3A1 variant (c.4109A>G; p.Tyr1370Cys) in exon 50, within the N-terminal region of type III procollagen. The patient was referred for genetic consultation, which concluded that, although this variant has not been previously reported, it is predicted to be deleterious. Notably, there was no prior history of vascular or visceral rupture, but family history across three generations revealed thin translucent skin, joint hypermobility, micrognathia, and positive wrist/thumb signs in the granddaughter, raising suspicion of mild vEDS with autosomal dominant inheritance.

Cerebral and cervical angio-MRI excluded dissection or malformations but identified a focal >70% left internal carotid artery stenosis. Hand MRI showed Jaccoud-like arthropathy, known acroosteolysis, and bone cysts without erosions. Immunological and metabolic workup was unremarkable.

Discussion: In this case, the primary diagnosis of Hajdu-Cheney syndrome is supported by the phenotype and a NOTCH2 variant. However, the additional connective tissue findings and a novel COL3A1 variant suggest dual pathology. The variant's position in the N-terminal domain may explain the attenuated phenotype (contrasting with the classic form of vEDS, which is typically associated with arterial and visceral rupture and vascular malformations), consistent with literature, indicating a milder later-onset disease with such mutations (1). The multigenerational family history, with autosomal dominant features, further supports this diagnosis.

The combination of these rare conditions has not been previously reported. Their coexistence may be coincidental, although both affect connective and skeletal tissues. We describe a unique case of coexisting Hajdu-Cheney syndrome and a mild form of vascular Ehlers-Danlos syndrome within the same patient and family, supported by clinical findings and novel genetic variants. The potential interaction or phenotypic overlap between these rare conditions warrants further investigation and contributes to the expanding spectrum of connective tissue disorders.

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ACKNOWLEDGMENT

Images referenced in this case report will be added to the final version of the poster.

124 - SÍNDROME COMPRESSIVA PÉLVICA EM CONTEXTO DE SÍNDROME DE SJÖGREN: QUANDO A DOR NÃO É SÓ PSICOGÉNICA

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Introdução: A síndrome de Sjögren (SS) é uma doença sistémica caracterizada por disfunção das glândulas exócrinas, com xeroftalmia, xerostomia e, ocasionalmente, secura vaginal. Pode coexistir com artralgias e fadiga generalizada. A dor neuropática na região pélvica , incluindo a meralgia pudenda e/ou a compressão do nervo cutâneo perineal, é uma entidade menos reconhecida mas potencialmente incapacitante, sendo frequentemente atribuída a doenças psiquiátricas, como a depressão ou a ansiedade, muito comuns na SS.
Caso clínico: Mulher de 42 anos, raça branca, com antecedentes de depressão, ansiedade e marsupialização da glândula de Bartholin esquerda em contexto de quisto recidivante, referenciada à consulta de Reumatologia em 2022 por xerostomia e secura vaginal. Associadamente, com cerca de 10 anos de evolução, apresentava dor vulvovaginal crónica com características disestésicas (queimor, alodinia, hiperestesia), agravada ao toque, sentar, marcha e uso de vestuário apertado, impossibilitando atividades da vida diária e relações sexuais. Estas queixas foram sendo atribuídas ao seu estado psiquiátrico, com escasso benefício de múltiplos tratamentos (tópicos, antifúngicos e neuromoduladores). Após investigação, foi diagnosticada com SS primário (anticorpos antinucleares e anti-SSA positivos, histologia de glândula salivar minor com infiltrado linfoplasmocitário e cintigrafia com hipofunção das glândulas submandibulares) e, dada a manutenção das queixas disestésicas genitais desde, pelo menos 2011, foi realizada uma ressonância magnética pélvica que revelou sinais compatíveis com compressão dos nervos pudendos na sua passagem junto à espinha isquiática, entre os ligamentos sacroespinhoso e sacrotuberoso espessados, sobretudo à esquerda, e possível compressão do ramo cutâneo perineal femoral posterior esquerdo, secundária a um quisto residual da glândula de Bartholin. Considerando estes achados, a doente foi referenciada a consulta de medicina física e reabilitação. Realizou um primeiro bloqueio anestésico do nervo pudendo esquerdo com benefício transitório e, em setembro de 2023, foi submetida a neuromodulação bilateral do nervo pudendo, incluindo bloqueio e radiofrequência pulsada do gânglio ímpar e dos nervos pudendos sob fluoroscopia. Apresentou uma franca melhoria da alodinia vulvovaginal e da dor neuropática, conseguindo retomar progressivamente atividades antes impossibilitadas. Iniciou fisioterapia pélvica dirigida à dessensibilização e reeducação neuromuscular, com ganhos funcionais sustentados. Do ponto de vista da SS, não foi necessária a introdução de tratamento imunossupressor.
Discussão: Este caso sublinha a importância de uma abordagem multidisciplinar e sistemática na avaliação de dor crónica na doença reumática. Apesar do diagnóstico de SS ter sido estabelecido, as queixas neuropáticas persistentes foram inicialmente atribuídas a fatores psicossociais, subestimando-se o sofrimento e o impacto funcional para a doente. A realização tardia da RM pélvica permitiu o diagnóstico de uma síndrome compressiva dos nervos pudendos e do ramo cutâneo perineal, potencialmente agravada por sequela da cirurgia prévia da glândula de Bartholin. Posteriormente, verificou-se a eficácia da neuromodulação pudenda, associada à reabilitação funcional, no alívio sintomático e na recuperação da qualidade de vida.

Concluíndo, nesta vinheta, ilustra-se a importância da escuta ativa e da valorização das queixas, especialmente, na presença de doenças reumáticas imunomediadas cuja comorbilidade psiquiátrica e a dor crónica são frequentes.

125 - SÍNDROME HEMOFAGOCÍTICO EM DOENTE COM LES E SAAF: DIAGNÓSTICO COMPLEXO E DESFECHO DE ALTO RISCO

Carlos Marques-Gomes[1, 2], Mariana Diz-Lopes [1, 2], Miguel Correia Natal[1], Bárbara Fernandes Esteves[1], Mariana R Sebastião[3], Inês Almeida[4], Sara Amaro Lopes[1], Ana Sá[1], Marina Oliveira[3], Miguel Bernardes[1, 2], Teresa Martins-Rocha[1, 2], Lúcia Costa[1]
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Introdução: A linfohistiocitose hemafagocítica/síndrome de ativação marcrofágica (LHH/SAM) é uma condição inflamatória grave de hiperativação imune associada a doenças reumáticas, como o lúpus eritematoso sistémico(LES), podendo levar a disfunção multiorgânica e mortalidade elevada. O diagnóstico é desafiante, sobretudo quando coexiste infeção — trigger em >65% dos casos com doença reumática subjacente —, alterações hematológicas e ausência de sinais de atividade imune(1).

Caso clínico: Mulher de 61 anos, diagnosticada desde os 41 anos com LES (envolvimento cutâneo e hematológico) e síndrome antifosfolipídico, sob hidroxicloroquina 400 mg/dia, azatioprina (AZA)125 mg/dia e varfarina. Admitida na unidade cuidados intensivos por choque séptico devido a provável infeção respiratória. Apresentava quadro de febre persistente, astenia, tosse não produtiva com 2 meses de evolução, hipotensão e agravamento do estado geral. Do estudo destacava-se pancitopenia, PCR 181 mg/L, VS 88 mm/h, insuficiência renal aguda, anti-dsDNA negativo, complemento normal, hemoculturas positivas para Staphylococcus aureus e TC torácica a demonstrar nódulos e consolidações bilaterais. Foi iniciado tratamento com antibioterapia de largo espetro e suspensa a terapêutica com AZA, mantendo-se hipocoagulada com enoxaparina. Contudo, verificou-se agravamento clínico e disfunção multiorgânica com febre persistente, confusão mental, anemia não hemolítica com necessidade transfusional, disfunção hepática, hipertrigliceridemia (303mg/dL), hiperferritinemia (5318mg/dL) e espelenomegalia. Neste contexto, considerando o diagnóstico de LES (embora sem imunologia sugestiva de atividade da doença) e, infeção, suspeitou-se de LHH/SAM (H-score calculado em 221 pontos, probabilidade 96–98%)(2). Foi doseado o CD25 solúvel (37892U/mL), efetuado mielograma, que revelou sinais sugestivos de hemofagocitose, e biópsia medular óssea, que excluiu patologia hematológica primária. Iniciou-se metilprednisolona (1g iv em pulsos,3 dias) seguido de prednisolona oral (60 mg/dia) e imunoglobulinas humanas (2 g/kg, iv, 3 dias). Dada a melhoria clínica progressiva, a doente foi transferida para a enfermaria de Reumatologia ao 10º dia, tendo-se decidido pelo início de anakinra (100 mg/dia). Todavia, após 8 dias, apresentou agravamento da astenia, dor abdominal aguda, agravamento da anemia (Hb 5 g/dL) e prolongamento do apTT e TP. Realizou angio-TC abdominal que revelou extenso hematoma retroperitoneal e do reto abdominal no contexto de coagulopatia. A doente foi transferida para a unidade de cuidados intermédios, onde faleceu ao 23º dia de internamento apesar de todas as medidas instituídas.

Discussão: A LHH/SAM ocorre em 0.9 a 4.4% dos doentes com LES(3). Este caso demonstra a sua complexidade diagnóstica em doentes com LES e infeção ativa, mesmo na ausência de alterações analíticas sugestivas de atividade de doença. A imunosupressão precoce permitiu a reversão do estado inflamatório, mas a evolução posterior com hemorragia destaca a importância da vigilância de complicações tardias, como a coagulopatia. Esta, embora rara, está descrita em doentes com LHH/SAM, quer pela disfunção hepática associada, quer por coagulopatias de consumo, agravadas por terapêuticas imunossupressoras intensas. Acresce o provável contributo da necessidade de hipocoagulação pelo SAAF para tal desfecho. Conclui-se que a LHH/SAM é uma emergência reumatológica desafiante, onde é exigente o equilíbrio clínico entre a contenção da hiperativação imune, o manuseamento da doença de base e a prevenção de complicações secundárias graves.

129 - A CASE REPORT OF SCLEREDEMA AND RETROPERITONEAL FIBROSIS

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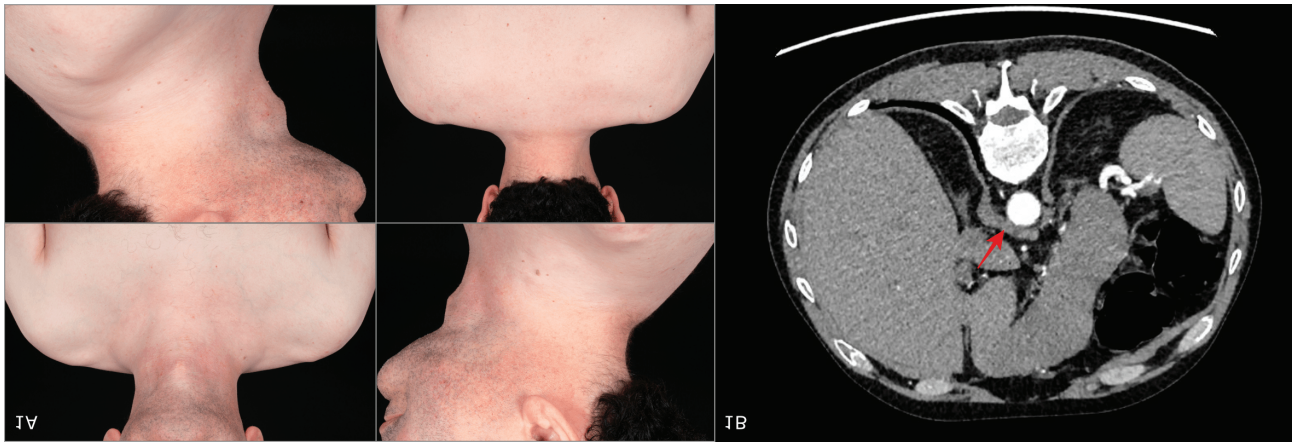
Introduction: Scleredema is a rare primary diffuse mucinosis, characterised by progressive, symmetrical skin hardening and thickening, most commonly involving the neck, shoulders, and upper trunk. It is part of the spectrum of scleroderma-like disorders. In contrast to systemic sclerosis, Raynaud's phenomenon and internal organ involvement are usually absent. It can be classified into three subtypes: type 1, associated with respiratory infections; type 2, associated with paraproteinaemia; and type 3, associated with diabetes mellitus. Retroperitoneal fibrosis (RPF) is a rare condition characterised by chronic inflammation and fibrosis of the retroperitoneum. Some cases are idiopathic, while secondary forms are usually associated with IgG4-related disease, infections, malignancies, radiotherapy and exposure to tobacco or certain medications. Although both conditions involve fibrotic processes, their coexistence has not been previously reported in the literature.

Case Report: We report the case of a 48-year-old male patient with a history of hypertension, who was referred to our tertiary care centre for evaluation. In November 2011, he first noticed neck stiffness and swelling that gradually extended to the shoulders, proximally to the elbows, and face (Figure 1A). The patient reported no Raynaud phenomenon, distal skin thickening, or other symptoms suggesting internal organ involvement. He is a non-smoker and has no exposure to chemicals or solvents. Laboratory investigations were reassuring, showing normal haemoglobin (13.8 g/dL), creatinine (84 µmol/L), erythrocyte sedimentation rate (5mm in the 1st hour), C-reactive protein (2mg/L), IgG4 [0.15 g/L (reference values 0-1.3 g/L)], fasting glucose and glycosylated haemoglobin (HbA1c 35 mmol/mol);



129 – Figure 1. 1A - skin thickening and hardening around the neck and upper trunk; 1B - CT showing retroperitoneal fibrosis (red arrow)

negative anti-nuclear antibodies (ANA) and extractable nuclear antigens (ENA). A low-grade IgG kappa paraprotein (2-4 g/L) was consistent with monoclonal gammopathy of undetermined significance (MGUS). The skin biopsy confirmed the diagnosis of scleredema, revealing thickened collagen fibres with mucoid degeneration and perivascular inflammation.

He was initially treated with prednisolone 30mg/day, oral methotrexate was initiated and titrated to 25mg weekly, and prednisolone was gradually tapered to 5mg/day. Despite treatment with methotrexate, the patient remained dependent on low-dose corticosteroids, experiencing relapses for doses below 5mg/day.

Cardiac assessment revealed a dilated left atrium and an estimated pulmonary artery systolic pressure of 49 mmHg on echocardiogram. A computed tomography pulmonary angiogram was performed to exclude thromboembolic disease before right heart catheterisation. While interstitial lung and thromboembolic disease were ruled out, the scan incidentally revealed retroperitoneal fibrosis around the abdominal aorta (Figure 1B). Due to the high risk of bleeding, biopsy of retroperitoneal fibrosis was not performed. Right heart catheterisation (RHC) confirmed elevated pressures initially, which normalised with blood pressure control, suggesting elevations were secondary to systemic hypertension rather than pulmonary hypertension.

Conclusion: We present a unique case of scleredema associated with retroperitoneal fibrosis in a patient with MGUS. This case highlights the need to consider scleredema in the differential diagnosis of skin thickening syndromes, particularly in patients with MGUS, and underscores the importance of thorough systemic evaluation to identify associated fibrosing conditions such as RPF.

130 - WHEN THE EYE COMES FIRST – CASE REPORT OF SCLERITIS AS THE FIRST MANIFESTATION OF RHEUMATOID ARTHRITIS

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Introduction. Rheumatoid Arthritis (RA) is an inflammatory systemic disease with several extra-articular manifestations, including eye involvement. Scleritis is associated with systemic disease in 35-48% of patients and RA is the most common diagnosis among them. In most patients, RA diagnoses precedes scleritis presentation.

Case Report. We present the case of a 52-years-old male patient in which disease onset starts in 2008, at 35 years of age, with a scleritis episode of the left eye that progressed to ulceration of the cornea and scleromalacia. Corneal transplant was performed but not successful as patient rejected the graft, resulting in severe visual impairment in the left eye. At time of scleritis presentation, the only relevant systemic symptoms mentioned were intermittent and short-lasting arthralgias. Evaluation by rheumatology at the time showed no signs of active synovitis but found positive rheumatoid factor (RF) and anti-cyclic citrullinated peptide antibodies (anti-CCP) titers. Oral methotrexate was then started but patient follow-up was compromised as medical appointments were not attended. In 2023, after discontinuation of this treatment due to poor adherence, patient presented with symmetrical inflammatory synovitis of the wrists, metacarpophalangeal

joints, and knees, along with elevated inflammatory markers on laboratory testing. Methotrexate therapy was then restarted, subcutaneously, and disease activity remains controlled ever since.

Discussion. Given the potential association between scleritis and rheumatoid arthritis (RA), a thorough medical history is essential to rule out polyarthritis or other systemic manifestations of RA. Additional diagnostic work-up, including laboratory testing for RF and anti-CCP, as well as immunologic panels for other rheumatologic diseases, is critical for identifying underlying systemic disease. Although RA typically precedes the onset of scleritis, our clinical case demonstrates that this sequence can be reversed. Patients presenting with scleritis and positive serologic markers can benefit from early referral to rheumatology.

132 - CUBITAL TUNNEL SYNDROME – CLINICAL CASE REPORT OF A NOT SO COMMON CAUSE

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Introduction. Cubital Tunnel Syndrome (CuTS) is a condition caused by compression or irritation of the ulnar nerve at the elbow, specifically where it passes through the cubital tunnel. Presentation includes numbness or tingling of the 4th and 5th fingers, worsened by elbow flexion, and regional muscle weakness and atrophy. Tinel's sign performed at ulnar groove is usually positive and electromyography confirms signs of ulnar dysfunction at the elbow. Treatment can be either conservative or surgical. CuTS' prevalence is rather elevated (about 5.9% of the population) but it is mostly associated with repetitive elbow movements, local trauma, anatomical variations of cubital tunnel or elbow synovitis or osteoarthritis. Rarer causes include compressing masses and systemic diseases like diabetes that make the nerve more susceptible to injury.

Clinical case report. We present the case of a 73-year-old man, with a medical history of osteoarthritis of the knee, hands and lumbar spine, and osteoporosis with a Colles' fracture of the left wrist treated conservatively in 2022. In March 2024, the patient presented with paresthesia on the palmar and dorsal aspects of the 4th and 5th fingers of the left hand that persisted for months and was worsened by elbow flexion. Physical examination showed hypothenar eminence and regional interossei atrophy and Tinel's sign was positive over the ulnar groove. Electromyography suggested findings of chronic left ulnar mononeuropathy localized in the forearm distal to the elbow, with both an axonal and demyelinating component, and elbow ultrasound showed a 28x12x7mm mass inside the cubital tunnel. The patient was then sent to plastic surgery consultation and did wrist X-ray and MRI which ruled out ulnar nerve compression at the wrist. With these findings, the plastic surgery team opted to perform cubital tunnel surgery in February 2025 with mass excision and ulnar nerve neurolysis, which underwent without complications. Biopsy report of the mass ruled out malignancy and revealed a vascular malformation composed by a combination of thickened wall blood vessels and capillaries of arterial, venous and lymphatic nature. After surgery, symptoms were strongly relieved although patient still reports some numbness.

Conclusion. CuTS is a fairly common condition in the general population. In a 73-years-old male with medical history of osteoarthritis, most probable causes would include elbow osteophytes or joint deformity. However, after ultrasound evaluation a compressing soft tissue mass was evident. While vascular malformations are not a common cause of CuTS, they should be considered in the differential diagnosis and soft tissue comprehensive imaging should be performed. Early recognition and appropriate management are crucial for optimal patient outcomes.

135 - IMMUNE CHECKPOINT INHIBITORS AND THE COST OF ACTIVATION: A CASE SERIES AND CLINICAL REFLECTIONS

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Introduction: Immune checkpoint inhibitors (ICIs), particularly PD-1 inhibitors such as pembrolizumab and nivolumab, have revolutionized cancer therapy. These monoclonal antibodies inhibit the interaction between the PD-1 receptor and its ligands, PD-L1 and PD-L2, thereby enhancing T-cell-mediated antitumor responses. Despite their generally good tolerability, ICIs can cause immune-related adverse events (irAEs). This case series details inflammatory arthritis in solid tumor patients treated with PD-1 inhibitors, highlighting clinical patterns, therapeutic approaches and the need for individualized management.

Case series: Case 1: A 77-year-old man with metastatic renal cell carcinoma developed high-titre seropositive rheumatoid arthritis two months after starting pem-

brolizumab, which was then replaced with second-line agents due to joint toxicity. Methotrexate and corticosteroids were initiated, with temporary improvement, however, a clinical flare subsequently required escalation to biologic therapy with infliximab.
Case 2: A 47-year-old woman receiving pembrolizumab for triple-negative breast cancer developed asymmetric oligoarthritis and extensor tenosynovitis. Hydroxychloroquine and nonsteroidal anti-inflammatory drugs were prescribed, with partial symptom control. Corticosteroids were recommended but declined.
Case 3: A 65-year-old woman who had been followed in rheumatology outpatient for seronegative axial spondyloarthritis, and who had recently initiated pembrolizumab for clear cell renal carcinoma, experienced worsening of previously controlled bilateral knee pain. Methotrexate dosage was progressively increased with clinical benefit. Corticosteroids were recommended but declined. The patient reported symptomatic improvement under maintained methotrexate therapy alone.
Case 4: A 78-year-old woman with squamous cell carcinoma of the gingiva developed acute monoarthritis of the right knee during pembrolizumab treatment, which led to its interruption. Symptoms improved with systemic corticosteroids, however, persistent swelling required continued management with a low-dose oral regimen.
Case 5: A 69-year-old woman with pleural mesothelioma had a longstanding history of occasional inflammatory back pain and arthralgias involving shoulders, knees, and small hand joints. Shortly after initiating nivolumab, these symptoms significantly worsened, with diffuse joint swelling and functional impairment. Despite treatment with methotrexate and corticosteroids, she experienced symptom flares associated with steroid tapering. Later diagnosed with a myxofibrosarcoma, she ultimately died due to febrile neutropenia as a complication of chemotherapy.
Case 6: A 58-year-old man with long-standing seropositive rheumatoid arthritis remained in sustained remission after discontinuation of methotrexate following a lung adenocarcinoma diagnosis. The patient underwent six cycles of pembrolizumab without experiencing a musculoskeletal flare but ultimately suffered a fatal outcome due to the natural progression of his oncological disease.
Discussion: PD-1 inhibitors enhance antitumor immunity but can also trigger inflammatory events. In this series, clinical presentations included seropositive polyarthritis, oligoarthritis, and tenosynovitis. Management strategies comprised corticosteroids, csDMARDs, and escalation to biological therapy. Most achieved symptom control, and treatment continuation was possible in selected cases. Interestingly, one patient with stable rheumatoid arthritis had no flare, suggesting autoimmune history alone may not predict irAEs.

137 - PSEUDO-RHEUMATOID NODULES: WHEN HISTOPATHOLOGY CONFUSES US

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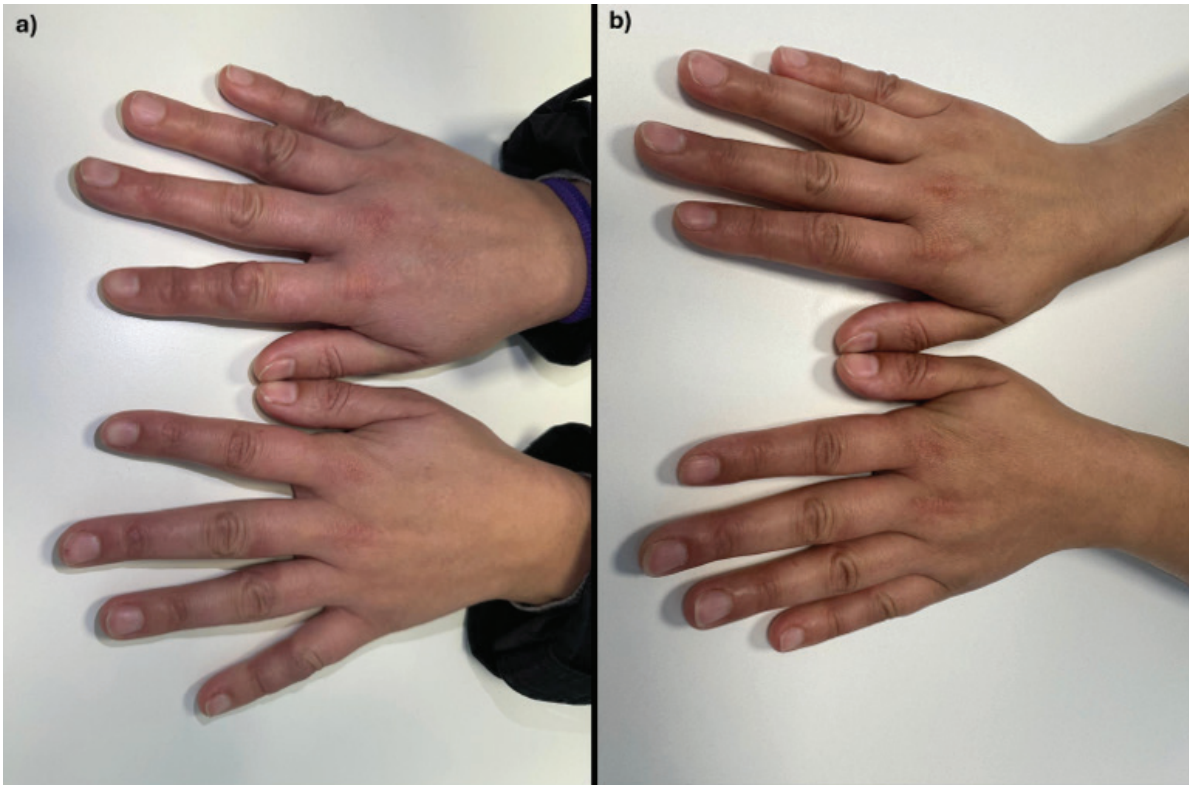
Introduction: Subcutaneous rheumatoid nodules (RNs) are a common extra-articular manifestation among rheumatoid arthritis (RA) patients. They manifest as a firm, nontender, and moveable mass, often found on extensor surfaces. The diagnosis is clinical, however, there are RN mimetics, so sometimes histology may be required.

Annular granuloma (AG) is a benign, typically self-limited granulomatous skin disorder, which histology may resemble RNs, with the main difference being the presence of mucin deposition. Their aetiology remains unclear, though it may be associated with diabetes mellitus, paraneoplastic syndromes, thyroid disorders, and certain medications. AG associated with topiramate is a rare adverse event, with only a limited number of cases reported in the literature.
Case: We present the case of a 40-year-old woman with a history of chronic migraine, treated with topiramate100mg/day since June 2022. In January 2023, she noticed the appearance of small, non-painful, non-pruritic nodules and hyperpigmented papules, on the surface of proximal interphalangeal joints of the 2nd, 4th, and 5th fingers bilaterally and the distal interphalangeal joint of the 2nd finger of the right hand (Fig.1a), as well as on the palmar side of the metacarpophalangeal joints of the 2nd and 4th fingers of the left hand. The patient had no limitation of active finger flexion and denied arthralgia or systemic symptoms. A Dermatology consultation was sought, and a skin biopsy was performed. Histological analysis revealed fibrous connective tissue with several nodules composed of palisading histiocytes surrounding areas of necrosis, compatible with RNs. Given this finding, she was referred to Rheumatology. Physical examination revealed the described cutaneous lesions, which were not typical of rheumatoid nodules, and there was no evidence of peripheral arthritis. Hand radiographs showed no relevant abnormalities. Autoimmune screening revealed negative results for anti-cyclic citrullinated peptide antibodies, rheumatoid factor, antinuclear antibodies and anti-extractable nuclear antigen antibodies. Serum protein electrophoresis, uric acid, angioten-



137 – Figure 1. Nodules and hyperpigmented papules, on the surface of proximal interphalangeal joints of the 2nd, 4th, and 5th fingers bilaterally and the distal interphalangeal joint of the 2nd finger of the right hand (a) suggestive of annular granuloma. Complete resolution of the lesions after topiramate suspension (b).

sin-converting enzyme levels and thyroid-stimulating hormone were normal. Given the suspicion of AG and a temporal relationship between the topiramate treatment and the occurrence of the lesions, a review of the histological specimen was requested, which confirmed the presence of histiocytic cells with palisade arrangement and necrobiotic granulomas with marked mucinous deposits and associated fibrosis, consistent with the deep variant of AG. Due to the possibility of topiramate-induced AG, the drug was discontinued in January 2025. One week after withdrawal, there was marked improvement in the hand lesions (Fig. 1b) and complete resolution was observed after 2 months, which confirmed the diagnostic suspicion.

Discussion: As an extra-articular manifestation, RNs occur in patients with RA, particularly in those who are seropositive. AG is an important differential diagnosis of RNs, with similar histopathologic features, such as the immune-mediated granulomatous process with histiocytic infiltrate in a palisading pattern. However, the presence of mucin deposition is a fundamental clue, being present mainly in AG. AG can develop as an adverse reaction to topiramate, being exceedingly rare, with the underlying pathophysiological mechanisms not yet fully understood. In reported cases, discontinuation of the drug typically leads to resolution of skin lesions within weeks, and recurrence is uncommon.

141 - WRIST DROP: A RARE CASE OF IATROGENIC RADIAL MONONEUROPATHY

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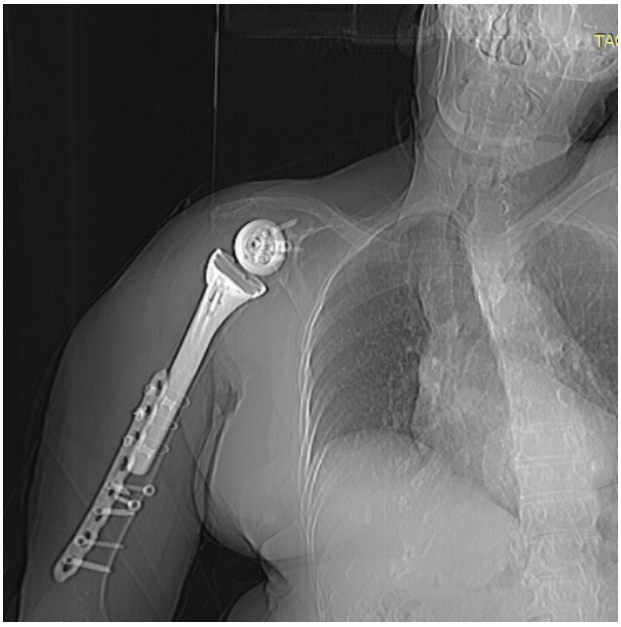
Introduction: Orthopedic procedures are essential in managing acute conditions such as fractures, but also in the treatment of chronic and deforming rheumatic disorders, with the aim of restoring anatomy, improving function and relieving pain. However, surgical complications are relatively common and may arise in both the early and late postoperative periods. The most frequent early/subacute complications include infection, bleeding, pain, joint stiffness and nerve injury. Chronic infection, prosthetic failure and periprosthetic fractures are among the most significant delayed complications.



141 – Figure 1. CT scan of right shoulder and arm

We present a clinical case illustrating a rare, late complication associated with orthopedic surgery, aggravated by periprosthetic fracture occurring at the time of surgery.

Case Report: We describe a 82-year-old female patient, with a past medical history of calcium pyrophosphate deposition disease (CPPD), osteoporosis (treated with Denosumab) and polyosteoarthritis, involving both axial and peripheral joints, with bilateral glenohumeral rapidly destructive osteoarthritis, secondary to CPPD. Given this, she undergone left shoulder arthrodesis in 2019 and right shoulder arthroplasty in 2016, the latter complicated by an intraoperative periprosthetic fracture requiring osteosynthesis and cerclage, during the same surgical procedure.

At present, the patient was evaluated for an 8-month history of right wrist drop, accompanied by pain and significant functional impairment in activities of daily live.

The patient underwent a physical rehabilitation program, which led to improvement in shoulder pain, though without clinical or functional improvement in the hands. Physical examination confirmed persistent inability to actively extend the right wrist, with evident wrist drop.

In this context electromyography (EMG) was performed, revealing severe axonal injury of the right radial nerve. A cervical spine computed tomography (CT) scan was subsequently conducted, which demonstrated multiple signs of uncovertebral arthrosis with likely nerve root involvement at C3–C4 (right), C4–C5 (left), and bilaterally at C5–C6 and C6–C7. A central osteophyte-disc complex at C5–C6 was also identified, potentially causing mild spinal cord indentation and possible C8 root compression.

Complementary ultrasonography of the limb revealed osteosynthesis material with probable displacement, with possible evolvement of the radial groove. New signs of fracture could not be excluded in this context. A new CT scan of the right shoulder and arm confirmed the presence of a reverse prosthesis, moderate joint effusion between prosthetic components, and multiple discontinuities along the humeral shaft as sequelae of a previous fracture, without features of recent fracture. One of the screws was found to be extrinsic to the humeral cortex, in close proximity to the topography of the radial nerve. Thus, the patient was diagnosed with a radial mononeuropathy secondary to prosthetic displacement and nerve compression. A newly developed periprosthetic fracture as a cause of nerve injury was excluded.

The patient continued regular rehabilitation and was referred for reevaluation by Orthopedic Surgery due to the late-onset radial mononeuropathy, secondary to nerve compression by the prosthetic and cerclage material in the right shoulder.

Conclusion: This case aims to highlight a rare, late, iatrogenic mononeuropathy of the radial nerve, secondary to orthopedic intervention – right shoulder arthroplasty, complicated by intraoperative periprosthetic fracture, treated with osteosynthesis and cerclage.

Images:

144 - ADALIMUMAB-INDUCED NEUTROPHILIC PANNICULITIS: A RARE CUTANEOUS ADVERSE REACTION

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Introduction: Anti-TNF agents are widely used in rheumatologic diseases, including psoriatic arthritis.1 Paradoxical immune-mediated conditions may occur during treatment, often affecting the skin. Psoriasiform eruptions are the most common, while other paradoxical skin reactions, such as neutrophilic dermatoses, are rarer.2 Among these, neutrophilic panniculitis is exceptionally rare, characterized by neutrophilic infiltration of subcutaneous fat tissue.3

Clinical case: A 52-year-old woman with peripheral psoriatic arthritis, diagnosed in 2022, was regularly fol-

lowed in the Rheumatology Department. She was initially treated with methotrexate but, due to persistent articular and cutaneous disease activity, adalimumab was added in November 2023. Three weeks after the first dose, she developed papulopustular skin lesions on one lower limb, which worsened and extended to all limbs. No systemic symptoms were reported. Laboratory results showed leukocytosis (10,850/µL) with neutrophilia, ESR 15mm/h, CRP 2.07mg/dL, and negative ANA and IGRA. Adalimumab was discontinued in August 2024. At dermatology evaluation in November 2024, she presented with multiple papulopustular lesions, some with serosanguineous discharge; most were superficial but some lesions appeared nodular and tender (Fig.1). A skin biopsy was performed. She was treated with oral prednisolone 60 mg/day and colchicine 1mg/day, rapidly tapered over two weeks, resulting in resolution. Histopathology revealed a predominantly neutrophilic infiltrate in the mid and deep dermis and the superficial hypodermis, with septal and peripheral lobular involvement, supporting the diagnosis of anti-TNF-induced neutrophilic panniculitis. Given the persistence of arthritis, treatment with ustekinumab was proposed.

Discussion: Paradoxical skin reactions occur in 0.6 to 5.3% of patients on anti-TNF therapy. Although the exact mechanisms remain unclear, current hypotheses point to a complex interplay between host-specific factors and biologic agent-induced shifts in cellular immune-response patterns. Neutrophilic dermatoses typically show pustules as a hallmark. Dermatologic evaluation and skin biopsy are essential for correlating clinical presentation with histopathology.2 Management of paradoxical cutaneous reactions includes topical therapies and systemic corticosteroids. More severe or refractory cases may require anti-TNF discontinuation and systemic immunosuppressants. Antineutrophilic drugs such as colchicine may also play a role.2,4 Anti-TNF-induced panniculitis is an exceedingly rare adverse event, with few cases reported. This case illustrates neutrophilic panniculitis secondary to adalimumab, marked by rapid onset and resolution after drug withdrawal and corticosteroid plus colchicine therapy.

Conclusion: Cutaneous adverse events often represent diagnostic challenges and frequently require skin biopsy for confirmation. This case highlights the importance of multidisciplinary collaboration with dermatology and the need to consider paradoxical reactions when new or changing skin lesions occur during biologic treatment.

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145 - ENVOLVIMENTO MIOCÁRDICO PRECOCE EM DOENTES COM ESCLEROSE SISTÉMICA

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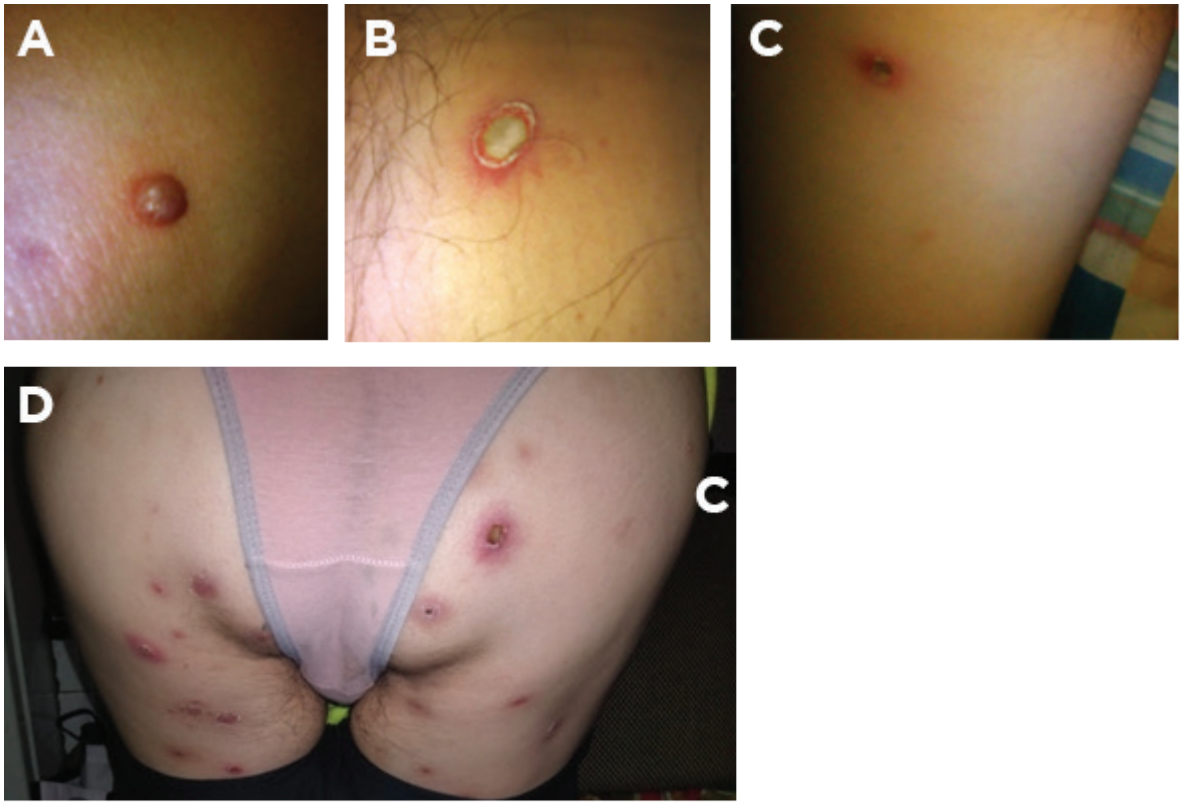
Introdução: Pacientes com esclerose sistémica (ES) e envolvimento cardíaco sintomático apresentam mau prognóstico, com taxa de mortalidade aos 2 anos de 60%. O envolvimento cardíaco parece ser mais severo em homens, e mais frequentemente associado a hipertensão pulmonar. No entanto, o envolvimento miocárdico é cada vez mais reconhecido nestes doentes.

Caso 1: Mulher de 29 anos com história de overlap entre ES e polimiosite conhecida desde 2019 (positividade para anticorpos anti-SCL70 e anti-PM/SCL100), com envolvimentos cutâneo difuso, articular, vascular, gastrointestinal e muscular, medicada com nifedipina 30mg/dia e metotrexato 20mg/semanal. Apresentou episódio de dor torácica aguda em 2022, com auscultação cardíaca arrítmica e crepitações pulmonares bibasais. À admissão, exibia taquicardia (150 bpm) e elevação do BNP (1002 pg/mL). O ecocardiograma revelou compromisso da fração de ejeção ventricular (FEV), sem alterações no cateterismo cardíaco. Realizou RM cardíaca, com descrição de cardiomiopatia dilatada, tendo a biópsia miocárdica demonstrado miocardite linfocítica. O painel de vírus cardiotrópicos requisitado foi negativo. Assumindo-se atingimento miocárdico pela doença, iniciou tratamento com ciclofosfamida (CYC) mensal (1g), com uma dose cumulativa de 6g, com posterior alteração para micofenolato mofetil (MMF) 2g/dia, levando a melhoria clínica e analítica inicial.

A doente foi readmitida em fevereiro de 2025 por insuficiência cardíaca agudizada de causa não esclarecida, com FEV estimada em 9%. Apesar da otimização da terapêutica modificadora de prognóstico, diurética e imunossupressora, foi verificado o óbito 22 dias após a admissão hospitalar.

Caso 2: Homem de 40 anos sem antecedentes pessoais relevantes, admitido no Serviço de Cardiologia em 2022 por dispneia para pequenos esforços, ortopneia



145 – Figure 1. (A, B, C, D, and E) Multiple skin lesions in different stages

e edemas periféricos dos membros inferiores, com 1 semana de evolução. Paralelamente, o doente referia queixas com 5 meses de evolução de poliartralgias simétricas e aditivas, com noção de tumefação articular a acometer cotovelos, punhos, metacarpofalângicas (MCFs) e tibiotársicas, bem como fenómeno de Raynaud. À observação, apresentava puffy hands e esclerodactilia proximal às articulações MCFs. À admissão, verificada elevação do BNP (286 pg/mL) e troponina I (696 ng/mL), bem como anticorpos anti-nucleares positivos (1/1000 padrão mosqueado), com positividade para anticorpos anti-histonas e anti-RNA polimerase III. Prosseguiu-se na marcha diagnóstica, tendo sido realizado ecocardiograma com achados sugestivos de cardiopatia infiltrativa. A RM cardíaca revelou compromisso ligeiro/moderado da FEV (42%), com fibrose focal de etiologia não-isquémica, em contexto provável de miocardiopatia inflamatória. Realizou ainda videocapilaroscopia que evidenciou alterações enquadráveis em padrão tipo esclerodérmico em fase inicial. Pelo diagnóstico de ES com manifestação inaugural cardíaca, o paciente iniciou CYC (1g) que cumpriu num total de 6 meses. Atualmente medicado com ácido micofenólico 1440 mg/dia, com recuperação total da função sistólica biventricular.

Discussão: O envolvimento miocárdico na esclerose sistémica pode manifestar-se precocemente, sendo que a sua deteção e tratamento agressivo com imunossupressores podem alterar o curso da doença. No entanto, a gravidade do envolvimento cardíaco nestes doentes dita um mau prognóstico, apesar de todos os esforços de otimização terapêutica, o que evidencia a necessidade de vigilância ativa e gestão multidisciplinar no follow-up destes doentes.

146 - PET/CT IN PRIMARY HYPERPARATHYROIDISM: A CASE REPORT

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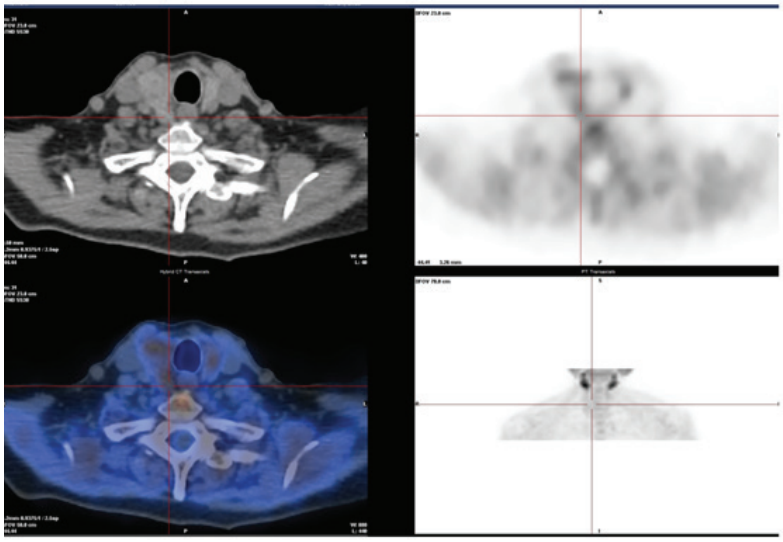
Introduction: Primary hyperparathyroidism (PHPT), most commonly caused by a parathyroid adenoma, can lead to secondary osteoporosis. Parathyroidectomy is the definitive treatment and improves BMD and bone microarchitecture. Standard imaging modalities, cervical ultrasound (US) and technetium-99m sestamibi scintigraphy, are effective for the detection of typical



145 – Figure 1. Fluorine-18 fluorocholine PET/CT scan showing a nodular lesion, measuring approximately 10x9 mm, posterior to the upper half of the right thyroid lobe, with mild radiotracer uptake, suggestive of a right parathyroid adenoma.

adenomas but less reliable for ectopic, small, or multiple gland disease, especially in the presence of thyroid pathology. Fluorine-18 fluorocholine (18F-FCH) PET/CT has emerged as a highly sensitive modality for localizing parathyroid adenomas, outperforming both US and scintigraphy in recent studies.

Case: We present the case of a 67-year-old woman referred to Rheumatology for severe osteoporosis refractory to treatment. She had been treated in primary care with oral bisphosphonates, with calcium and vitamin D supplementation, for four years. However, at the end of that period, the patient sustained a right wrist fracture and the bone densitometric reevaluation showed persistently low T-scores [L2-L4 with -3.7, total femur -3.3, femoral neck -3.8 and distal radius -3.0], despite good therapeutic compliance. Laboratory workup showed elevated parathyroid hormone (PTH: 109 pg/mL) with increased ionized calcium (2.70 mEq/L) but normal total calcium (4.9 mEq/L), 25-OH-vitamin D (41 ng/mL) and phosphorus (3.4 mg/dL). Alkaline phosphatase, albumin, thyroid stimulating hormone, serum protein electrophoresis and magnesium were normal. Bone turnover markers (β-crosslaps and osteocalcin) were within the normal range, and a 24-hour urine study revealed normal excretion of calcium, phosphorus, and magnesium. Given the findings of hyperparathyroidism, the patient underwent cervical US, which revealed multiple thyroid nodules, the biggest in the right lobe, but no abnormalities in the parathyroid glands. Scintigraphy evidenced a small focus of increased radiotracer uptake superior to the right thyroid lobe, posterior to the hyoid bone, suggesting the presence of an abnormal parathyroid gland. Therefore, the patient was referred to General Surgery. Because the blood test only revealed elevated ionized calcium, additional investigation was conducted. A neck CT scan showed no significant findings, and an abdominal CT ruled out nephrolithiasis. To clarify the findings, an 18F-FCH PET/CT scan was performed, which confirmed a nodular lesion measuring approximately 10×9 mm, located posterior to the upper half of the right thyroid lobe, with mild radiotracer uptake, highly suggestive of a right parathyroid adenoma. The patient underwent successful parathyroidectomy of the right superior parathyroid gland, and ionized calcium normalized the following day.

Discussion: With this case, the authors aim to highlight the new clinical utility of 18F-FCH PET/CT in the study of PHPT, particularly when different conventional imaging methods are incongruous. Its superior diagnostic performance supports its use as a second-line or problem-solving modality in the diagnosis and preoperative workup of PHPT, facilitating targeted surgical approaches, reducing the need for bilateral neck exploration and optimizing patient outcomes.

147 - LATE DIAGNOSIS OF AXIAL SPONDYLOARTHRITIS IN PATIENTS WITH GIANT CELL ARTERITIS

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Introduction: Giant cell arteritis (GCA) is a large vessel

vasculitis (LVV) that affects individuals over 50 years old. Coexisting LVV has previously been seldom reported in cases of established spondyloarthritis (SpA). Overlapping immunopathogenic pathways may allow for this coexistence. Herein we describe two patients with GCA and late diagnosis of axial SpA.

Case 1: A 75-year-old man presented with shoulder and pelvic girdle pain, prolonged morning stiffness and elevated ESR (81mm/h) and CRP (15.5mg/dL). A diagnosis of polymyalgia rheumatica (PMR) was established and treatment with prednisolone (15mg qd) ensured clinical remission, without recurrence after glucocorticoid (GC) withdrawal.

Five years later, he presented with sudden unilateral vision loss, frontal headache and elevated ESR and CRP. Ophthalmologic evaluation suggested arteritic anterior ischemic optic neuropathy and, given the high index suspicion for GCA, pulse treatment with methylprednisolone (1g qd for 3 days) followed by prednisolone (60mg qd) was started. Temporal artery ultrasound revealed nonspecific intima-media thickening that normalized on follow-up.

During GC tapering, despite methotrexate therapy, the patient experienced PMR-like recurrences with predominantly pelvic girdle pain and ankle arthritis. PET-CT excluded active LVV. Unexpectedly, pelvic radiography revealed partial sacroiliac joint ankylosis and chronic active sacroiliitis was confirmed by MRI.

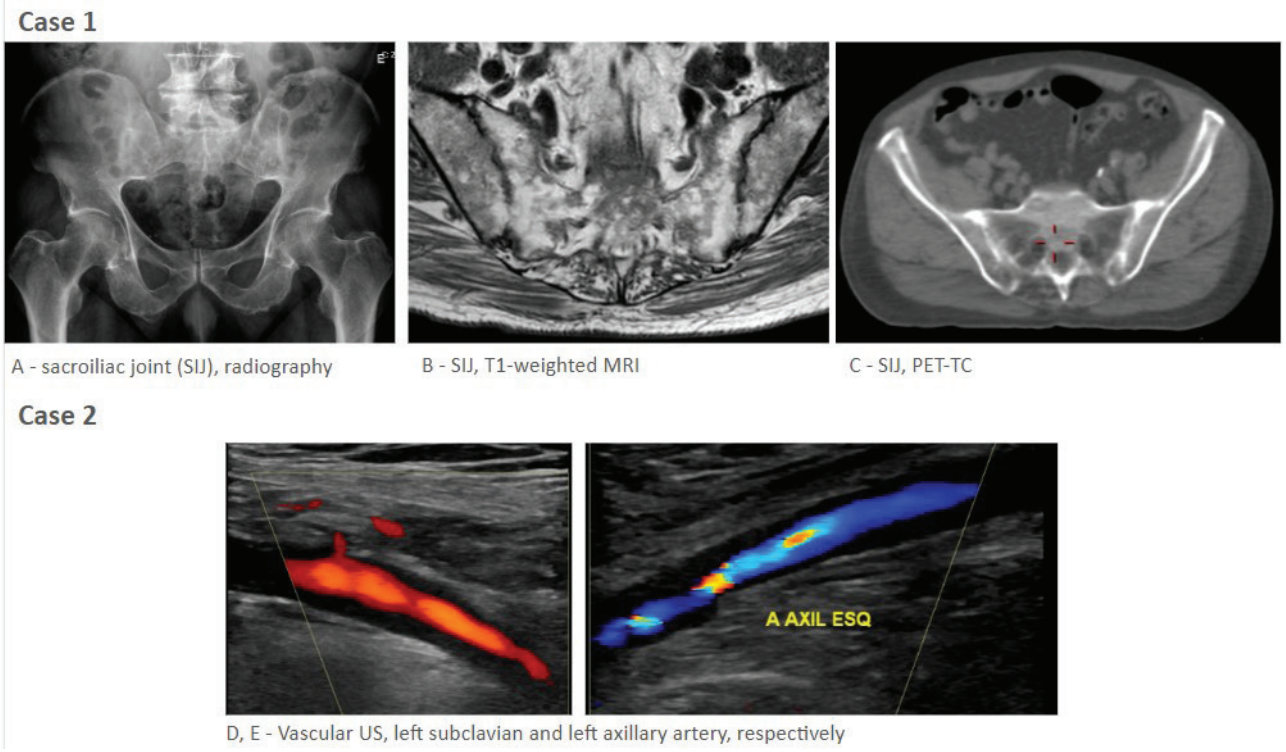
After the diagnosis of axial SpA (HLA-B27 positive) with high-disease activity (ASDAS-CRP 4.6), treatment with adalimumab combined with methotrexate ensured sustained GC-free clinical remission.

Case 2: A 78-year-old woman presented with a 2-year diagnosis of relapsing LV-GCA. She first presented with a history of pelvic girdle and low back pain, morning stiffness and elevated ESR (81mm/h) and CRP (9.2mg/dL). Presence of the ultrasonographic halo sign of the axillary and subclavian arteries by US confirmed the diagnosis and treatment with prednisolone (60mg qd) was started. After failure to taper GC below 20mg qd, methotrexate was initiated with partial benefit.

On clinical review, the patient reported long standing low back pain with mixed rhythm. Follow-up vascular US revealed chronic halo of the subclavian, axillary and left temporal arteries. PET-CT excluded active LVV but exposed chronic sacroiliitis, confirmed by MRI.

After the diagnosis of axial SpA (HLA-B27 negative), treatment with adalimumab was started with clinical improvement and GC tapering (5mg qd). ESR and CRP remained mildly elevated and PET-CT reassessment showed mild-to-moderate FDG uptake in the thoracic aorta, suggestive of subclinical aortitis.

Conclusion: Evidence suggesting an association between LVV and SpA is emerging, although GCA-SpA coexistence remains rarely reported. We describe two



147 – Figure. Case 1: imaging of SI joints. Case 2: vascular ultrasoun

additional cases, adding to the few published to date. Both conditions may share genetic and immunological pathways. While HLA-B27 is commonly associated with SpA, possible links with HLA-DRB104 and HLA-DRB101 – both strongly related to GCA – have been reported. Th1/Th17-mediated inflammation with elevated TNF-α, IL-6, IL-17, and IL-23 also plays a key role in both diseases.

These cases highlight the need to consider SpA in GCA patients with persistent inflammation or atypical musculoskeletal symptoms, particularly in elderly patients with incomplete treatment response.

148 - LOMBOSSACRALGIA PERSISTENTE EM DOENTE COM HISTÓRIA DE BRUCELOSE

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Introdução: A brucelose é uma zoonose bacteriana causada por espécies do género Brucella, geralmente adquirida por contacto com produtos de origem animal contaminados ou através da ingestão de laticínios não pasteurizados. A apresentação clínica é variada, podendo evoluir com complicações osteoarticulares em até 40% dos casos, sendo a artrite periférica, espondilodiscite e sacroileíte as manifestações mais frequentemente reportadas. Embora o tratamento antibiótico seja geralmente eficaz, sintomas residuais como artralgias podem persistir, levantando dúvidas entre possível recidiva infecciosa e sequelas pós-infecciosas.
Caso clínico: Mulher de 67 anos, com história médica prévia de Diabetes Mellitus Tipo 2, Hipertensão Arterial, Síndrome da Apneia Obstrutiva do Sono e quisto ovárico submetido a remoção cirúrgica, sem história de doença inflamatória intestinal ou psoríase. Em 2017, apresentou quadro de poliartralgias simétricas dos punhos e pequenas articulações das mãos e ombros, bem como gonalgia e coxalgia direitas persistentes de ritmo misto. Referia ainda lombossacralgia de ritmo inflamatório, associada a astenia e febre. Após estudo complementar, foi diagnosticada brucelose e iniciada terapêutica com doxiciclina durante 6 semanas, com resolução do quadro infecioso agudo e melhoria das queixas. Cerca de um ano após o tratamento, apresentou-se de novo com queixas de poliartralgias e lombossacralgia esquerda de ritmo misto com cedência apenas parcial a anti-inflamatório, bem como astenia, com reação de Wright positiva (1:160), motivo pelo qual foi internada por suspeita de recidiva infeciosa. Ao exame objetivo não apresentava artrite periférica mas sim manobras de FABER e compressão direta da sacroilíaca esquerda positivas. Foi realizada RMN das sacroilíacas, que demonstrou edema medular ósseo em ambas as vertentes da articulação sacroilíaca esquerda, compatível com sacroileíte assimétrica. Completou então 12 semanas de tratamento com doxiciclina e rifampicina com boa evolução clínica e analítica. A doente foi orientada para Consulta de Reumatologia após resolução da recidiva infeciosa por manter lombossacralgia mecânica, interpretando-se o quadro em contexto de sequela pós-infecciosa, motivando o seu acompanhamento multidisciplinar, com necessidade de analgesia otimizada e medidas de reabilitação.
Discussão: Este caso sublinha não só a importância de um diagnóstico precoce e adequado da brucelose, como também chama a atenção para a possibilidade de complicações pós-infeciosas de natureza crónica. Apesar do tratamento antibiótico dirigido e da resolução aparente da infeção ativa, a doente desenvolveu lombossacralgia crónica, compatível com um quadro de sequela pós-infeciosa. Este tipo de manifestação, embora menos descrito na literatura, pode ter um impacto funcional e psicossocial relevante, exigindo uma abordagem terapêutica centrada no controlo sintomático e reabilitação. O reconhecimento destas complicações tardias é fundamental para um seguimento clínico adequado e para a otimização da qualidade de vida dos doentes.

150 - O IMPACTO TERAPÊUTICO DE SUSPEITAR DE SÍNDROMES DE SCHNITZLER E SCHNITZLER-LIKE

Ana Sá[1], Sara Amaro Lopes[1], Carlos Marques-Gomes[1,2], Bruno Miguel Fernandes[1, 2], Salomé Garcia[1, 2], Miguel Bernardes[1, 2], Lúcia Costa[1, 2]
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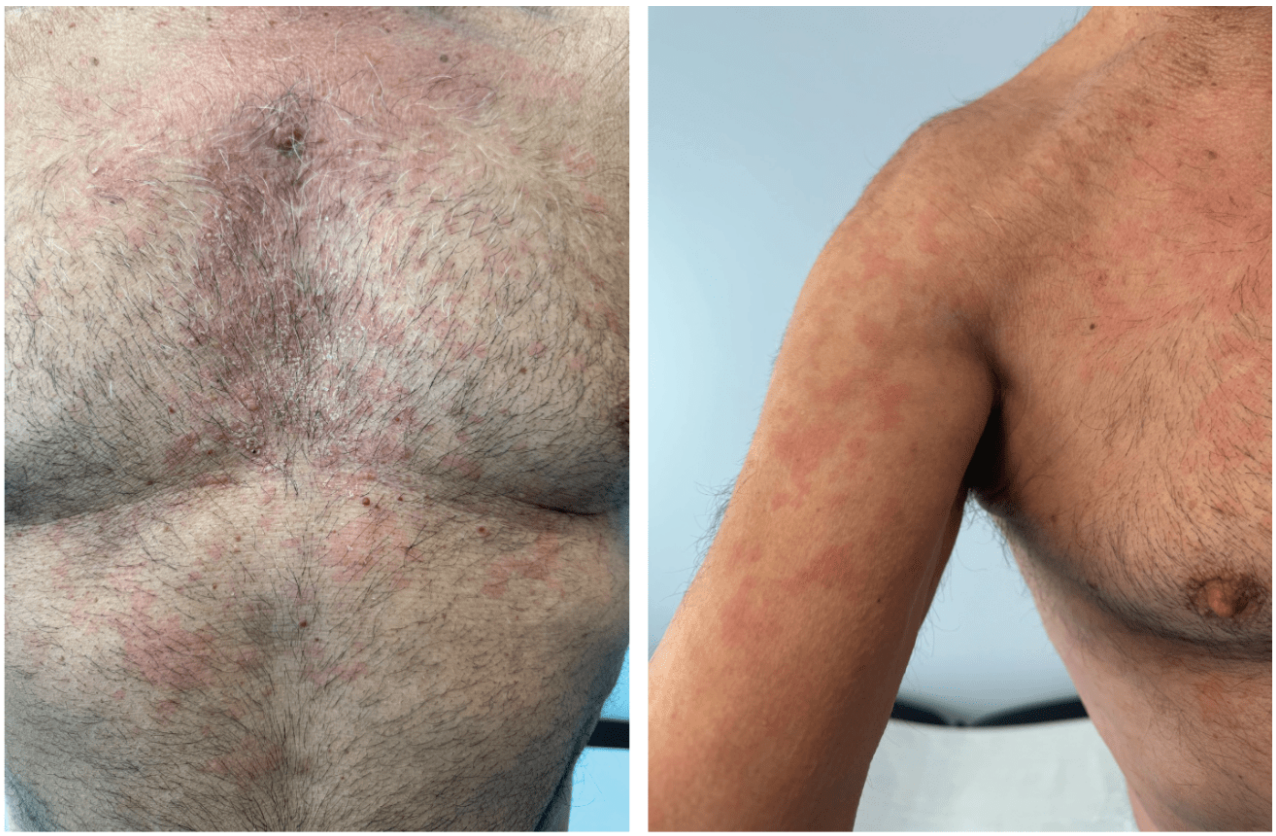
Introdução: A Síndrome de Schnitzler é uma doença rara de início na idade adulta, frequentemente subdiagnosticada. As suas principais características incluem erupção urticariforme, febre recorrente, artralgias ou artrite, gamopatia monoclonal de significado indeterminado (MGUS) e inflamação sistémica. A importância do seu reconhecimento reside na resposta proeminente ao bloqueio da interleucina (IL)-1 ou IL-6. As síndromes Schnitzler-like, que não preenchem os critérios diagnósticos clássicos, também apresentam uma excelente resposta a estes fármacos.
Caso 1: Homem de 68 anos, com antecedentes de hipertensão arterial, dislipidemia e gota, foi referenciado à

consulta de Reumatologia em 2022 devido a episódios frequentes de artralgias nas pequenas articulações das mãos e punhos, oligoartrite, bem como episódios de exantema eritematoso e pruriginoso disseminado, previamente diagnosticado como urticária. Além disso, o doente relatava episódios transitórios de febre de baixa intensidade. As análises laboratoriais revelaram elevação dos marcadores inflamatórios (leucócitos 14.890x10^9/L; PCR 11,2 mg/L) e uma gamopatia IgM/Kappa completa. O doente preenchia os Critérios de Estrasburgo para o diagnóstico de síndrome de Schnitzler. A biópsia cutânea corroborou o diagnóstico, apresentando caraterísticas de dermatose urticariforme neutrofílica. A biópsia da medula óssea excluiu doença linfoproliferativa primária e o doente iniciou tratamento com inibidor de IL-1 (anakinra), com excelente resposta clínica e laboratorial.

Caso 2: Mulher de 36 anos, com antecedentes de carcinoma folicular da tiroide, submetida a hemitiroidectomia direita, que foi referenciada à consulta de Reumatologia por apresentar exantema pruriginoso disseminado maculopapular e vesicular, assim como poliartralgias assimétricas nas mãos e adenomegalias axilares palpáveis e indolores. Havia sido previamente medicada com anti-histamínicos, glucocorticóides sistémicos e anti-inflamatórios não esteróides, com escassa melhoria. Objetivamente, apresentava artrite das articulações interfalângicas proximais e lesões urticariformes dispersas pelo tegumento. As análises laboratoriais mostraram aumento dos marcadores inflamatórios (VS 40 mm/1ªh; PCR 13 mg/L) e higergamaglobulinémia de base larga com IgM sérica elevada apesar do resultado da imunofixação ser negativo. Os estudos imunológicos revelaram positividade para anticorpos antinucleares (ANA) em título baixo (1/80, padrão homogéneo), com resultados negativos para anti-dsDNA, anti-ENA, ANCA e crioglobulinas. Com a persistência dos sintomas, foi realizada uma biópsia cutânea, cujos resultados foram compatíveis com dermatite leucocitoclástica granulocítica, sugerindo evidência de síndrome de Schnitzler. Apesar da doente não cumprir os critérios diagnósticos necessários, a apresentação clínica e a biópsia cutânea apontavam para o diagnóstico provável de síndrome Schnitzler-like. Após exclusão de doença linfoproliferativa e outros diagnósticos diferenciais relevantes, como vasculite urticariforme, a doente iniciou tratamento com inibidor de IL-1, com boa resposta inicial, mas desenvolvendo reações cutâneas locais repetidas no local de administração. Posteriormente à transição para inibidor de IL-6 (tocilizumab),



150 – Figure 1. Exantema urticariforme no tronco e braço direito, à esquerda e direita, respetivamente, em doente com Síndrome de Schnitzler.

a doente apresentou melhoria clínica sustentada.
Discussão: Apesar de serem condições raras, com manifestações clínicas inespecíficas, estes casos demonstram a importância de diminuir o limiar de suspeição relativamente às síndromes de Schnitzler e Schnitzler-like, devido à excelente resposta terapêutica dirigida que apresentam.

151 - VASCULITE POR IGA NO ADULTO COM MANIFESTAÇÃO PRODRÓMICA PARANEOPLÁSICA?

Ana Sá[1], Sara Amaro Lopes[1], Salomé Garcia[1, 2], Bruno Miguel Fernandes[1, 2], Lúcia Costa[1, 2]
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Introdução: A Vasculite por IgA, previamente designada púrpura de Henoch-Schönlein, é uma vasculite sistémica mediada por imunocomplexos, que afeta predominantemente pequenos vasos, com deposição de IgA na parede vascular. Embora mais frequente em idade pediátrica, a apresentação em adultos tende a ser mais grave, pelo risco acrescido de evolução para doença renal crónica (DRC). A etiologia permanece muitas vezes indefinida, sendo, no entanto, importante excluir causas secundárias, como infeção, introdução de fármacos e neoplasia.
Caso clínico: Homem de 65 anos, com antecedentes de gastrite crónica, foi admitido no Serviço de Reumatologia por quadro de púrpura palpável no tronco, membros inferiores (MIs) e membros superiores (MSs), bem como oligoartrite simétrica, afetando os joelhos e articulações tibiotársicas. Negava introdução de fármacos de novo ou clínica sugestiva de foco infecioso/infeção recente. Analiticamente, apresentava leucocitose ligeira (11x10'^9/L) e elevação dos parâmetros inflamatórios (VS 50mm/1ªh; PCR 40.6 mg/L), sem alteração da função renal; o exame sumário de urina revelava apenas eritrocitúria ligeira. As serologias víricas (hepatites B e C, HIV, CMV, EBV, parvovirus B19, sarampo e varicela) foram negativas, bem como o estudo microbiológico e imunológico (anticorpos anti-nucleares, anti-citoplasma do neutrófilo, anti-double stranded DNA, fator reumatóide, anti-peptídeo citrulinado, anti-antigénios nucleares extraíveis, crioglobulinas e complemento). Realizou ainda TC toraco-abdomino-pélvico, sem alterações de relevo. A biópsia cutânea evidenciou vasculite leucocitoclástica, com expressão de c3c na parede dos vasos e expressão fraca e focal de IgA e C1a. Iniciou corticoterapia sistémica com Prednisolona (PDN) 20mg/dia, com posterior redução para 10mg/dia. Por agravamento de astenia e edemas discretos dos MIs 9 meses após o internamento, procedeu-se à colheita de urina de 24h, a qual revelou proteinúria subnefrótica (1.23g/24h). A biópsia renal revelou proliferação mesangial ligeira e focos de necrose e atrofia tubular, com depósitos mesangiais de IgA na imunofluorescência, confirmando o envolvimento renal pela doença. Manteve PDN 10mg/dia em esquema de desmame e iniciou azatioprina 1mg/kg/dia por manutenção de lesões purpúricas nos MIs e MSs, titulada até 1.5mg/kg/dia, permitindo a sua posterior resolução. Paralelamente, completou-se o estudo de causas secundárias, com elevação do antigénio específico da próstata total (16 ng/mL). Após estudo complementar, foi diagnosticado adenocarcinoma prostático. O doente foi submetido a prostatectomia radical e 6 meses de hormonoterapia adjuvante. Após o tratamento da neoplasia, manteve-se assintomático, tendo sido reduzida a imunossupressão até à sua suspensão 5 anos após a apresentação inicial, sem evidência de recidiva até à data.
Discussão: Este caso ilustra uma apresentação de vasculite por IgA no adulto, sem etiologia identificada ao diagnóstico. A posterior identificação de adenocarcinoma da próstata levantou a possibilidade de uma associação paraneoplásica, já descrita na literatura e em principal relação com tumores sólidos (urogenitais, gastrointestinais e do pulmão (1,2,3)), apesar de nestes casos o diagnóstico de neoplasia parecer preceder o de vasculite por IgA (3). A evolução clínica favorável, com boa resposta à imunossupressão e ausência de recidiva após o tratamento da neoplasia, reforça a importância do seguimento e investigação exaustiva de potenciais causas secundárias nestes doentes, bem como pelo seu risco acrescido de evolução para DRC (4,5,6).

152 - MULTISYSTEMIC MANIFESTATIONS OF IGG4-RD: CHALLENGES IN TIMELY RECOGNITION AND TREATMENT

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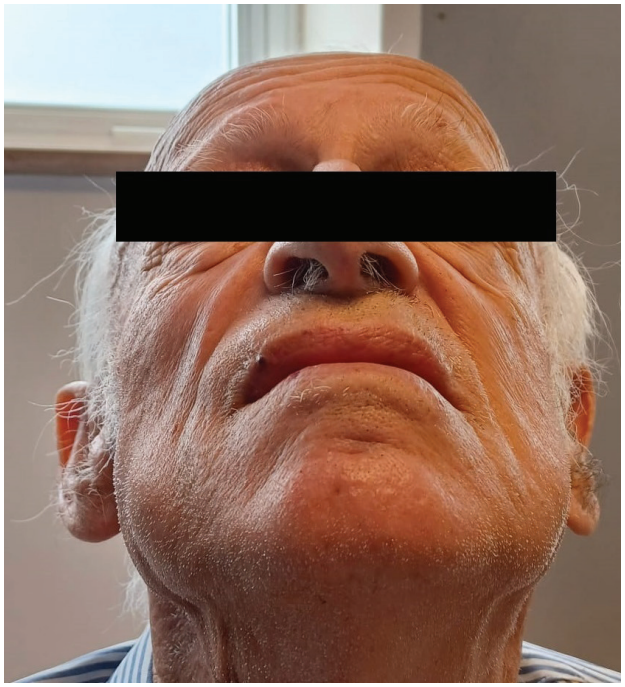
Background: Immunoglobulin G4-related disease (IgG4-RD) is a systemic immune-mediated fibroinflammatory condition characterized by multi-organ involvement. Its highly variable and often insidious presentation contributes to frequent diagnostic delays and

misclassification, particularly in cases with vascular or retroperitoneal involvement.
Case-presentation: We report the case of a 75-year-old man initially referred to Rheumatology for generalised pain and elevated inflammatory markers. An 18F-FDG PET scan revealed increased uptake in the thoracic and abdominal aorta, leading to a presumptive diagnosis of large-vessel vasculitis. The patient received corticosteroid treatment followed by methotrexate but was subsequently lost to follow-up. Eight years later, he was referred for consultation again. A comprehensive review of his medical background revealed a history of abdominal aorta aneurysm repair and a right parotidectomy for unspecified reasons. The physical examination was remarkable for submandibular enlargement (Figure 1.), and laboratory analysis revealed elevated creatinine, near nephrotic-range proteinuria, eosinophilia, and increased levels of acute phase reactants, IgE, and IgG4. Imaging studies, including CT and PET scans, showed aneurysmal dilation of the abdominal aorta with increased tracer uptake, thoracic lymphadenopathy, and pancreatic calcifications. Submandibular gland biopsy showed a dense lymphoplasmacytic infiltration of mainly IgG4-positive plasma cells, consistent with IgG4-RD. The patient was treated with prednisolone and rituximab due to recurrent disease, resulting in a favourable clinical and analytical response.
Conclusion: This case underscores the diagnostic challenges associated with IgG4-RD due to its heterogeneous presentations. Clinicians should consider IgG4-RD in the differential diagnosis of patients with unexplained multisystemic manifestations, recognising the characteristic patterns of organ involvement. Timely identification can enable earlier diagnosis and appropriate management, ultimately improving outcomes.



152 – Figure 1. Submandibular gland enlargement

153 - METHOTREXATE AND BONE HEALTH: WHEN COMMON THERAPY TURNS INTO A RISK

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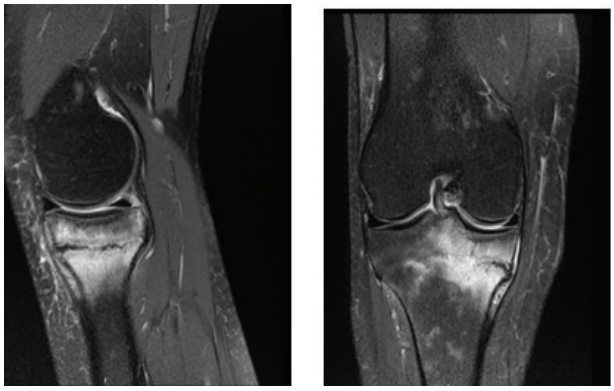
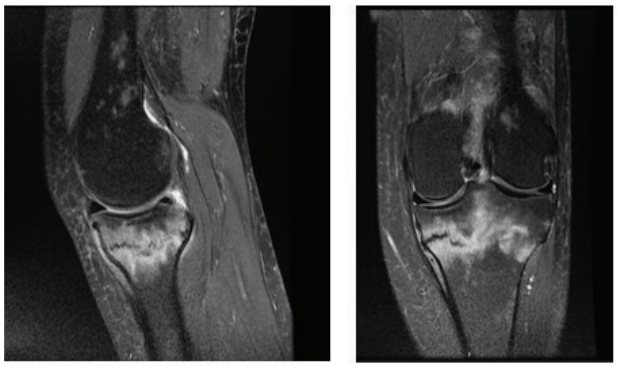
Introduction: Methotrexate (MTX) is a folate antagonist with anti-inflammatory and immunomodulatory effects. MTX in doses ≤ 25 mg per week is commonly used a first-line disease-modifying antirheumatic drug in rheumatic diseases, such as peripheral psoriatic arthritis.1,2 MTX osteopathy is a rare adverse effect characterized by the triad of bone pain, osteopenia/osteoporosis, and stress fractures.3 The first case of MTX osteopathy in rheumatology was described in 1983: a 72-year-old man with psoriatic arthritis who developed a spontaneous stress fracture of the medial femoral condyle.4 Since then, several cases have been reported.3
Clinical case: A 51-year-old woman presented to the rheumatology outpatient clinic with bilateral knee pain without trauma. Diagnosed with psoriatic arthritis in 2019, she had been treated with methotrexate 20 mg/week for four years. The treatment was effective, and at presentation, there were no signs or symptoms of active psoriatic arthritis. Physical examination showed significant tenderness on palpation of both tibial plateaus, without visible local abnormalities. Magnetic resonance imaging (MRI) revealed bilateral stress fractures involving the tibial metaphyses (Fig. 1 and 2). Dual-energy X-ray absorptiometry (DXA) confirmed osteopenia – a femoral neck T-score of -2.3 and a lumbar spine T-score of -1.8. Based on these findings, a diagnosis of MTX osteopathy was established. MTX was discontinued, and treatment with denosumab was initiated. The patient showed marked improvement, with no recurrence of psoriatic arthritis.
Discussion: Stress fractures due to MTX osteopathy most often affect the tibia, followed by the calcaneus.5 Patients usually present with multiple, bilateral, and sometimes recurrent stress fractures.6 While DXA frequently shows osteopenia or osteoporosis, typical osteoporotic fractures are rare in the cases.5 MRI is the preferred diagnotic tool due to its high sensitivity for early and subtle bone changes.3,6 Stress fractures with



152 – Figure 1. MRI of the right knee



152 – Figure 2. MRI of the left knee

a characteristic band- or meander-shaped appearance along the growth plate (epimetaphyseal osteolysis and band-like sclerosis) are considered pathognomonic.6 In most cases, the prognosis is favorable after MTX withdrawal. In some cases, anti-osteoporotic drugs such as antiresorptive and osteoanabolic agents may be used to promote bone healing and improve bone density.5 This case illustrates MTX osteopathy with bilateral tibial stress fractures, osteopenia, and improvement after MTX discontinuation and anti-osteoporotic treatment.
Conclusion: MTX osteopathy is a rare but important adverse effect. It should be considered in patients presenting with unexplained lower limb pain, particularly in the absence of trauma and when the pain does not correspond to typical sites of osteoporotic fractures. Early recognition is essential to prevent complications and guide appropriate therapy.

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155 - SARCOIDOSIS DIAGNOSIS AFTER SUCCESSFUL TREATMENT OF CUSHING'S DISEASE

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Introduction: Cushing's syndrome (CS) is characterized by chronic glucocorticoid excess and is associated with a range of complications, including cardiovascular, metabolic, and musculoskeletal disorders. A less commonly highlighted consequence is its immunosuppressive effect, primarily mediated through lymphoid tissue atrophy and lymphopenia. Following remission of CS, this immunosuppression is reversed, potentially leading to immune reconstitution phenomena. In some cases, this immune rebound may unmask or trigger immune-mediated conditions responsive to glucocorticoids.
Case Report: A 49-year-old woman presented to the Endocrinology clinic with a history of recent significant weigh gain, buffalo hump, easy bruising, lower limb oedema, hypertension and osteoporosis. Personal history was notable for an episode of left eye anterior uveitis and right eye panuveitis at the age of 18. Abdominal computed tomography scan (CT scan) and magnetic resonance imaging (MRI) described a 1.1cm adenoma in the left adrenal gland. However, laboratorial work-up suggested ACTH-dependent hypercortisolism and desmopressin stimulation was suggestive of Cushing's Disease. Accordingly, pituitary MRI confirmed a right pituitary adenoma with 8x11x12mm. Transsphenoidal tumorectomy was performed confirming a corticotroph adenoma and the patient was discharged with hydrocortisone 20+10 mg and desmopressin 60 mcg daily.

In the following weeks, a cervical/shoulder girdle inflammatory pain appeared along with inflammatory arthralgias of the proximal interphalangeal joints of the hands (without arthritis). She also reported dry coughing and xerostomia. Additional evaluation revealed an erythrocyte sedimentation rate of 120 mm, a

C-reactive protein of 1.84 mg/dL, and an elevated angiotensin converting enzyme (77 IU/L). The interferon gamma release assay (IGRA) was negative. Ultrasound of shoulders and hands had no evidence of synovitis or tenosynovitis. Chest CT scan revealed the presence of supraclavicular, mediastinal and hilar lymph nodes, with diffuse areas of nodular opacities of the pulmonary parenchyma, mainly in the lower lobes. Transbronchial lymph node biopsy was carried out and confirmed the presence of non-necrotizing, non-caseating granulomas. The bronchoalveolar lavage had lymphocytosis (26%) with a CD4/CD8 ratio of 3. Bacteriologic, fungal and mycobacterial cultures were negative. Lung function tests showed a slight decrease in DLCO of 68% (diffusing capacity of the lungs for carbon monoxide), with normal forced vital capacity (88%). These findings led to the diagnosis of sarcoidosis (stage II in terms of pulmonary involvement) and treatment with methotrexate was initiated.

Discussion/ Conclusion: Autoimmune diseases are a recognized complication following the resolution of CS. Sarcoidosis has been described, although rarely, typically involving the skin, lungs, or both. Glucocorticoid-withdrawal syndrome should also be considered in the differential diagnosis, as symptoms such as myalgia and asthenia can overlap with adrenal insufficiency and immune-mediated disorders. The episode of panuveitis at age 18 raises the possibility that the disease was already present but unrecognized, and further symptoms only developed after CS cure.

With this case, we want to highlight a rare complication in the posttreatment period of CS, when a close follow-up is of utmost importance along a high level of suspicion when clinical presentation deviates from the expected course of recovery.

156 - DERRAME PLEURAL NA ARTRITE REUMATOIDE: REFORÇANDO A IMPORTÂNCIA DA SUSPEIÇÃO CLÍNICA

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Introdução: Uma das complicações extra-articulares mais comuns da Artrite Reumatoide (AR) é o envolvimento pulmonar, que pode afetar até 70% dos doentes com AR. [1]

Caso clínico: Apresenta-se o caso de uma mulher de 64 anos, diagnosticada na consulta de Reumatologia há oito anos com AR seropositiva e erosiva, medicada com metotrexato (MTX) e ácido fólico desde o diagnóstico. Encontra-se em menopausa desde os 51 anos, sob terapêutica hormonal de substituição (THS) com estradiol e noretisterona.

A doente encontrava-se previamente controlada do ponto de vista osteoarticular, tendo sido observada em consulta de rotina, onde apresentou astenia e dispneia, desencadeadas por um quadro gripal e associadas a um «flare» da AR, com poliartrite das pequenas articulações das mãos. Foi medicada com esquema curto de prednisolona, após o qual manteve apenas quadro clínico de dispneia e astenia, com perda de peso associada. Para esclarecimento do quadro e para rastreio de eventual doença intersticial pulmonar (DIP) associada à patologia de base, foi pedida radiografia do tórax e provas de função respiratória (PFR). As PFR revelaram alterações restritivas e obstrutivas moderadas, e a radiografia do tórax evidenciou derrame pulmonar (DP) bilateral, o que motivou o pedido de tomografia computorizada (TC) do tórax que confirmou a presença do DP bilateral com 70mm de espessura máxima à direita e 58mm de espessura máxima à esquerda (loculado), sem outros achados, nomeadamente, presença de nódulos reumatoides ou de alterações sugestivas de DIP.

O caso foi discutido com a Pneumologia e, atendendo à presença de DP bilateral extenso numa doente com AR controlada, e perante a ausência de patologia intersticial pulmonar e nódulos reumatoides, foi sugerido estudo por toracocentese para esclarecimento diagnóstico.

Procedeu-se à toracocentese com drenagem de 1750ml de conteúdo sero-hemático; analiticamente, o líquido pleural (LP) era um exsudado estéril com pH de 7.44, com presença de abundantes leucócitos (predomínio de mononucleares) e eritrócitos, glucose 100 mg/dL, proteínas totais 4.4 g/dL, albumina 2.8 g/dL, lactato desidrogenase (LDH) 287.0 U/L, colesterol total 93 mg/dL, triglicéridos 33 mg/dL e adenosina desaminase 9,6 UI/L. O exame bacteriológico foi negativo.

Iniciou furosemida e prednisolona em esquema de desmame.

Entretanto no exame citológico do LP foi revelada a presença de células neoplásicas com padrão de expressão imunocitoquímica de neoplasia primitiva genital. Após estudo diagnóstico alargado, foi estabelecido o diagnóstico de carcinoma seroso de alto grau do ovário, em estadio IV, BRCA1/2 negativo.

Conclusão: O DP é uma manifestação que pode, de facto, ser causada pela AR, sobretudo se existir elevada atividade da doença, presença de nódulos pulmonares reumatoides e sinais de doença pulmonar intersticial no exame de imagem, bem como achados pleurais típicos (pH baixo, hipoglicorraquia, LDH e colesterol elevados).

Neste caso clínico, o DP foi a manifestação inaugural de uma neoplasia maligna oculta, o que dificultou

o processo diagnóstico, mas o elevado índice de suspeição clínica, sustentado pela maior incidência de neoplasias em doentes com AR, permitiu orientar a doente para o percurso diagnóstico adequado.

163 - CHALLENGES IN MANAGING NECROTIZING FASCIITIS IN A PATIENT WITH SYSTEMIC LUPUS ERYTHEMATOSUS: A COMPLEX CASE

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Introduction: Necrotizing fasciitis (NF) is a highly aggressive soft tissue infection causing extensive and fulminant destruction of the skin, underlying subcutaneous fat, and fascia. Although rare, NF is associated with a high mortality rate due to its rapid progression and challenges in early recognition, often resulting in multiorgan dysfunction.

Patients with systemic lupus erythematosus (SLE) are at increased risk for severe infections, including NF, due to immune dysregulation, chronic immunosuppressive therapy, and comorbidities.

Case report: We report the case of a 50-year-old woman an diagnosed with SLE at the age of 41, presenting with polyarthritis, hair loss, oral ulcers, and pancytopenia, alongside positive anti-dsDNA antibodies and complement consumption. Her medical history included osteoporosis and an episode of necrotizing herpetic retinitis requiring hospitalization in 2018. She was on methotrexate, hydroxychloroquine, prednisolone 5mg daily and weekly alendronate.

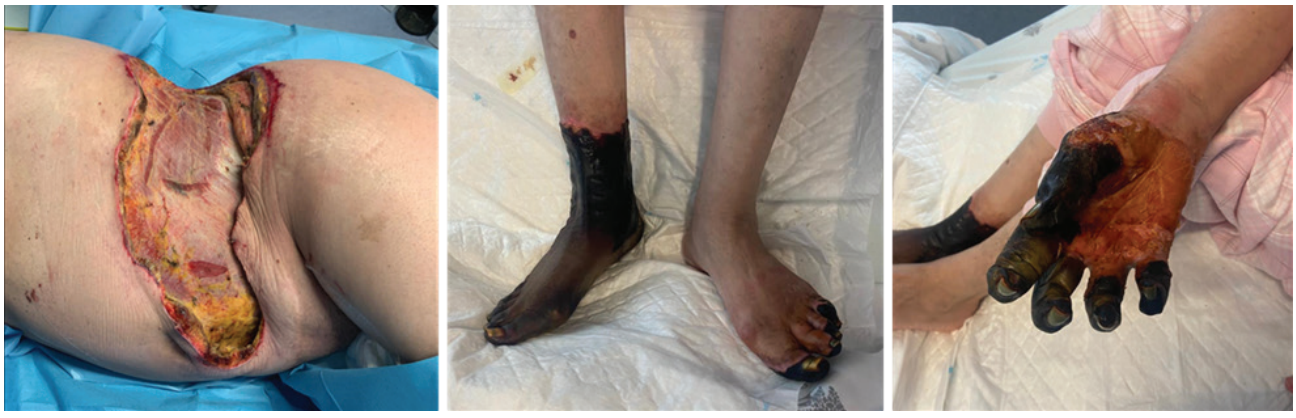
In December 2023, she presented to the emergency department with fever, vomiting, abdominal pain, and oliguria. Examination revealed a necrotic plaque on the left flank measuring 10×15 cm with inflammatory signs at its periphery. She was admitted to intensive care in septic shock with cardiovascular (hypotension requiring vasopressors), respiratory, renal (acute kidney injury, with serum creatinine 3.53 mg/dL) and hematologic (hemoglobin 9.2 g/dL, platelets 102×10^9/L) dysfunctions, and markedly elevated C-reactive protein (432.4 mg/L) and procalcitonin (21.75 ng/mL).

Empirical antibiotics (meropenem, vancomycin, clindamycin) were initiated, and surgical debridement of the affected abdominal region was performed. Streptococcus pyogenes was isolated in both blood cultures and tissue cultures, confirming the diagnosis of NF complicated by streptococcal toxic shock syndrome. Antibiotic therapy was de-escalated to ceftriaxone and clindamycin, alongside three days of intravenous immunoglobulin.

During hospitalization, the patient experienced further deterioration with disseminated intravascular coagulation, resulting in necrosis of multiple digits of the hands and feet and requiring below-knee amputation of the right leg. She underwent multiple surgical interventions (four in total) for debridement and abdominal wall reconstruction.

Having achieved clinical stability, she was discharged after nearly two months of hospitalization. Approximately one month later, she was electively readmitted for amputation of all fingers of the right hand and left foot. Since then, she has had no new complications or SLE flares and continues treatment with hydroxychloroquine, methotrexate, and prednisolone.

Discussion: Infections are a frequent cause of morbidity and mortality in patients with autoimmune diseases, with NF representing an extreme example of this risk. Balancing immunosuppression is imperative to prevent



163 – Figure 1. Photographic registry of severe complications in a lupus patient: necrotizing fasciitis requiring abdominal debridement (left) and disseminated intravascular coagulation leading to peripheral tissue necrosis of feet (center) and hand (right).

flares while minimizing the risk of severe infections. This case highlights the complexity of therapeutic management in NF and its potential for rapidly progressive tissue destruction and resulting life-threatening complications.

A high index of suspicion is essential in the presence of suggestive skin changes in SLE patients. Early recognition and prompt surgical exploration are critical in this population, given their increased vulnerability due to immunosuppression and systemic inflammation. Multidisciplinary collaboration plays a key role in optimizing outcomes, underscoring the need for close monitoring in this high-risk group.

164 - CHONDRITIS – A DERMATOMYOSITIS MANIFESTATION, PARANEOPLASTIC SIGN, OR BOTH?

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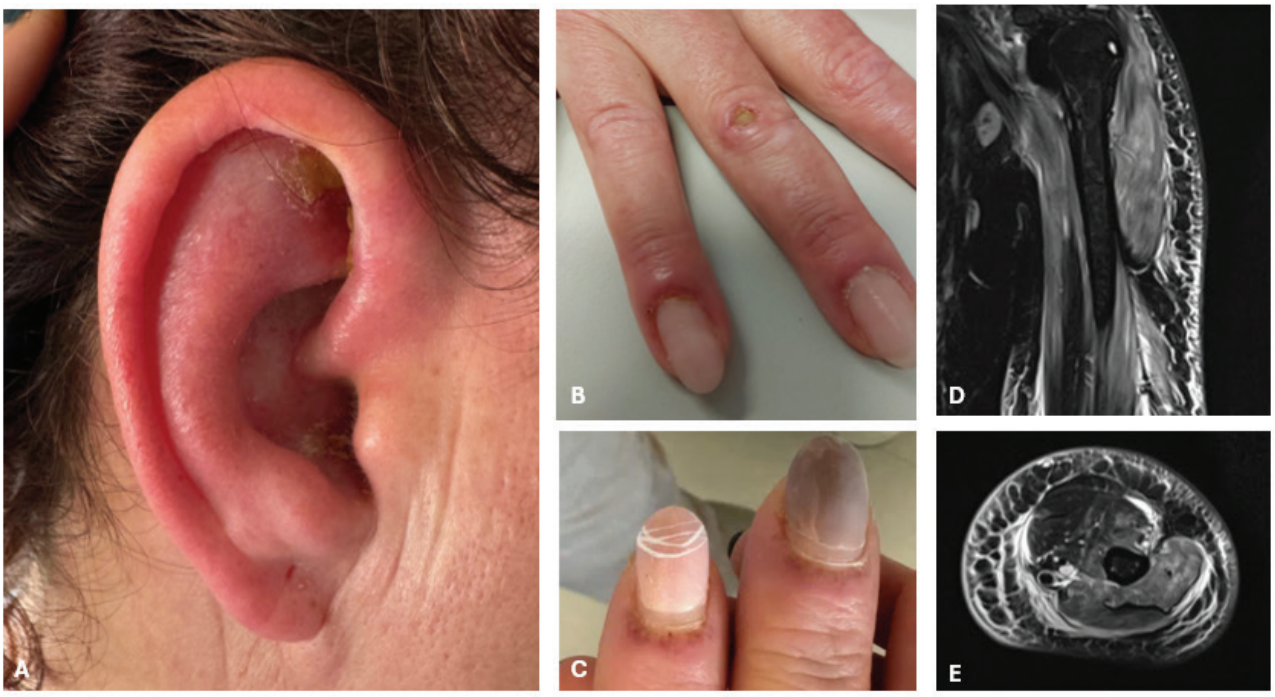
Introduction: Idiopathic inflammatory myopathies (IIMs) are a group of rare systemic autoimmune rheumatic diseases (SARDs) with a well-documented association with malignancy. However, the co-occurrence of chondritis and myositis is exceedingly rare.

Clinical vignette: A 44-year-old Caucasian female presented to the emergency department with facial swelling and ear pain, erythema, and oedema. She had a 2-month history of erythematous rash on her neck, dorsum, abdomen, and limbs and a 2-week history of proximal upper limb myalgia and perceived muscle weakness. She had been previously treated with prednisolone 60 mg daily in a weaning scheme for 2 weeks, which led to a partial and temporary improvement of the complaints, followed by worsening after treatment suspension. The patient recently had an endoscopy for dyspepsia, revealing H. pylori gastritis and antral bulging. A subsequent abdominal ultrasound showed a peri-vesicular nodule and peri-pancreatic lymphadenopathy.

At physical examination, the patient had generalized facial oedema, more striking in the periorbital area and



164 – Figure 1. A - Right auricular chondritis; B - Ulcerative Gottron papule on the extensor surface of the 31d metacarpophalangeal joint C - Exuberant periungueal erythema- D, E -Left arm magnetic resonance imaging showing diffuse myofibrillar edema involving nearty all muscle groups, with relative sparing of the biceps and triceps, with extension to the deltoid, subscapularis, infraspinatus muscles, and the pectoralis major—showing T2 hyperintensity and T1 isointensity. There are also marked inflammatory changes in the subcutaneous tissue, with skin thickening and a nonspecific infiltrative pattern.

malar rash affecting the nasolabial fold. Additionally, erythema, edema, and tenderness of the auricles was noted, V-neck and holster sign, a maculopapular rash of the upper limbs up to the dorsal side of the hands, dorsal and abdominal area, Gottron sign on the extensors surfaces of the metacarpophalangeal joints and knees, periungueal erythema, livedo reticularis of the lower limbs, and proximal muscle weakness (manual muscle testing MMT8- 116/140). Lab tests revealed elevated C-reactive protein (2.57 mg/dL), lymphopenia (0.75x10E9/L), elevated creatine kinase (2649 U/L), and aldolase (33.8 U/L). Anti-nuclear antibodies with a nuclear coarse speckled pattern were detected in indirect immunofluorescence up to the 1:160 titer, and immunoblotting revealed weakly positive anti-TIF1y autoantibodies. Left arm magnetic resonance imaging (MRI) documented increased signal on T2-weighted sequences, with diffuse and generalized myofibrillar oedema, involving practically all muscle groups, suggesting myositis. Skin and muscle biopsies were compatible with dermatomyositis (DM). An abdominal MRI was also performed, showing two adenopathic conglomerates at the hepatic hilum.

A diagnosis of DM with cutaneous and muscular involvement and associated acute non-infectious chondritis was assumed. The patient started hydroxychloroquine 400mg and prednisolone 60mg daily, with skin and strength improvement (MMT8-T 128/150), although she developed ulcerative Gottron papules on her hands. At the time of discharge, the patient was referred to our Myositis Clinic and to a Hepatobiliary Oncology Reference Center, where she underwent biopsy of the lymph nodes and was diagnosed with large cell neuroendocrine cancer. Three months later, the patient passed away from cancer-related complications.

Discussion: We present a case of cancer-associated DM with concurrent acute chondritis. Chondritis is a rare manifestation within the spectrum of rheumatic diseases. Only four cases describing the coexistence of DM and relapsing polychondritis have been reported in the literature, with only one occurring in the context of an underlying malignancy. Although the coexistence of DM and chondritis is exceptionally rare, clinicians should maintain a high index of suspicion, as both conditions can present as paraneoplastic syndromes.

166 - PACINIAN CORPUSCLE HYPERPLASIA: A CASE SERIES OF TWO PATIENTS

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Introduction: Pacinian corpuscles are specialized mechanoreceptors located in the dermis and hypodermis, primarily involved in the detection of vibration and pressure. Hyperplasia of these corpuscles is a rare phenomenon that can cause bothersome symptoms, with limited documentation in the medical literature. Here, we present two cases of Pacinian corpuscle hyperplasia (PCH), characterized by chronic pain, paresthesias, and localized swelling.

Case 1: A 64-year-old female patient, followed in Neurology for due to a history of severe right-sided peripheral facial paralysis (December 2021), associated with incomplete recovery despite physiotherapy and botulinum toxin therapy, was referred for Rheumatology evaluation of paresthesias in the palmar aspect of the proximal phalanges and fingertips of both hands.

The patient denied Raynaud's phenomenon but described nocturnal pain, a "throbbing" sensation in the fingers, and occasional erythema, particularly after wetting the hands. Physical examination revealed crepitation of the flexor tendons in the hands, with no peripheral arthritis.

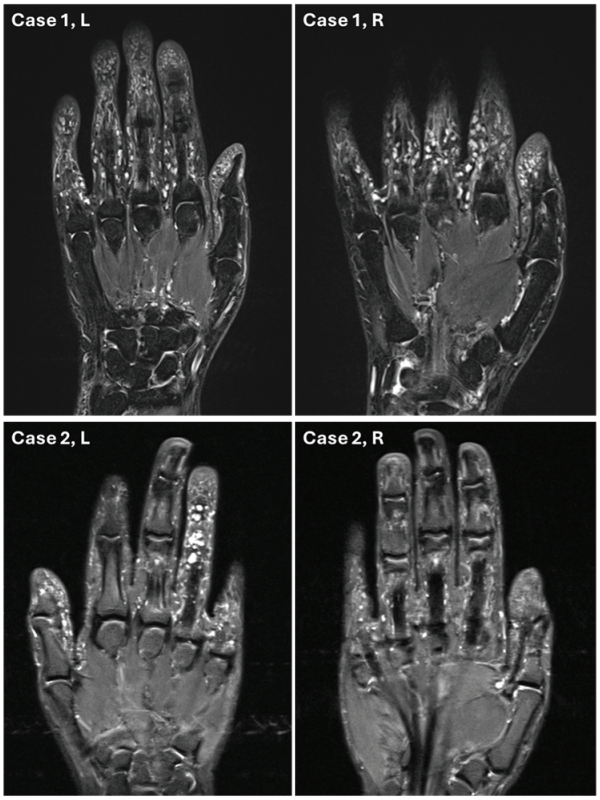
Investigations included an EMG showing chronic neurogenic lesions of the right facial nerve, laboratory studies (normal inflammatory markers, negative autoantibodies and serum protein eletrophoresis), and a hand MRI showing an increase in the number and prominence of Pacinian corpuscles in the palmar subcutaneous tissue, prompting the diagnosis of PCH. This was later confirmed through a skin biopsy, which revealed multiple enlarged Pacinian corpuscles (measuring up to 3 mm in diameter) and perineural and perivascular fibrosis.

Case 2: A 60-year-old female patient with a history of osteoarthritis and autoimmune thyroiditis was referred for evaluation of a 2-year history of recurrent bilateral digital pain and "pricking" sensations, followed by localized swelling and ecchymosis along the affected fingers. The symptoms were not associated with trauma, Raynaud's phenomenon, or digital ulcers.

On examination, there was no evidence of active arthritis or skin changes; however, crepitus was noted in the metacarpophalangeal and interphalangeal joints.

Laboratory evaluation showed mildly elevated inflammatory markers (ESR 47 mm/h, CRP 7.8 mg/L) but no significant autoantibodies. Ultrasound of the hand showed no significant changes. Hand MRI identified multiple rounded, hyperintense formations on fluid-sensitive sequences, consistent with hyperplasia of Pacinian corpuscles, localized in the palmar subcutaneous tissue.

Discussion: PCH is a rare and likely underdiagnosed



166 – Figure 1. MRI T2 STIR and T1 TIRM scans of both hands from cases 1 and 2, respectively, showing identical increase in the number and prominence of Pacinian corpuscles, seen as high-signal rounded formations in the subcutaneous tissue.

condition. Potential links to chronic mechanical stimulation or trauma, neuropathic changes, hematologic malignancies, and occasionally neurofibromatosis have been proposed. Symptoms like digital pain, paresthesias, and ecchymoses are nonspecific, often leading to delays in diagnosis.

MRI (and in some case ultrasound) may provide important information and significantly contribute to the diagnosis, while excluding conditions that may mimic it. Recognizing these imaging patterns and their clinical correlation is key to diagnosis, as seen in both cases exposed. However, histopathologic examination remains essential to establish a definitive diagnosis.

This condition should be considered in patients with unexplained digital pain. Early diagnosis using advanced imaging can prevent unnecessary interventions and aid appropriate management. Further research is needed to clarify its pathophysiology and systemic associations.

167 - TRANSIENT LUPUS-LIKE SYNDROME: RETHINKING THE SPECTRUM OF AUTOIMMUNE DISEASE

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Case report: A 58-year-old woman with a personal history of Hashimoto's thyroiditis and a maternal family history of systemic lupus erythematosus (SLE) presented in May 2024 with disabling polyarthritis preceded by odynophagia, partially responsive to NSAIDs and systemic corticosteroids. Laboratory tests showed normal blood count, liver and renal function; ANA 1:320 (speckled pattern) with negative remaining immunology; parvovirus B19 serology positive for IgM. The clinical picture was interpreted as possible post-infectious arthritis versus inaugural rheumatological disease, and sulfasalazine (SSZ) was initiated.

In August 2024, one week after starting SSZ and following intense sun exposure, she developed a malar rash sparing the nasolabial folds, trunk rash, pleuritic chest pain, fever (39°C), and worsening arthralgias. She was hospitalized due to severe leukopenia (1.45×10^9/L), anemia with a positive direct Coombs test (Hb 9.2 g/dL), hepatic cytolysis (AST/ALT 5× ULN), elevated CRP (50.2 mg/dL), and immunologic seroconversion: ANA 1:640, anti-dsDNA 796.3 U/mL, and positive anti-nucleosome and anti-histone antibodies. Skin biopsy showed interface dermatitis compatible with acute cutaneous lupus. SSZ was discontinued due to suspected hypersensitivity reaction, and high-dose corticosteroids plus hydroxychloroquine were initiated, leading to progressive clinical and laboratory improvement.

Approximately one week after hospital discharge, she developed cognitive changes, confusion, and disorientation. Brain MRI revealed subtle focal areas suggestive of vascular stenoses. She was admitted to the Neurology department for evaluation of possible encephalopathy. Cerebrospinal fluid analysis was normal, and cervical and transcranial Doppler as well as angiography revealed no significant findings. Repeat immunologic testing showed ANA 1:320 with all other autoantibodies negative (including anti-dsDNA, nucleosomes, histones). She was treated with intravenous methylprednisolone (1g/day for 5 days), with marked neurological recovery.

Three months later, complete immunologic negativation was documented. She remained in sustained clinical and laboratory remission on hydroxychloroquine monotherapy, without requiring corticosteroids or additional immunosuppressants.

Discussion: This case presents features that do not fully align with conventional diagnostic patterns of SLE. Drug-induced lupus was considered given temporal relationship with sulfasalazine and anti-histone positivity; however, severe systemic manifestations, extremely elevated anti-dsDNA titers, and neurological

involvement argue against this hypothesis. Conversely, inaugural SLE precipitated by parvovirus B19 is inconsistent with rapid clinical resolution and complete immunological negativation within just three months.

A plausible explanation is a transient lupus-like syndrome secondary to sulfasalazine, potentially amplified by prior immune dysregulation triggered by parvovirus B19 infection. This hypothesis could account for both the atypically exuberant clinical and immunologic presentation and the subsequent spontaneous resolution. Sustained remission with hydroxychloroquine monotherapy reinforces the self-limited nature of the process.

Challenging cases such as this one highlight the heterogeneity of transient autoimmune syndromes and question the rigidity of current diagnostic criteria, emphasizing the need for a deeper understanding of the underlying pathophysiological mechanisms of these atypical syndromes.

174 - LOW-TITER MPO-ANCA AND PULMONARY FIBROSIS: UNMASKING A SILENT VASCULITIS

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Case report: A 70-year-old woman had been followed in Pulmonology since 2022 for presumed idiopathic pulmonary fibrosis (usual interstitial pneumonia pattern on HRCT scan). Autoimmune screening was negative, and genetic sequencing did not identify pathogenic variants, despite a family history of pulmonary fibrosis. She was treated with pirfenidone and remained clinically stable for two years.

In June 2024, she developed fatigue and new-onset hypertension associated with progressive kidney dysfunction (creatinine increased from 0.64 to 2.78 mg/dL over 6 months), microscopic hematuria, and non-nephrotic proteinuria (P/Cr ratio 636 mg/g). MPO-ANCA was borderline positive (20.0 U/mL; N<3.5), with negative remaining immunological panel. She was referred to Nephrology.

ANCA-associated vasculitis (AAV) was suspected and immunosuppression with methylprednisolone pulses was initiated (1g x 3 days). Renal biopsy demonstrated glomerulonephritis with fibrous, cellular, and fibrocellular crescents, fibrinoid necrosis, and marked interstitial fibrosis (70% of parenchyma). Immunofluorescence showed vestigial mesangial C3 deposits. Chest CT revealed progression of fibrotic changes without other vasculitic involvement, particularly no alveolar hemorrhage (AH). Pirfenidone was discontinued due to GFR <30 mL/min.

After multidisciplinary discussion, CYCLOPS protocol with cyclophosphamide (CYC) was initiated and oral prednisolone tapering according to PEXIVAS scheme. Evolution was favorable, with improvement in renal function (serum creatinine ~1.4 mg/dL) and proteinuria (P/Cr ratio ~400mg/g), normalization of urinary sediment and ANCA negativation. The patient remained asymptomatic and normotensive. Pirfenidone was resumed at full dose in January 2025. After 10 cycles of CYC, maintenance rituximab was initiated, with the first cycle completed in May 2025.

Discussion: AAV are systemic necrotizing vasculitides that preferentially affect small vessels with highly variable clinical presentations, with renal involvement being frequent. The presence of ANCA at low titers should not be undervalued when associated with compatible clinical manifestations. In this case, AAV as the etiology of rapidly progressive renal insufficiency was unequivocal, with compatible renal biopsy changes demonstrating marked chronicity but also significant inflammatory activity.

Pulmonary involvement in AAV typically presents as AH. However, pulmonary fibrosis is increasingly recognized as a manifestation of AAV, particularly in patients with MPO-ANCA, and may precede, coincide with, or follow the diagnosis of vasculitis, occurring even without AH. This raises the hypothesis that the pulmonary disease in this case could also be AAV-related, supported by the observed radiological progression.

It is noteworthy that ongoing studies are exploring whether antifibrotic agents used in pulmonary fibrosis, such as pirfenidone, may also delay fibrosis progression in other organs, particularly the kidney, in chronic inflammatory diseases. Early pirfenidone reintroduction following partial renal recovery could potentially provide dual benefits in the long term: maintaining lung function while simultaneously halting progression of kidney fibrosis.

This case illustrates the complexity of diagnosing AAV, particularly when initial manifestations are atypical. Although a causal relationship between pulmonary fibrosis and AAV remains uncertain, its possibility should be considered. Timely biopsy and multidisciplinary management were key to guiding treatment and reintroducing antifibrotic therapy appropriately.

175 - NAIL–PATELLA SYNDROME: UNCOVERING VASCULAR INVOLVEMENT BEYOND SKELETAL ABNORMALITIES

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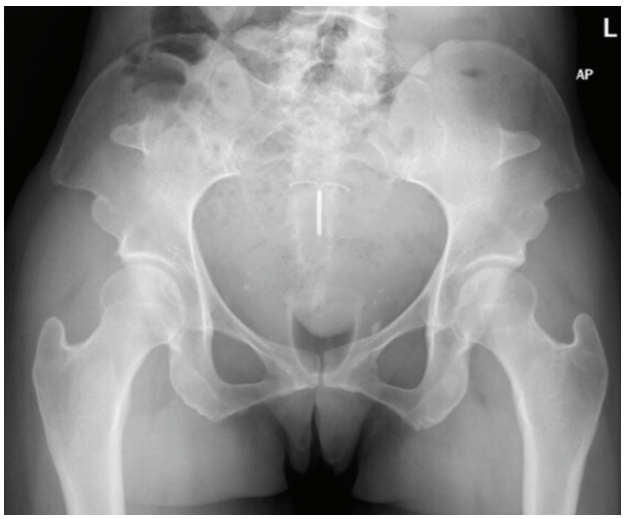
Nail–patella syndrome (NPS) is a rare autosomal dominant disorder caused by pathogenic variants in the LMX1B gene. It typically presents with nail dysplasia, absent or hypoplastic patellae, iliac horns and elbow abnormalities. The syndrome results from altered transcription of genes involved in limb, renal, and ocular development, and may affect extracellular matrix proteins such as collagen types I and III.

The authors present the case of a 37-year-old woman with chronic joint pain, functional impairment due to limitation in pronation and supination from elbow abnormalities. She also exhibited nail abnormalities, hypoplastic patellae, and iliac horns identified on radiography (figure 1). Genetic testing confirmed NPS with a pathogenic LMX1B variant. Her past medical history includes asthma, allergic rhinitis, extrasystoles, and notably, a hemorrhagic stroke in the left thalamo-capsular region, extending into the lateral ventricle and causing tetraventricular hemorrhage. This resulted in gait imbalance, mild right hemiparesis, sensory and proprioceptive deficits, and mild bilateral fourth cranial nerve paresis. Extensive workup revealed no identifiable etiology for the stroke.

Literature on vascular anomalies in NPS is scarce. To the best of our knowledge, this is the first documented case of stroke associated with this syndrome. Literature reports on vascular involvement include internal carotid artery aplasia, spontaneous coronary artery dissection and ST-segment elevation myocardial infarction associated with anomalous coronary anatomy and aneurysms. Recently, ocular microvascular involvement was also reported, in which the investigation excluded other causes and suggested that NPS was the underlying cause of retinal ischemia and neovascularization. This case raises the possibility of vascular involvement in NPS, as suggested by previous reports, supporting the hypothesis that LMX1B mutations might impair endothelial development or vascular integrity, considering the crucial role of Type I and III collagen in vascular walls.



175 – Figure 1. Pelvic radiography of the patient, demonstrating iliac horns, characteristic of the NPS.

We emphasize the importance of recognizing non-musculoskeletal complications in NPS, particularly vascular events. Further research is needed to clarify whether these are coincidental findings or inherent to the disease. Thus, physicians should maintain a high index of suspicion for systemic vascular anomalies in these patients. Multidisciplinary follow-up is essential to address the full clinical spectrum of this syndrome.

179 - ACUTE MYOCARDIAL INFARCTION AS A RARE MANIFESTATION OF BEHÇET'S DISEASE: A CASE REPORT

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Introduction: Behçet's disease (BD) is a variable vessel vasculitis characterized by inflammation, endothelial destruction, and coagulation abnormalities. The most frequent manifestations of this disease are recurrent oral and genital aphthous ulcers and uveitis. Coronary artery involvement presenting as acute myocardial infarction is rare, with fewer than 70 cases reported in literature worldwide. The authors present a case of an acute myocardial infarction as a presentation of Behçet's disease.

Case report: A 47-year-old male with a history of BD presented to the emergency department with pleuritic chest pain. His medical history included recurrent oral and genital ulcers, bilateral panuveitis at the age of 38, and central retinal artery occlusions in both eyes during his early 30s. He had been treated with prednisone and adalimumab, achieving sustained remission and discontinuing therapy at the age of 44.

At presentation, electrocardiogram (ECG) showed

sinus rhythm without ST-T abnormalities. Transthoracic echocardiography (TTE) revealed a non-dilated left ventricle with preserved ejection fraction and a slightly hyperrefringent pericardium with a small effusion. Troponin levels were elevated (>400 ng/L), leading to a presumptive diagnosis of perimyocarditis, and he was treated with nonsteroidal anti-inflammatory drugs and muscle relaxants. Four months later, he returned with malaise, left-sided chest pain radiating to the left upper limb, and fever (>38°C). ECG demonstrated ST-segment elevation in leads V1-V5, T-wave inversion in V4-V6, and deep Q waves in V1 and V2. He was admitted to cardiology, received acetylsalicylic acid (ASA) 300 mg and unfractionated heparin 5000 units IV, and was transferred for catheterization. Coronary angiography revealed total occlusion of the mid-left anterior descending artery immediately after the first diagonal branch, with no lesions in other coronary arteries. Angioplasty was not pursued due to probable lack of myocardial viability. TTE showed moderate systolic dysfunction (left ventricular ejection fraction 37%, global longitudinal strain 10.9%), consistent with new-onset heart failure. He was managed with ASA 100 mg, clopidogrel 75 mg, and guideline-directed medical therapy (rosuvastatin 20 mg) due to poor systolic function. Post-discharge, at the Rheumatology appointment, he reported inflammatory low back pain with morning stiffness lasting less than one hour and a recurrence of oral ulcers for six months. He denied other systemic symptoms. After exclusion of other infectious and non-infectious etiologies, Behçet's disease was assumed as the etiology of the myocardial infarction. Despite the presence of heart failure, given that it was classified as NYHA class I, and following interdisciplinary discussion, adalimumab was reintroduced, leading to sustained clinical and analytical remission. However, after six months, he developed hepatotoxicity, prompting a switch to infliximab 5 mg/kg every six weeks, with a bridging course of prednisone 15 mg. He remained clinically and analytically stable thereafter.

Conclusion: This case illustrates the rare occurrence of myocardial infarction in BD. The differential diagnosis of chest pain in BD should include acute myocardial infarction, given the possibility of coronary vasculitis or a higher cardiovascular risk compared to the general population. The strategy to mitigate the likelihood of future events should include close clinical and laboratory monitoring, along with tailored immunosuppressive therapy.

183 - HYPOTHYROID MYOPATHY: A RARE YET REVERSIBLE CAUSE OF MUSCLE WEAKNESS

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Introduction: Hypothyroidism affects multiple systems due to reduced thyroid hormone levels. While symptoms like fatigue and cold intolerance are well known, musculoskeletal manifestations are less commonly recognized and often misattributed to neuromuscular disorders, which leads to delayed diagnosis[1].

We present a case of severe hypothyroid myopathy with rhabdomyolysis as the initial manifestation of hypothyroidism.

Case presentation: A 42-year old locksmith male was referred to our clinic with a one-year history of intense asthenia, muscle weakness, lower limb pain and difficulty climbing stairs or lifting his arms. History includes past smoking, past heavy alcohol use, depression, and dyslipidemia. The patient was medicated with fluoxetine, alprazolam, rosuvastatin and ezetimibe. No relevant family history.

The symptoms began in early 2024, initially affecting the lower limbs and later the upper body. Over time, the symptoms became progressively worse, and the patient started to lose a significant amount of weight (6% bodyweight in 6 months). The patient also developed hair loss and bilateral upper eyelid swelling. Other systemic symptoms were denied.

Initial workup showed a normal EKG and normal leg doppler. The echocardiogram revealed a mildly dilated left atrium and small pericardial effusion with normal function. Labs showed creatinine 1.24 mg/dL and 24-hour proteinuria of 352 mg, suggesting non-nephrotic range proteinuria. Diuretics were started but ineffective.

In November 2024, rosuvastatin and ezetimibe were stopped due to suspected statin-induced myopathy, but symptoms persisted. Further laboratory evaluation showed TSH 52.9 mUI/L (N-0.4 and 4.0 mIU/L) and anti-TPO 301 IU/mL.CK was 24,998 U/L (N- 24- 204 U/L) and aldolase was 145.6 U/L (N 1-7.5 U/L). Thyroid ultrasound supported the diagnosis of Hashimoto's thyroiditis. Levothyroxine 50 mcg/day was started.

By January 2025, mild clinical improvement was noted; TSH dropped to 16.8 mUI/L. Liver enzymes were elevated [TGO (AST) at 87 U/L (N: 5–40), GGT at 71 U/L (N: 9–48), and TGP (ALT)114 U/L (N: 7–56)]. Anti-nuclear antibodies were negative, as well as myopathy specific or associated antibodies. Levothyroxine dosage has been titrated to 100 mcg/day.

Follow-up showed improved strength and improvement of muscle enzymes: CK 269 and aldolase 10.5

U/L. Proteinuria and pericardial effusion also improved with treatment.

Due to the delay in diagnosis, the patient suffered from severe muscle loss and rhabdomyolysis, which led to multiple disfunctions, including renal, hepatic and cardiac involvement. At this point, the patient hasn't yet returned to work. In order to maximize his rehabilitation potential, the patient has been prescribed physical therapy.

Conclusion: Hypothyroid myopathy is often misdiagnosed due to nonspecific symptoms. In this case, delayed diagnosis resulted in severe rhabdomyolysis and multisystem involvement. Clinicians must consider thyroid dysfunction in patients with unexplained muscle weakness, as it can be the sole manifestation or presentation of hypothyroidism. Early diagnosis and appropriate thyroid hormone therapy are essential for full recovery, to minimize unwarranted investigations and to prevent complications and therapeutic delay.

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184 - RHEUMATOID, PSORIATIC OR REACTIVE ARTHRITIS? A CLINICAL CASE REPORT

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Introduction. Anti-cyclic citrullinated peptide (anti-CCP) antibodies are usually associated with rheumatoid arthritis (RA) with a specificity of 86-99%. However, they can also be presented in a small portion of (<10%) of psoriatic arthritis (PsA) patients, being associated with polyarticular and more erosive joint involvement. Rheumatoid factor (RF) may also be found positive in around 10% of PsA patients. When both this serum markers are positive, differential diagnosis between RA and PsA may be complicated and clinical and imaging evaluations become even more relevant.

Clinical case report. We present the case of a 59-year-old caucasian man, smoker and with past history of infectious urethritis. In 2014 (at 48 years of age) he started complaining of cervical and lumbar inflammatory pain as well as inflammatory pain and swelling of both wrists. Morning stiffness was present and symptoms were relieved by nonsteroidal anti-inflammatory drugs (NSAIDs). Physical examination showed positive sacroiliac maneuvers on the right side, reduced cervical mobility and evidence of synovitis of both wrists. Blood work revealed elevated inflammatory parameters, positive RF (50 UI/mL) and anti-CCP (81 Uml), positive Chlamydia trachomatis IgG and negative human leukocyte antigen B27 (HLA-B27). X-ray imaging showed right grade 3 sacroiliitis, syndesmophytes of thoracic spine, erosions of atlas' anterior arch and axis' odontoid process as well as marginal erosions on carpal joints and distal radioulnar. Systemic treatment with leflunomide was started after methotrexate intolerance, achieving only partial clinical improvement and the patient is currently on routine screening to start anti-TNF therapy. In June 2025, patient presents with foot nail dystrophy and psoriatic-like skin lesions of the foot and is currently waiting a dermatology evaluation.

Discussion and conclusions. In a patient with clinical arthritis, positive RF and anti-CCP, erosive marginal carpitis and atlantoaxial joint erosions, RA becomes a possible diagnosis. However, asymmetrical joint involvement, presence of radiographic sacroiliitis, syndesmophytes favor the hypothesis of spondylarthritis. In this context, psoriatic-like skin lesions may indicate a RF and anti-CCP positive PsA, whereas urethritis history and positive Chlamydia trachomatis IgG may raise reactive arthritis as a possible diagnosis. Differential diagnosis may also include an overlap of this diseases. In rheumatology clinical practice, differential diagnosis can sometimes be difficult and must rely on clinical, laboratory and imaging findings.

185 - BEYOND BEHÇET: UNVEILING TRISOMY 8 – ASSOCIATED AUTOINFLAMMATORY DISEASE (TRIAD) IN A PAEDIATRIC PATIENT

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Introduction: Behçet's disease (BD) is an autoinflammatory (AI) disorder of unknown cause, marked by recurrent oral and genital ulcers, uveitis, and skin lesions. Trisomy 8, a chromosomal anomaly often linked to myelodysplastic syndromes, is also associated with Behçet-like and other AI features. TRIAD (Trisomy 8–associated AI disease) has been recently proposed to describe this overlap, though reported cases remain limited.
Objectives: To describe a case of TRIAD.
Methods: Case-report.
Results: We report the case of a now 12-year-old girl with an early-onset, persistent autoinflammatory syndrome. Family history included recurrent oral aphthosis (mother) and Behçet's disease (second-degree maternal cousin). Symptoms began at 18 months with recurrent high-grade fevers (40ºC), painful oral ulcers, myalgias and arthralgias. She was initially diagnosed with Periodic Fever, Aphthous stomatitis, Pharyngitis and Adenitis (PFAPA) and treated with corticosteroids during flares, with only partial response. At age 4, she developed periumbilical abdominal pain and marked perianal aphthosis. Endoscopic studies were unremarkable. At age 10, she presented panniculitis. During flares — marked by oral aphthous ulcers and severe abdominal pain — she required frequent hospital admissions and total enteral nutrition, with limited improvement. Diagnostic work-up revealed leucocytosis, elevated C-reactive protein and persistently high serum amyloid A levels (peak 710 mg/L). HLA-B51 was negative. Urinary mevalonic acid, urinalysis and proteinuria were normal. Faecal calprotectin was intermittently elevated (max 9531 µg/g). New endoscopic study showed nonspecific mucosal erosions and ulcerations in the cecum, with histopathology revealing mild nonspecific inflammation. Enteral MRI was normal. Genetic testing identified a mosaic trisomy 8, confirmed by FISH in peripheral blood but not in buccal cells, alongside a pathogenic variant in TNFRSF13B inherited from her asymptomatic father.

Therapeutic management was challenging. Initial treatment with azathioprine and colchicine was ineffective. Adalimumab provided partial relief, while tofacitinib showed no benefit. Infliximab was recently started. She currently experiences episodic abdominal pain and perianal aphthosis, with some improvement, alongside stable inflammatory markers. Haematological evaluation shows no evidence of myelodysplastic changes.
Conclusion: This case illustrates the diagnostic and therapeutic challenges of early-onset BD-like syndromes and the importance of investigating genetic or chromosomal causes in atypical or refractory cases. Mosaic trisomy 8 has recently been linked to AI diseases, often with gastrointestinal involvement and poor therapeutic response. It is also a known cytogenetic abnormality in myelodysplastic syndromes, raising concerns about long-term risk. These overlapping features highlight the need for multidisciplinary care and follow-up.

190 - ESCLEROSE ÓSSEA AXIAL – O DESAFIO DO DIAGNÓSTICO DA OSTEOMESOPICNOSIS

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Introdução: A osteomesopicnosis é uma displasia óssea benigna extremamente rara, de provável transmissão autossómica dominante. Dos cerca de 20 casos descritos mundialmente, cerca de 50% tem origem em França, Espanha e Portugal. Caracterizada por esclerose do esqueleto axial e ausência de alterações no metabolismo fosfo-cálcico. Trata-se de um diagnóstico de exclusão, frequentemente detetado de forma incidental em indivíduos em idade adulta e adolescência. O quadro clínico apresenta-se com sintomas minor, tais como lombalgia ligeira. Os autores apresentam um caso de osteomesopicnosis numa doente com espondilartrite axial, em que o reconhecimento imagiológico foi determinante.
Descrição do caso: Mulher de 50 anos, seguida em consulta de Reumatologia desde 2018 por espondilartrite axial radiográfica associada a doença inflamatória intestinal (Doença de Crohn) e fibromialgia. Outros antecedentes patológicos relevantes: síndrome de apneia obstrutiva do sono sob CPAP e litíase renal submetida a litotrícia em 2020. De salientar história de menopausa precoce aos 43 anos. Sob terapêutica com golimumab mensal, salazopirina 1,5gr dia, prednisolona 5mg dia, celecoxib 200mg dia e vitamina D mensal com controlo clínico da doença traduzido por BASDAI 5,2 e ASDAS-PCR 2,8.

Ao longo do seguimento, constatou-se nas radiografias da bacia alterações sugestivas de scrolileite grau II à direita e aumento marcado da densidade óssea do esqueleto axial (coluna cervical, lombar, bacia e porção proximal do fémur) e osteofitose marginal lombar dis-

creta. Densitometria óssea em 2018 revelava T-score muito aumentado: coluna lombar Tscore +5,9; colo do fémur Tscore +6,0; rádio 1/3 Tscore +1,5. Valores mantidos em 2019 e 2022: L1-L4 com Tscore +5,4 (BMD 1,645 g/cm2); colo do fémur Tscore +5,6 (BMD 1,629g/cm2). Analiticamente apresentava: calcémia 9,26 mg/dL, fosfatémia 4,0 mg/dL, hormona da paratiroide (PTH) 28 pg/mL, vitamina D 30 ng/mL e fosfatase alcalina 63 U/L, dentro dos intervalos de referência.

Discussão: O padrão de esclerose axial acentuada, na ausência de alterações analíticas ou envolvimento do esqueleto apendicular, motivou a consideração de osteomesopicnosis, uma displasia rara e podendo estar subdiagnosticada. O diagnóstico é eminentemente imagiológico e de exclusão, não existindo atualmente teste genético específico. A associação com litíase renal, embora rara, encontra-se descrita na literatura e esteve presente nesta doente. A coexistência com espondilartrite axial representou um desafio adicional no raciocínio clínico, exigindo análise criteriosa. Salienta-se, ainda, que o diagnóstico diferencial de esclerose óssea difusa inclui outras entidades como a patologia metastática, o que reforça a importância de um diagnóstico atempado e rigoroso.

Conclusão: A osteomesopicnosis deve ser considerada no diagnóstico diferencial de esclerose óssea axial marcada, especialmente em doentes com densidade mineral óssea aumentada e estudo laboratorial fosfo-cálcico sem alterações. Este caso ilustra o desafio clínico que representa uma doença rara, o papel essencial da radiologia e a importância da exclusão cuidadosa de diagnósticos alternativos.

193 - QUANDO A HIDROXICLOROQUINA NÃO É A MELHOR OPÇÃO: UM CASO DE TROMBOCITOPENIA SECUNDÁRIA A HIDROXICLOROQUINA

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Introdução: A trombocitopenia induzida por fármacos é uma entidade clínica complexa, subdiagnosticada e potencialmente associada a complicações hemorrágicas graves. A hidroxicloroquina é um fármaco antimalárico usado comummente como tratamento de 1ª linha em doentes com Lúpus Eritematoso Sistémico (LES). Embora seja um fármaco seguro e bem tolerado, há casos descritos de citopenias secundárias à sua toma, incluindo trombocitopenia. Este efeito adverso pode surgir dias ou semanas após a introdução do fármaco ou, menos frequentemente, após o seu uso prolongado.

Caso Clínico: Doente de 37 anos, do sexo feminino, reformada por invalidez, com LES diagnosticado aos 16 anos, com envolvimento cutâneo, hematológico (anemia) e renal (glomerulonefrite lúpica classe IV com necessidade de progressão para tratamento dialítico). Sem seguimento regular durante anos e com má adesão à terapêutica. Dada a progressão da doença renal, foi observada em consulta de Nefrologia, onde foi iniciado tratamento com hidroxicloroquina 400mg id. Foi referenciada para observação por Reumatologia, por apresentar trombocitopenia grave, sem melhoria após introdução de prednisolona 60mg id. À observação, em internamento, apresentava IMC>30 Kg/m2, fácies cushingóide e múltiplos hematomas espontâneos dispersos pelo corpo, sem outras alterações no exame objetivo geral e articular. Analiticamente, com positividade para ac anti-dsDNA, anti-histonas, anti-cromatina e negatividade para ac anti-plaquetários e anti-fosfolipídicos, alteração grave da função renal (creatinina 9.65 mg/dL, ureia 261 mg/dL), anemia (hemoglobina de 9.2 g/dL) e trombocitopenia de novo (plaquetas 24.000/uL). Neste contexto foi medicada com pulsos de metilprednisolona (500mg id durante 3 dias) e imunoglobulinas (2g/Kg durante 5 dias), seguidos de prednisolona 30mg oral id. Apesar da terapêutica, manteve anemia, assumida no contexto da doença renal crónica em estadio terminal, e sem qualquer melhoria do quadro de trombocitopenia. Tendo em conta o timing de desenvolvimento de trombocitopenia e, excluídas outras causas, foi colocada a hipótese de iatrogenia medicamentosa, pelo que se suspendeu a hidroxicloroquina e foi iniciado desmame da corticoterapia. Em avaliações analíticas seriadas após suspensão de hidroxicloroquina, verificou-se melhoria progressiva até normalização da contagem plaquetária, apresentando aos 6 meses de seguimento plaquetas de 167.000/uL (evolução representada na tabela 1). Concomitantemente, doente com desmame completo de corticoterapia ao final de 6 meses de seguimento, sem agravamento de anemia crónica, flares ou sintomatologia de novo, mantendo tratamento dialítico 3x/semana.

Conclusão: Embora se trate de um fármaco seguro e de uso frequente na prática clínica, a hidroxicloroquina pode ser responsável por quadros de trombocitopenia em doentes com LES, com consequências potencialmente graves e eventualmente fatais. A trombocitopenia secundária a hidroxicloroquina é habitualmente imunomediada, embora anticorpos anti-plaquetários possam não ser detetados, e é expectável que a contagem plaquetária normalize após a sua suspensão. Para se estabelecer o diagnóstico, devem ser excluídas outras causas de trombocitopenia, nomeadamente não

193 – TABELA 1. Evolução da contagem plaquetária e desmame de corticoterapia após suspensão de Hidroxicloroquina

Tempo após suspensão de Hidroxicloroquina	Valor inicial	1S	1M	2M	3M	4M	6M
Contagem Plaquetária	24.000/uL	29.000/uL	32.000/uL	75.000/uL	105.000/uL	115.000/uL	167.000/uL
Dose de Prednisolona	Pulsos de Metilprednisolona 500mg EV seguidos de Prednisolona 30mg	20mg id	15mg id	12,5mg id	5mg id	2,5mg id	0

Legenda: S – Semanas; M – Meses.

farmacológicas. A recorrência de trombocitopenia após nova exposição ao fármaco, embora fortaleça a relação de causalidade, não é exequível nem ética na prática clínica.

194 - DELAYED DIAGNOSIS OF X-LINKED HYPOPHOSPHATEMIA IN ADULTHOOD: A CASE REPORT

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Background: X-linked hypophosphatemia (XLH) is the most common inherited form of phosphate-wasting osteomalacia. It typically presents in childhood. However, in some cases, diagnosis may be missed or delayed into adulthood, especially when comorbidities such as inflammatory joint disease obscure the clinical picture.
Case Presentation: We report a case of a 45-year-old female followed in the Rheumatology clinic for rheumatoid arthritis (RA) in clinical remission on methotrexate. She had a longstanding history of progressive bilateral varus deformity of the lower limbs since early childhood and underwent two corrective orthopedic surgeries between the ages of 6 and 9. Despite this, she continued to report diffuse joint pain, particularly in the knees, without a history of trauma, fractures, infections, or features suggestive of connective tissue disease overlap.

On examination, she exhibited significant skeletal abnormalities including shortened lower limbs, bilateral bowing of the femurs and humeri, and joint stiffness in the shoulders, elbows, hips, and knees, with fixed 20º flexion contractures in both elbows and knees. No active arthritis, lymphadenopathy, or organomegaly was noted.

Laboratory evaluation revealed marked hypophosphatemia (1.7 mg/dL), low-normal serum calcium (8.8 mg/dL), elevated parathyroid hormone (PTH 82 pg/mL), and low 25-OH-vitamin D (39.3 nmol/L). Alkaline phosphatase was normal (72 IU/L), as were renal function markers, magnesium, and serum bicarbonate. Urinary studies revealed hypocalciuria (24 mg/24h) and normophosphaturia.

Bone scintigraphy identified increased uptake in multiple skeletal regions including the right rib, carpus, tarsus, femurs, and sacrum, compatible with sites of prior microfractures. DEXA scan demonstrated osteoporosis in the lumbar spine (Z-score -2.8), with near-normal bone density at the femoral neck (Z-score -0.3). Plain radiographs showed classic features of osteomalacia: bowing of long bones, epiphyseal widening of the radius and ulna, valgus deformities of the knees, and multiple enthesopathies.

FGF23 was markedly elevated (1250 RU/mL; normal <180), a key diagnostic marker for XLH. There was no family history of similar skeletal disorders, but de novo mutations in the PHEX gene are common in sporadic cases.

The patient was started on oral phosphate (monosodium phosphate titrated to 80 mmol/day), calcifediol (0.266 mg biweekly), calcium/colecalciferol (1500 mg/400 IU twice daily), and alendronate (70 mg weekly). Follow-up showed normalization of phosphate and 25-OH-vitamin D levels, with stabilization of bone symptoms.
Discussion: This case illustrates the diagnostic complexity of XLH in adults. Although initial differential diagnoses included acquired causes of phosphate wasting, such as Fanconi syndrome, the absence of metabolic acidosis, glycosuria, or renal dysfunction, along with the characteristic skeletal deformities and elevated FGF23 levels, strongly supported a diagnosis of XLH. Mild PTH elevation likely reflected secondary or tertiary hyperparathyroidism, possibly from prolonged hypocalcemia due to phosphate supplementation with-

out active vitamin D.

XLH results from loss-of-function mutations in PHEX, leading to excess FGF23, reduced renal phosphate reabsorption, and suppression of 1α-hydroxylase activity, impairing activation of vitamin D.

Conclusion: This case underscores the importance of recognizing hereditary causes of osteomalacia in adulthood. Elevated FGF23, early-onset skeletal deformities, and compatible biochemical and imaging findings were essential for diagnosis.

198 - WHEN ARTHRITIS ISN'T JUST ARTHRITIS: THE HIDDEN THYROID FACTOR

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Introduction: Psoriatic arthritis is a chronic inflammatory arthropathy frequently requiring immunosuppressive therapy. Long-term immunosuppression increases susceptibility to infections, complicating management. Concomitant autoimmune diseases, such as Graves' disease, may also influence disease activity and treatment decisions.

Case Report: A 51-year-old woman has been followed in our Rheumatology department since 2001 for psoriatic arthritis with peripheral and axial involvement. Her past medical history is notable for latent tuberculosis diagnosed in 2016, for which she completed a 9-month course of isoniazid. The patient is bio-experienced, previously treated with etanercept (suspended due to recurrent infections) and with adalimumab and methotrexate for the last 2 years, but with several interruptions due to recurrent urinary tract infections, despite infectious prophylactic attempts. These are related to structural renal abnormalities, under urological follow-up.

The patient was admitted due to worsening arthralgias involving the hands, wrists, and ankles, resulting in significant functional impairment. Additional symptoms included low-grade fever, asthenia, dyspnea on moderate exertion, xerostomia, dysgeusia, unquantified weight loss, and symmetrical peripheral edema with a two-week duration. The patient also reported psychomotor agitation and insomnia. She denied genitourinary complaints.

On physical examination, the patient appeared anxious and had tachycardia (140 bpm) with sinus rhythm. There was pain and swelling of the elbows, wrists, several metacarpophalangeal and proximal and distal interphalangeal joints, and the tibiotarsal joints. Additionally, firm, bilateral, symmetrical pretibial edema was present.

Inpatient workup excluded infectious causes and pulmonary embolism. Laboratory evaluation revealed severe hyperthyroidism (TSH <0.01 mUI/L; free T4 >5.00 ng/dL) secondary to Graves' disease. Treatment with methimazole and propranolol was initiated in collaboration with Internal Medicine and Endocrinology, leading to marked clinical and laboratory improvement.

During hospitalization, the patient developed chest pain, and further evaluation led to the diagnosis of acute pericarditis of undetermined etiology, with a possible association with severe hyperthyroidism. She was started on anti-inflammatory therapy and colchicine, with clinical improvement and no recurrence of symptoms.

Due to polyarthritis likely related to withdrawal of immunosuppressive therapy and possibly exacerbated by thyroid dysfunction, the patient was re-started on methotrexate, with concomitant prednisone, resulting in improvement of joint symptoms, but still maintaining relevant disease activity. Considering her history of recurrent infections, ustekinumab was requested to optimize disease control while trying to minimize infection risk.

Conclusion: Multidisciplinary management is essential in complex autoimmune diseases with overlapping pathologies and treatment complications. Adapting immunosuppressive regimens to individual risk profiles can improve outcomes and reduce adverse events.

203 - EXPANDING THE SPECTRUM OF AIRE-RELATED AUTOIMMUNITY: A CASE OF PRIMARY SJÖGREN'S DISEASE IN EARLY CHILDHOOD

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Introduction: Sjögren's disease (SjD) is a chronic autoimmune disorder characterised by lymphocytic infiltration of exocrine glands, though its molecular pathogenesis remains poorly understood. The autoimmune regulator gene (AIRE) plays a key role in central immune tolerance, and its dysfunction predisposes to

autoimmunity, including SjD. In AIRE-deficient mice, early lacrimal gland inflammation and altered signalling pathways mimic SjD, but in humans, this association is rarely reported.
Objectives: To describe a paediatric case of primary SjD likely associated with a heterozygous variant in the AIRE gene.
Methods: Case-report.
Results: A 4-year-old girl, with no relevant past medical history and a family history of systemic lupus erythematosus (maternal grandmother), was referred for bilateral submandibular swelling persisting for over a year, associated with intermittent parotid gland enlargement and recurrent episodes of unexplained high-grade fever (~39 ºC, lasting 1–2 days per month). She also reported xerostomia, with no ocular symptoms. Physical examination revealed elastic, non-tender submandibular swellings measuring ~3.5 cm. Laboratory work-up was unremarkable, except for a positive ANA (1:320) and elevated serum amyloid A (max 37 mg/L). Extractable nuclear antigens (ENA) panel, anti-dsDNA, and rheumatoid factor were negative. IgG4 and angiotensin-converting enzyme (ACE) levels were normal. Cryoglobulinemia, monoclonal component (free light chain ratio), hepatitis C and HIV were excluded.

Ultrasound revealed enlarged parotid and submandibular glands with heterogeneous echotexture and poorly defined hypoechoic areas, suggestive of chronic inflammation. Ophthalmologic evaluation was normal. Submandibular biopsy showed dense B- and T-cell infiltration forming seven lymphoid aggregates (>50 cells) with germinal centres; with a focus score of 2.1, compatible with SjD. No evidence of IgG4-related disease or lymphoma. Given the early onset and systemic features, genetic testing was performed and revealed a heterozygous variant of uncertain significance (VUS) in the AIRE gene: c.816G>T (p.Arg272Ser), not previously reported in the literature. Pathogenic AIRE mutations have been described in both autosomal dominant and recessive forms of autoimmune polyendocrine syndrome type 1 (APS-1). Cytokine profiling and familial segregation studies are ongoing. Treatment with hydroxychloroquine was initiated, with clinical improvement.
Conclusion: This case highlights a rare paediatric presentation of primary SjD likely associated with a previously unreported AIRE variant, pending further clarification through familial segregation studies. It supports the hypothesis that AIRE dysfunction may contribute to early-onset autoimmunity beyond classical APS-1. Findings from AIRE-deficient mouse models reinforce a potential pathophysiological link with salivary gland inflammation. Further studies are needed to elucidate the role of AIRE variants in isolated autoimmune phenotypes such as SjD.

210 - CUTANEOUS LUPUS-LIKE PRESENTATION OF T/NK-CELL LYMPHOMA IN AN IMMUNOSUPPRESSED PATIENT: A DIAGNOSTIC CHALLENGE

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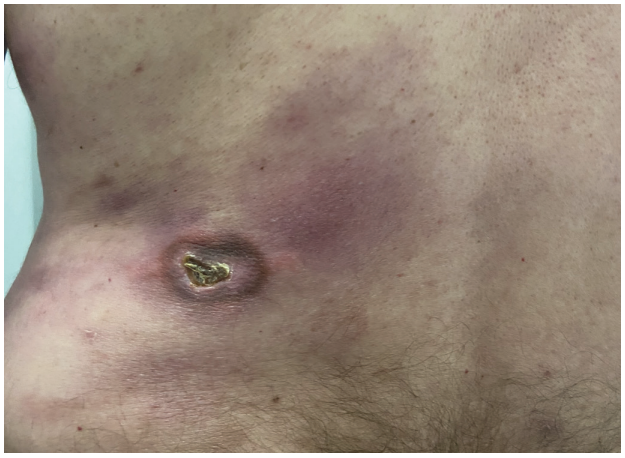
Introduction: Patients with immune-mediated inflammatory diseases (IMID), such as psoriatic arthritis, receiving immunomodulatory therapy have an increased risk of infectious and neoplastic complications. Extranodal nasal-type T/NK-cell lymphoma associated with Epstein-Barr virus (EBV) is a rare and aggressive malignancy that may present with cutaneous lesions mimicking benign dermatoses, delaying diagnosis. Recent evidence suggests a higher incidence of lympho-haematologic malignancies in psoriasis, particularly cutaneous T-cell lymphomas (CTCL), associated with chronic inflammation and immunosuppression.[1]
Case Report: A 55-year-old man with psoriatic arthritis since 2011 was on methotrexate (10 mg/week) and etanercept since 2017 (every 10 days). History included subclinical hepatitis B infection, hypertension, and dyslipidaemia.

In 2022, he developed two erythematous infiltrated plaques on the anterior thorax, worsened by sun exposure. Dermatology evaluation and biopsy suggested tumid lupus. Laboratory tests showed ANA 1:160; ENA, dsDNA, ANCA and viral serologies were negative. Hydroxychloroquine and topical therapy led to near-complete lesion resolution.

In January 2024, with lesion recurrence, a new biopsy again showed features compatible with cutaneous lupus; immunological tests remained negative. Hydroxychloroquine was increased to 400 mg/day.

In February 2025, new annular, polymorphic erythematous lesions and painless pretibial nodules appeared. Biopsy of these revealed primary cutaneous involvement by nasal-type T/NK-cell lymphoma. Lymph node biopsy confirmed EBV-associated cytotoxic T-cell lymphoma.

Staging PET-FDG revealed metabolically active subcutaneous, nodal and osseous disease (D12). Immunosuppressants were stopped, prednisolone 0.75 mg/kg/day was started, and haemato-oncology initiated chemotherapy with PEG-asparaginase, gemcitabine and oxaliplatin.



210 – Figure 1. Lesão ulcerada no dorso

Discussion: The initial presentation and histology suggested cutaneous lupus, despite absent autoantibodies and no clear temporal link with anti-TNF therapy. The diagnosis remained presumptive, and etanercept was continued for arthritis control following multidisciplinary discussion.

Patients with moderate-to-severe psoriasis have an increased lymphoma risk (HR 1.27), particularly for CTCL/mycosis fungoides (HR 6.22).[1] Chronic immunosuppression with methotrexate or anti-TNF agents has also been associated with lymphoma, especially in the setting of EBV and sustained T-cell activation.[2,3]

This case emphasises the importance of re-evaluating diagnoses when clinical or laboratory features are atypical and highlights the role of repeated biopsies when disease patterns evolve.

Conclusion: This case illustrates the complexity of cutaneous differential diagnosis in IMID patients under immunomodulatory therapy. The possible link between psoriasis, immunosuppression and T/NK-cell lymphoma, though debated, is clinically relevant and warrants close surveillance and a multidisciplinary approach.

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212 - RENAL THROMBOTIC MICROANGIOPATHY INDUCED BY ETANERCEPT: A CASE REPORT

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Introduction: Thrombotic microangiopathy (TMA) is a condition resulting from endothelial injury, characterized by microangiopathic hemolytic anemia, thrombocytopenia, and ischemic damage to organs. Renal-limited disease is a less recognized manifestation of TMA. Drug-induced thrombotic microangiopathy (DITMA) accounts for 10%-13% of all TMA cases. This case report describes a patient with renal-limited TMA caused by Etanercept.

Case report: A 59-year-old male with a history of cured hepatitis C and psoriatic arthritis (PA), treated with Etanercept 50mg weekly since 2018, presented to the emergency department in February 2024 with malignant hypertension (blood pressure measured at 254/146mmHg). Initial blood work reported acute kidney failure with a serum creatinine of 4.28mg/dl and an albumin/creatinine ratio of 800mg/g. Hemoglobin and platelet levels were normal, and there was no increase in inflammatory markers. He was admitted to the Nephrology ward for treatment and an etiologic workup. A renal biopsy revealed glomerular thrombi and fibrinoid necrosis, as well as arterial mucoid hyperplasia, leading to the diagnosis of TMA. Histochemical search for amyloid substance was negative. Immunofluorescence study was negative for IgA, IgG, IgM, C1q, C3, and C4. Anti-nuclear, anti-neutrophil cytoplasmic, anti-phospholipid, and anti-glomerular basement membrane antibodies were negative. Haptoglobin levels and coagulation parameters were normal. Viral testing and culture studies were negative. ADAMTS13 activity was 41%, excluding deficiency of this enzyme. Genetic and molecular complement studies were conducted, but no genetic variants were detected. Thoraco-abdomino-pelvic CT scan showed no sign of malignancy. No extra-renal involvement was found. The possibility of renal-limited DITMA was considered, and Etanercept was discontinued. Due to severe kidney failure, he was started on Eculizumab. Adequate tensional control was achieved with Nifedipine and Clonidine. Due to a relapse of his PA, after a multidisciplinary discussion with Dermatology and Nephrology, he was started on anti-IL17 treatment with Ixecizumab in December 2024. Thus far, PA remission was achieved, and although there is sequelar chronic kidney disease, the need for renal replacement therapy has been halted.

Discussion: DITMA is a common cause of TMA which may present without any extra-renal manifestations.

To the authors knowledge, this is the first report of Etanercept induced TMA. Other monoclonal antibodies against tumor necrosis factor alpha have been previously reported to cause TMA. While the exact relationship between drugs and TMA is often unclear, it is important to rule out other causes and stop the offending drug to reduce associated morbidity and mortality.

213 - ERYTHEMA MULTIFORME IN A PATIENT WITH RS3PE ON CORTICOSTEROIDS: A DIAGNOSTIC PARADOX

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Introduction: Remitting Seronegative Symmetrical Synovitis with Pitting Edema (RS3PE) is a rare inflammatory arthritis of the elderly, marked by sudden-onset symmetrical distal synovitis, pitting edema, and negative rheumatoid factor. Erythema multiforme (EM) is an acute immune-mediated mucocutaneous condition, most often triggered by infection, with herpes simplex virus (HSV) being the most frequent precipitator, and less frequently triggered by drugs or autoimmune disorders. The simultaneous occurrence of RS3PE and EM is unusual, particularly considering the paradox of immune mediated cutaneous lesions emerging during corticosteroid therapy. This case seeks to explore this incongruity and reflect on the most plausible etiologies.
Clinical Case: An 81-year-old woman was referred to the Rheumatology outpatient clinic in September 2024, for evaluation of new-onset polyarthritis. Over the preceding month, the patient developed bilateral pain, prolonged morning stiffness and marked swelling of the joints in her hands and feet. Her past medical history included hypertension, dyslipidemia, and hypothyroidism. Regular medications included levothyroxine, rosuvastatin, and lisinopril. On physical examination, she presented with an antalgic gait using a walking stick, symmetrical arthritis of the hands with pronounced pitting edema, and bilateral pedal edema. Laboratory investigations revealed elevated inflammatory markers (ESR 67 mm/h, CRP 7.83 mg/L), leukocytosis (14,800/mm^3), negative rheumatoid factor, and no other autoimmune serologies of note. Based on the above findings, a diagnosis of RS3PE was made and the patient started oral prednisolone with a tapering plan. Due to high osteoporotic fracture risk (FRAX: 24% major, 10% hip), weekly risedronate, calcium, and vitamin D were also prescribed. A workup including bone densitometry was requested. By October 2024, the patient reported significant improvement of joint pain. At that time, she mentioned having experienced a herpes simplex labialis outbreak earlier that month, which had already been resolved by the time of the consultation. There were no signs of active arthritis. prednisolone was reduced with plans for a further taper by January 2025. However, in December 2024, the patient returned to clinic with new erythematous, raised skin lesions on the dorsum of both hands, left lower eyelid, knees, feet, and forearms. These lesions were tender to touch but not pruritic. There was no evidence of arthritis recurrence. Dermatology was urgently consulted, and a skin biopsy was performed. Histology of the biopsy specimen from the right-hand dorsum was not specific but consistent with a dermal variant of EM. Patient was prescribed a potent topical corticosteroid (clobetasol propionate). At follow-up on May 2025, patient was asymptomatic with no recurrence of skin lesions or joint inflammation.
Discussion: RS3PE is an under-recognized cause of acute polyarthritis in older adults, with typical presentation and excellent corticosteroid response. This case illustrates diagnostic complexity when new cutaneous lesions arise during treatment. EM is most commonly triggered by infections, especially HSV, and sometimes by medications. The patient's recent HSV labialis suggests viral reactivation as a possible EM trigger. Bisphosphonates, while uncommon causes, can induce inflammatory reactions including EM. The temporal association of both HSV reactivation and risedronate use suggests a multifactorial etiology. This case emphasizes the need for multidisciplinary care in rheumatologic patients.

214 - CAUSA OU CONSEQUÊNCIA? – UM CASO DE PNEUMONITE INDUZIDA POR METOTREXATO

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A pneumonite induzida por metotrexato (MTX) é uma complicação rara com uma incidência estimada de 0.3% a 7.5%/doentes tratados. Consiste numa pneumonite de hipersensibilidade, de apresentação subaguda, que se não for corretamente identificada e tratada pode conduzir a falência respiratória. O diagnóstico é essencialmente clínico, requerendo um elevado grau de suspeição dada a ausência de clínica específica.

Sexo feminino, 73 anos, seguida em consulta de Reumatologia desde 2022 por Esclerose Sistémica, diagnóstico estabelecido com base na presença de fenómeno de Raynaud, puffy fingers, pitting scars, capilaroscopia com padrão esclerodérmico precoce segundo a definição de Cutolo et al. e positividade de ANA's com doseamento de anti-RP11 e anti-RP155 positivos.

A doente encontrava-se clinicamente estável sob medicação habitual com nifedipina 30mg id, sem indícios de envolvimento órgão-alvo, nomeadamente pulmonar.

Em 2023 inicia espessamento cutâneo com progressão proximal (score Rodnan = 12) e microstomia, pelo que inicia terapêutica com MTX 12.5mg/semana, com titulação até aos 15 mg/semana.

Seis meses após o início do MTX, a doente apresentou quadro de instalação subaguda e com agravamento progressivo, caracterizado por tosse seca, dispneia, febre e anorexia, motivo pelo qual recorreu ao Serviço de Urgência.

Apresentava-se febril (TT 38°C), polipneica, com saturação de oxigénio de 93% em ar ambiente. A auscultação pulmonar revelava diminuição do murmúrio vesicular bibasal. Gasometricamente com hipoxémia normocapnica (paO_2 61mmHg; $paCO_2$ 35mmHg).

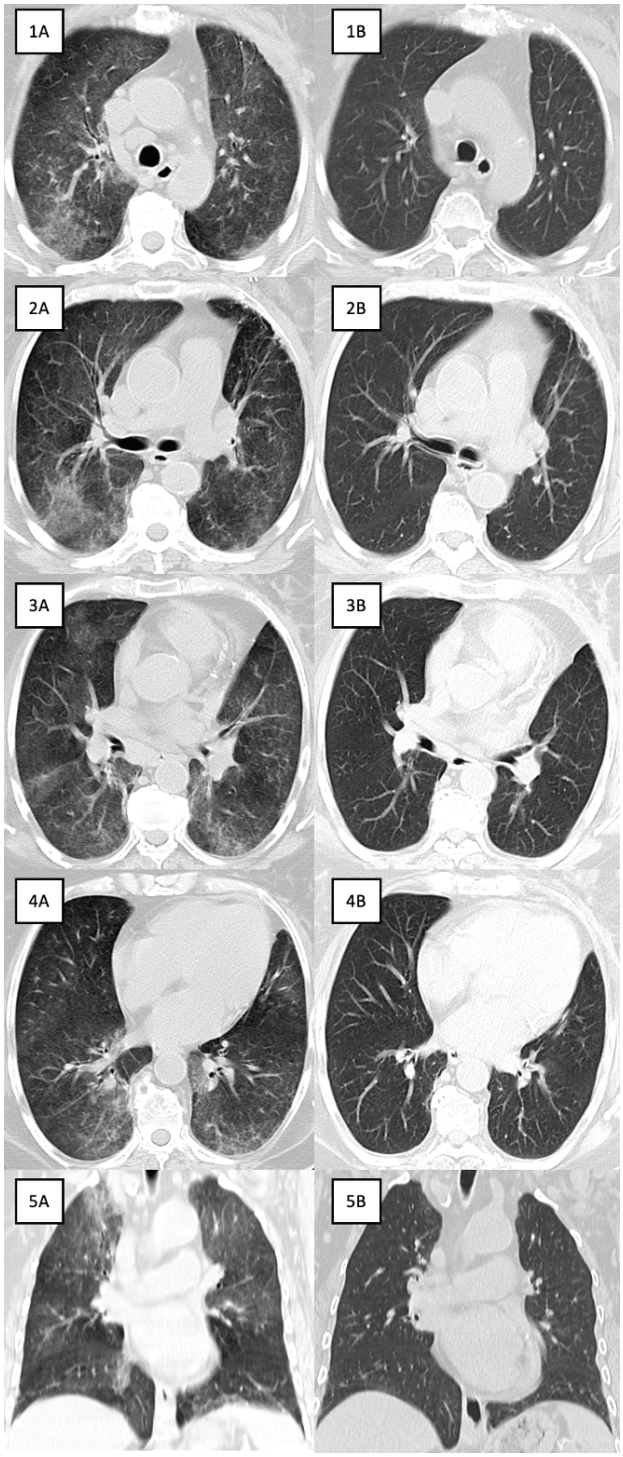
Analiticamente com ausência de leucocitose, elevação da PCR (78,9 mg/L) e procalcitonina e teste SARS-CoV-2 negativos. A radiografia torácica revelou um padrão reticulado peri-hilar, sem evidência de consolidações parenquimatosas.

Perante a suspeita de infeção respiratória em doente imunodeprimida, iniciou antibioterapia empírica com piperacilina-tazobactam e foi internada no serviço de Pneumologia. Por ausência de melhoria clínica ao terceiro dia de internamento, realizou Tomografia computarizada (TC) torácica de alta resolução, que mostrou infiltrados algodonosos nos lobos superior direito e inferiores, com densificação alveolar em vidro despolido, sugestivos de pneumonite. As hemoculturas vieram negativas.

Após discussão com a Reumatologia e perante o elevado grau de suspeita clínica de pneumonite induzida por MTX, o fármaco foi suspenso e iniciou-se prednisolona 40 mg id, com excelente resposta clínica. A doente teve alta hospitalar com plano de redução gradual da dosagem de corticoide e agendamento de reavaliação em consulta a curto prazo.

Realizou nova TC torácica de alta resolução, 6 meses após a alta, que revelou uma resolução completa dos achados imagiológicos prévios

Atualmente, encontra-se assintomática do ponto de vista respiratório, sem progressão do espessamento cutâneo, sob terapêutica com prednisolona 5 mg id, hidroxicloroquina 400 mg id e nifedipina 30 mg id.



214 – Figure 1. TC-AR tórax antes da suspensão do metotrexato (cortes 1-5A) e depois da sua suspensão (cortes 1-5B)

A pneumonite induzida por MTX, embora rara, constitui uma complicação potencialmente grave, pelo que requer um alto grau de suspeição clínica. Neste caso existia uma relação temporal e de causalidade provável com o início da terapêutica com MTX, ausência de resposta clínica com a antibioterapia de alto espetro e resolução completa dos achados imagiológicos com a suspensão do MTX e o início da corticoterapia. Este caso sublinha o desafio diagnóstico inerente a este tipo de pneumonite de hipersensibilidade, especialmente em doentes com patologia reumática sistémica de base sob imunossupressão crónica.

215 - SÍNDROME DA ESCÁPULA ALADA ASSOCIADA A INFEÇÃO POR PARVOVIRUS B19 – UMA ASSOCIAÇÃO MUITO RARA

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Introdução: A síndrome da escápula alada é uma condição rara, causada por disfunção dos músculos estabilizadores escapulotorácicos, mais frequentemente o serratus anterior, por lesão do nervo longo torácico. Pode surgir após trauma, compressão, causas iatrogénicas, infeções ou de forma espontânea. A infeção por Parvovírus B19 raramente se associa a neuropatia periférica, embora estejam descritas formas motoras, aparentemente imunomediadas.

Caso clínico: Doente do sexo feminino, 38 anos, previamente saudável, que recorreu ao serviço de urgência (SU) por exantema generalizado, dores músculo-esqueléticas difusas e talalgias, com início no próprio dia. Relatava síndrome gripal na semana anterior. Foi pedida observação por Reumatologia, destacando-se, ao exame objetivo, edema dos tornozelos, com dor apenas à palpação retro-maleolar externa. Sem alterações analíticas relevantes, com negatividade para Influenza, Covid19, VSR, Treponema pallidum, EBV, HBsAg, HCV e HIV 1 e 2. O quadro foi interpretado como pós-viral e teve alta medicada com Celecoxib 200mg 2x dia.

Recorreu ao SU 48 horas depois, com omalgias bilaterais de novo. Foi administrada hidrocortisona endovenosa e contactada de novo a Reumatologia. Ao exame objetivo, sem limitação/dor à mobilização ativa e passiva dos ombros. Realizou ecografia dos tornozelos, que mostrou edema dos tecidos moles, e dos ombros, sem alterações. Foi realizado switch do AINE para Diclofenac 75mg 2x dia e agendada consulta de Reumatologia após 1 semana. Na consulta, focalizava apenas omalgias bilaterais. Exame objetivo sobreponível. Do estudo analítico, com ANAs/ENAs/ANCAs negativos, serologias para CMV IgG + e IgM -, e serologias para Rubéola IgG + e IgM -. Contudo, salientava-se consumo de complemento (C3 68mg/dl; C4 6mg/dl) e aumento de LDH (347U/L) e transaminases (AST 46U/L; ALT 54U/L). Associou-se prednisolona oral 10mg/dia, com posterior ajuste para 15mg. 14 dias depois, focalizava

215 – Figure 1. A e B – Síndrome de escápula alada

apenas omalgia direita. Repetiu ecografia dos ombros, que não mostrou alterações. Analiticamente, com positividade igG e IgM para Parvovirus B19 (PvB19) (pendente nas consultas passadas), com normalização do complemento/LDH/transaminases. Foi iniciado esquema de desmame do corticoide. Após 4 semanas, já sem corticoterapia e sem omalgia, a doente descrevia, de novo, quadro de fraqueza da omoplata direita. Ao exame objetivo, apresentava escápula alada (figura). Realizou estudo eletromiográfico que documentou "lesão do nervo torácico direito, recente, muito grave". A RM do ombro/omoplata documentou extenso edema muscular do supra e infraespinhoso, e do serratus anterior, sem sinais de rotura, associado a involução lipomatosa grau I nestas localizações. Assumiu-se o diagnóstico de síndrome da escápula alada pós infeção PvB19. Iniciou fisioterapia, com benefício gradual. Repetiu a eletromiografia após 3 meses, compatível com lesão axonal crónica do nervo longo torácico (acometendo apenas o serratus anterior, sem envolvimento do supra ou infraespinhoso).

Conclusão: Este caso ilustra uma complicação neurológica muito rara da infeção por PvB19. A associação temporal com a infeção, serologias positivas e achados eletromiográficos sustentam a relação causal. Embora raramente descrita, a neuropatia periférica por PvB19 pode apresentar envolvimento motor isolado. Este caso destaca a importância de incluir etiologias víricas no diagnóstico diferencial de neuropatias periféricas, sobretudo após infeção recente, com investigação serológica e neurológica dirigida.

216 - ESPONDILODISCITE E TENOSSINOVITE DO TIBIAL ANTERIOR – UM ACHADO RARO, NO CONTEXTO DE INFEÇÃO POR PASTEURELLA MULTOCIDA

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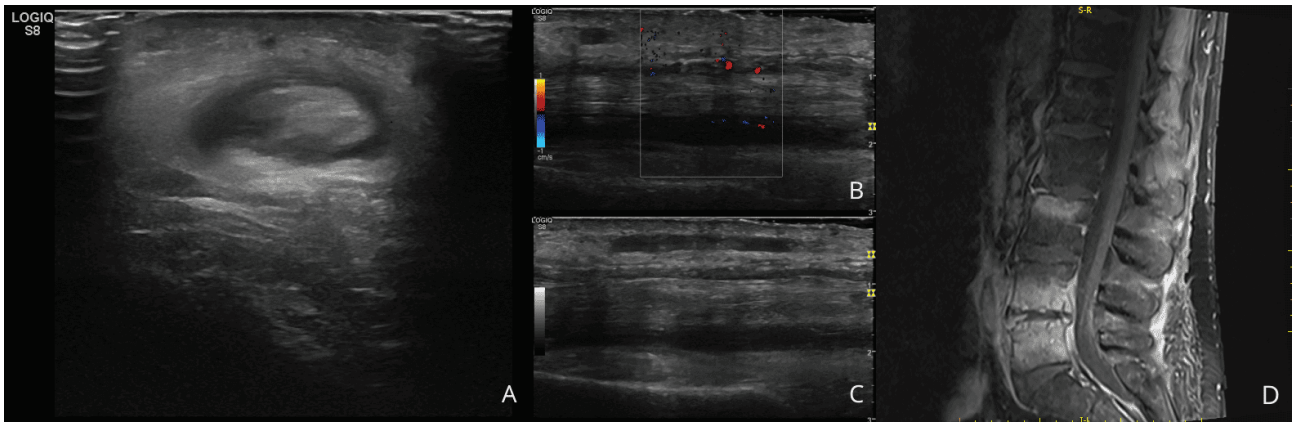
Introdução: A Pasteurella multocida é um cocobacilo gram-negativo comensal da orofaringe de animais domésticos, particularmente gatos e cães. A infecção humana, frequentemente associada a mordeduras ou arranhões, pode assumir apresentações clínicas variadas, desde celulite localizada a quadros sépticos com envolvimento sistémico, especialmente em doentes imunocomprometidos.

Caso clínico: Doente do sexo masculino, 55 anos, de nacionalidade alemã, que recorreu ao serviço de Urgência por quadro de febre, mialgias, lombo-sacralgia e oligoartalgias (ombro direito e tornozelo esquerdo), com 3 dias de evolução, após mordedura de gato. Antecedentes de esplenectomia. Foi admitido em internamento em Medicina Interna, e iniciou antibioterapia endovenosa com amoxicilina e ácido clavulânico. Do estudo analítico colhido, positividade das hemoculturas para Pasteurella multocida, com PCR de 23.19 mg/dL e Procalcitonina de 36.43ng/mL. Foi solicitada colaboração de Reumatologia para esclarecimento das queixas articulares, que o doente descrevia como intensas, que agravavam com movimentos, mas que persistiam em repouso. Objetivamente, era incapaz de mobilizar o ombro direito e tornozelo esquerdo, tanto de forma ativa ou passiva por dor intensa; o tornozelo esquerdo apresentava ainda edema, rubor e calor locais. Foi realizada ecografia articular que, para além de bursite subacromial do ombro direito, documentou tenossinovite do tendão do tibial anterior do tornozelo esquerdo (figura A, B e C). A RM da coluna lombar documentou espondilodiscite de L4-L5 e plataforma



216 – Figure 1. Tenossinovite do tibial anterior do tornozelo esquerdo (A, B e C); RM da coluna lombar, plano sagital (D)

superior de L3 (figura D). Face à apresentação clínica e achados do estudo complementar, foi possível assumir o diagnóstico de quadro séptico associado a Pasteurella multocida, com atingimento multifocal do aparelho músculo-esquelético. A esplenectomia prévia constituiu um fator de risco relevante, dada a natureza capsulada do agente infecioso envolvido. Durante o internamento, o doente manteve antibioterapia dirigida e tratamento sintomático com AINE, com melhoria clínica e analítica progressivas. À data de alta, manteve antibioterapia oral, com posterior normalização dos parâmetros inflamatórios.

Conclusão: Este caso destaca a importância da suspeição e reconhecimento precoce das complicações sistémicas de infeções por Pasteurella multocida, em doentes esplenectomizados. A apresentação clínica com envolvimento multifocal músculo-esquelético e espondilodiscite reflete o potencial de disseminação hematogénea deste agente. A abordagem multidisciplinar e o tratamento antimicrobiano adequado permitiram uma evolução favorável, com recuperação clínica e laboratorial.

223 - SÍNDROME DE ERASMUS: UMA ENTIDADE RARA

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A síndrome de Erasmus é uma entidade rara que se caracteriza pela associação entre Esclerose Sistémica (ES) e exposição prévia à sílica, geralmente por inalação. Com uma prevalência entre 30-290/milhão de habitantes, o termo foi primeiro utilizado por Erasmus em 1957, que descreveu a associação em trabalhadores das minas expostos à sílica.

Doente do sexo masculino, 73 anos, reformado da construção civil, ex-fumador (13 UMA), referenciado à consulta de Reumatologia para estudo etiológico de doença intersticial pulmonar.

Apresentava história de fenómeno de Raynaud bifásico com evolução desde os 40 anos, sem história de úlceras digitais (UD) e sem noção de espessamento cutâneo. Associadamente com queixas de dispneia para médios esforços, com episódios de tosse seca ocasional. Sem outras queixas sistémicas ou focalizadoras de órgão-alvo.

Ao exame objetivo, apresentava espessamento cutâneo dos dedos (score de Rodnan=4) e telangiectasias faciais, sem UD ou pitting scars. À auscultação pulmonar, identificavam-se fervores crepitantes bibasais discretos.

Analiticamente com positividade de ANA 1/320, padrão mosqueado, com anti-Scl-70 positivo (388 UQ; N < 20 UQ) e enzima de conversão da angiotensina de 104,1 U/L (N < 70 U/L).

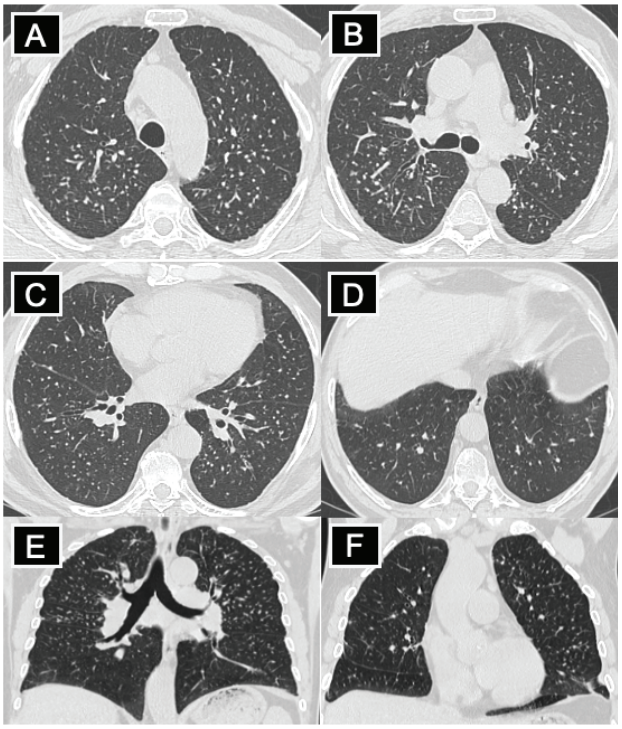
A Tomografia Computorizada (TC) torácica de alta resolução revelava a presença de adenopatias mediastínicas e hilares, e múltiplos micronódulos, bem definidos, distribuídos de forma bilateral e predominando nos lobos superiores e nos segmentos superiores dos lobos inferiores.

Realizou broncofibroscopia, sendo que o lavado broncoalveolar (LBA) evidenciou, uma razão CD4+/CD8+ <3.5 e exames culturais negativos. Foi também realizada biópsia brônquica (BB), na qual estavam ausentes células gigantes multinucleadas e granulomas, afastando a hipótese diagnóstica de sarcoidose.

A videocapilaroscopia revelou a existência de um padrão esclerodérmico precoce, de acordo com os padrões definidos por Cutolo et al.

Estabeleceu-se o diagnóstico de ES e iniciou estudo de envolvimento de órgão-alvo. Após revisão do caso e discussão em reunião multidisciplinar Pneumologia/Reumatologia, concluiu-se que as alterações intersticiais não seriam resultado do envolvimento pulmonar da ES. Assim, perante o contexto profissional de risco, os resultados da LBA e BB e o padrão imagiológico característico da TC tórax, foi colocado o diagnóstico concomitante de Silicose.

Iniciou terapêutica com hidroxicloroquina 400 mg id, metotrexato 12,5 mg/semana, ácido fólico 10 mg semanal e prednisolona 5 mg id, mantendo estabili-



223 – Figure 1. Padrão de silicone em TC-AR tórax (A-D: vista axial; E e F: vista coronal)

dade clínica atual.

Embora ambas as patologias compartilhem o mesmo fator de risco (inalação de sílica), a coexistência de Esclerose Sistémica com Silicose é uma associação rara com potencial de agravamento sinérgico do prognóstico destes doentes.

Este caso clínico destaca a importância da abordagem multidisciplinar para a correta orientação diagnóstica e terapêutica destes doentes

224 - AN UNUSUAL CULPRIT BEHIND JOINT DESTRUCTION: A CASE OF SYRINGOMYELIA-ASSOCIATED CHARCOT ELBOW

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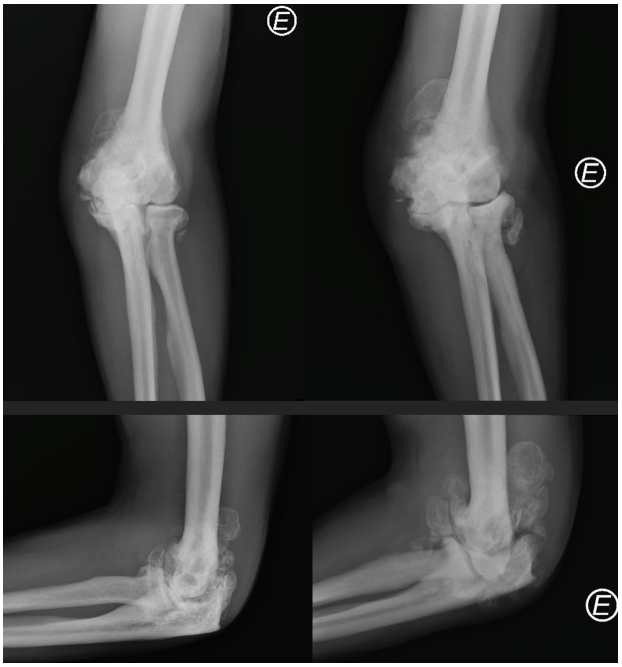

224 – Figure 1. Elbow xray: images on the left from 2018 and images on the right from 2025.

Introduction: Neuropathic arthropathy, or Charcot joint, is a rare but severe form of joint degeneration caused by underlying neurological dysfunction. While it is most observed in the lower extremities—particularly in the context of diabetes mellitus—its manifestation in the upper limbs is uncommon. Syringomyelia is recognized as the predominant cause of Charcot joints in the upper extremity. The condition results from the formation of a syrinx, a fluid-filled cavity within the spinal cord, which disrupts pain and temperature pathways and, over time, leads to motor and sensory deficits. The ensuing joint destruction arises through neurotraumatic mechanisms—repetitive microtrauma and abnormal pressure due to proprioceptive loss—and neurovascular alterations, such as increased osteoclastic activity driven by hyperaemia.

Case report: We present the case of a 64-year-old man with a history of Chiari type I malformation and associated syringomyelia, diagnosed after several years of occipital headache and back pain. He underwent suboccipital craniectomy and C1 laminectomy in 2019. The patient had previously been diagnosed with long-standing ulnar neuropathy and underwent cubital tunnel release in 2016. He was referred due to progressive swelling and reduced mobility of the left elbow over several years, associated with only mild discomfort. Clinical examination revealed marked joint swelling, limitation of both active and passive movement (flexion-extension restricted to 30–90º), and sensory deficits in the upper limbs, predominantly on the left. Radiographs of the elbow showed severe arthrosis with extensive extra-articular ossifications, with clear radiographic progression compared to previous imaging from 2018. Laboratory tests showed no elevation of inflammatory markers, negative rheumatoid factor and anti-CCP antibodies, and normal calcium/phosphate metabolism. Based on the radiographic findings, clinical history, and known syringomyelia, a diagnosis of neuropathic arthropathy of the elbow secondary to syringomyelia was made.

Discussion: Charcot elbow is an uncommon but important differential diagnosis in patients presenting with monoarticular swelling and functional impairment, especially when associated with ulnar nerve involvement (e.g., paraesthesia, atrophy, clawing). Up to 50% of patients report pain, although many remain painless. Imaging is central to diagnosis: plain radiographs often reveal advanced joint destruction and heterotopic ossifications, while cervical MRI is essential for identifying a syrinx in undiagnosed patients. Management should be individualized, focusing on joint protection and treatment of the underlying neurological condition. In the presence of a syrinx, neurosurgical decompression is indicated to halt progression.

Conclusion: This case highlights the importance of considering neuropathic arthropathy of the elbow in the differential diagnosis of chronic monoarthritis with atypical radiographic features, particularly in patients with known syringomyelia or with suggestive symptoms, such as occipital headache exacerbated by Valsalva manoeuvre, back pain, or radicular symptoms.

227 - UNCOMMON SKIN MANIFESTATIONS IN ADULT-ONSET STILL'S DISEASE: THE ROLE OF HISTOLOGY AND BIOMARKERS

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Background: Adult-onset Still's disease (AOSD) is a rare systemic autoinflammatory disorder characterized by heterogeneous clinical presentations. Skin lesions in AOSD typically include a transient, salmon-pink, maculopapular rash; however, persistent or unusual cutaneous findings may mimic other dermatologic or systemic conditions.
Case: We report the case of a 19-year-old female with no relevant medical history who presented with urticarial papular lesions on the thighs and gluteal region following a self-limited episode of odynophagia in September 2024. One month later, the patient developed symmetric polyarthralgia affecting the hands, wrists, elbows, and ankles, and progression of the cutaneous lesions into confluent, erythematous plaques. These plaques extended beyond the initial areas to involve the shoulders, dorsum of the hands and periorbital region. Antibiotic therapy was ineffective, while administration of prednisolone (PDN) at 40 mg/d resulted in full remission of symptoms. However, symptom recurrence occurred during PDN tapering, with new-onset fever.

Upon hospital admission, physical examination revealed periorbital edema, a widespread urticarial rash with dermatographism, polyarthralgia, and generalized hyperalgesia. Laboratory evaluation demonstrated leukocytosis (19,350/µL), neutrophilia (16,400/µL), elevated C-reactive protein (17.34 mg/dL), erythrocyte sedimentation rate (95 mm/h), and hyperferritinemia (1241 ng/mL). Immunologic screening, infectious and neoplastic workups were negative. Dermatology consultation proposed differential diagnoses of urticarial vasculitis (UV) and AOSD. Despite the atypical persistence and morphology of the rash, as well as the absence of classical AOSD features such as serositis or hepatic involvement, AOSD was considered the most probable diagnosis. PDN was reintroduced at 30 mg/day, resulting in marked improvement.

Subsequent skin biopsy revealed neutrophilic urticarial dermatosis (NUD) and serum calprotectin levels were elevated (15 µg/mL), further supporting the diagnosis. The patient was discharged on a regimen of PDN, hydroxychloroquine, and naproxen, and later started on methotrexate, allowing for further tapering of PDN.

Discussion: The differential diagnosis in this case centred on UV and AOSD, both capable of presenting with urticarial lesions and systemic symptoms such as fever and arthralgia. Clinically, UV is characterized by painful or burning lesions lasting over 24 hours, often with residual hyperpigmentation, and histologically by leukocytoclastic vasculitis (1). In contrast, AOSD may manifest with atypical rashes, including persistent urticarial or flagellate lesions, typically associated with intense systemic inflammation (2).

In our patient, the skin biopsy revealed NUD, a pattern commonly associated with autoinflammatory conditions like AOSD (3). The absence of vasculitic changes, combined with markedly elevated ferritin and calprotectin levels and a robust response to corticosteroids, strongly favoured AOSD over UV.
Conclusion: This case underscores the critical role of histology and inflammatory biomarkers in distinguishing clinically overlapping conditions, enabling accurate diagnosis and timely initiation of appropriate therapy.

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228 - SILENT ADRENALS, CRACKING BONES: A GLUCOCORTICOID-NAIVE CAH CASE WITH OSTEOPOROSIS

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Introduction: Congenital adrenal hyperplasia (CAH) is an autosomal recessive disorder, most commonly (over 90% of cases) caused by 21-hydroxylase deficiency (21OHD). This enzymatic defect impairs the conversion of 17-hydroxyprogesterone to 11-deoxycortisol, leading to reduced cortisol synthesis and a compensatory increase in adrenocorticotropic hormone (ACTH) secretion. Chronic ACTH stimulation results in adrenal hyperplasia and excessive androgen production. The clinical severity of CAH correlates with the extent of residual 21-hydroxylase activity, ranging from se-

vere to mild presentations.

Low bone density index (BDI) is a well-recognized concern in both children and adults with CAH, often attributed to prolonged exposure to supraphysiological doses of glucocorticoids (GC). However, the role of GC therapy in bone loss remains controversial, and additional contributing factors such as low body mass index and impaired growth during childhood have been proposed.

We report a case of osteoporosis in a glucocorticoid-naive male patient with undiagnosed CAH, highlighting alternative mechanisms of bone fragility in this context.

Case report: A 62-year-old male was referred to rheumatology for persistent osteoporosis despite three years of treatment with alendronate and cholecalciferol. Dual-energy X-ray absorptiometry revealed a lumbar spine T-score of –3.1 and a femoral neck T-score of –2.0. He had sustained a right femoral fracture at age 49 following a fall from a stepladder but denied any additional fractures or height loss. He reported no history of smoking and occasional alcohol consumption.

His medical history was notable for precocious puberty, infertility, reduced libido, and absence of morning erections. There was no family history of hip fractures or hereditary diseases. His adult height was 145 cm. On physical examination, he presented with small testes within the scrotum and otherwise normal external genitalia.

Hormonal workup revealed elevated ACTH (84.7 pg/mL), markedly increased 17-hydroxyprogesterone (90.1 ng/mL), low total testosterone (148 ng/dL), and low morning cortisol (4.9 µg/dL). A Synacthen stimulation test confirmed adrenal insufficiency, with a peak cortisol of 7.7 µg/dL. Testicular ultrasound identified bilateral, irregularly hyperechoic lesions, consistent with testicular adrenal rest tumours (TARTs).

Genetic analysis identified a homozygous pathogenic variant in the CYP21A2 gene [c.1273G>A p.(Gly425Ser)], confirming the diagnosis of classic congenital adrenal hyperplasia (CAH).

The patient was started on hydrocortisone replacement therapy and antiresorptive treatment with denosumab.

Conclusion: This case highlights an uncommon presentation of CAH, diagnosed in late adulthood in a glucocorticoid-naive patient with severe osteoporosis. It challenges the notion that bone loss in CAH is solely attributable to glucocorticoid therapy and suggests a multifactorial aetiology, potentially involving chronic androgen excess, hypogonadism, and early growth disturbances. Clinicians should maintain a high index of suspicion for CAH in patients with unexplained osteoporosis and signs of long-standing hormonal imbalance.

229 - THE WRISTS THAT WOULDN'T HEAL: REFRACTORY STILL'S DISEASE REQUIRING BILATERAL ARTHRODESIS

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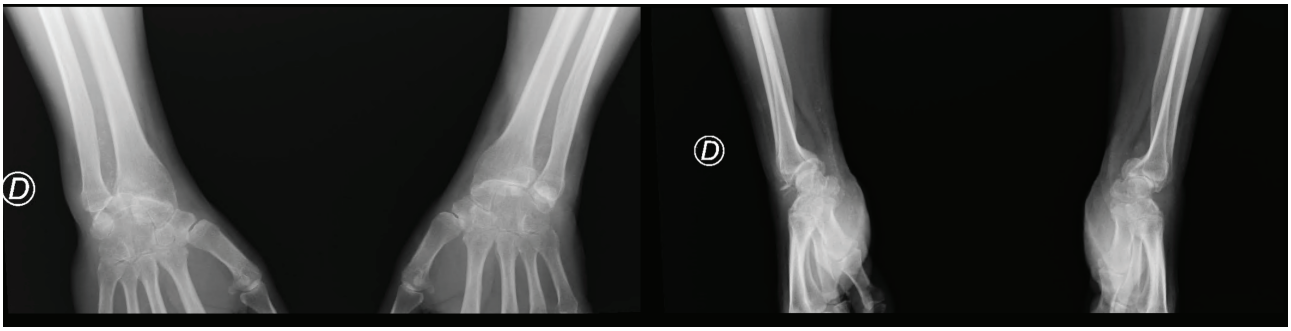
Introduction: The management of Still's disease has evolved significantly with the introduction of biologic disease-modifying anti-rheumatic drugs (bDMARDs), particularly IL-1 and IL-6 inhibitors, which have demonstrated efficacy in both systemic and articular phenotypes. Despite therapeutic advances, a subset of patients may develop refractory disease, particularly with persistent articular involvement, leading to irreversible joint damage and functional disability. To our knowledge, this is the first reported case Still's disease with refractory, destructive arthritis requiring total wrist arthrodesis.

Case report: We report the case of a 44-year-old man diagnosed with Still's disease in 2019. Initial symptoms included headache, odynophagia, myalgia, fever, and inflammatory polyarthralgia involving the shoulders, wrists, and knees. He also reported abdominal discomfort and a transient erythematous rash predominantly on the trunk. Cardiac symptoms prompted further evaluation, revealing myopericarditis. Additionally, he developed pleural effusion with dyspnoea and peripheral desaturation, requiring oxygen supplementation (3 L/min).

Initial therapy with methotrexate, corticosteroids, and colchicine proved insufficient to control disease activity, and multiple bDMARDs were sequentially introduced. Anakinra (Dec 2019–Aug 2020) was discontinued due to secondary failure. Tocilizumab (Aug 2020–Jan 2021) did not control oligoarthritic with persistent hyperferritinaemia. Canakinumab (Apr 2021–Mar 2023) achieved systemic control but with ongoing articular activity. After a brief suspension, Canakinumab was reintroduced (Nov 2023–Apr 2024), again with limited articular response. Rituximab (from Apr 2024) and adalimumab (May–Jun 2024) also failed to provide clinical benefit. Infliximab (from Sep 2024) was well tolerated systemically, but did not resolve wrist arthritis. Disease control was further complicated by two infectious events: a hepatitis B virus infection and pneumocystosis (both in 2024), which delayed optimization of immunosuppressive therapy.

By early 2025, the patient presented with marked



229 – Figure 1. Wrist xray

functional impairment and pain in both wrists. MRI revealed advanced osteoarthritis with cartilage loss and multiple erosions, rendering radiosynovectomy unfeasible. Although JAK inhibitors were considered, they were ultimately dismissed due to the extent of joint destruction and severe disability. Considering the refractory articular disease and irreversible structural damage, total wrist arthrodesis was proposed. The procedure was successfully performed on the right wrist in June 2025, with the left wrist surgery planned subsequently. Routine blood tests at the time of surgery were unremarkable, including inflammatory markers.

Discussion: Although Still's disease is often responsive to immunosuppressive therapy, a small subset of patients develops refractory articular manifestations despite multiple lines of targeted biologic treatment. In this case, the patient showed inadequate articular response to IL-1 and IL-6 inhibitors, anti-CD20, and TNF inhibitors, ultimately progressing to severe bilateral wrist damage over a 6-year disease course. Joint destruction in Still's disease is rarely reported, and this case underscores the potential for aggressive, treatment-resistant articular involvement despite optimal pharmacologic management. To the best of our knowledge, this is the first reported case of Still's disease requiring total wrist arthrodesis due to persistent arthritis and advanced joint destruction.

231 - A RARE CASE OF JUVENILE-ONSET IGG4-RELATED DISEASE INVOLVING THE CENTRAL NERVOUS SYSTEM

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Introduction: IgG4-related disease (IgG4-RD) is a chronic, immune-mediated fibroinflammatory disorder with multisystem involvement including the central nervous system (CNS). Although rare, IgG4-RD CNS involvement most commonly manifests as hypertrophic pachymeningitis with a range of neurological symptoms including headache, cranial neuropathies, seizures, and visual disturbances. Even less frequently, IgG4-RD may present with cerebral mass-like lesions mimicking neoplasia.

Case Report: A 9-year-old boy presented with a history of headache and right sided proptosis and was found to have a right anterior cranial fossa mass, extending in the ethmoidal air sinuses and, intracranially, in the right frontal lobe. He had two needle biopsies of the right ethmoidal mass that were inconclusive: one at 10 years old, which showed a mixed inflammatory infiltrate, with predominantly lymphocytes and no neoplastic infiltration; and at 16 years old, after he had developed seizures for the first time, a second biopsy confirmed findings consistent with an inflammatory fibroblastic tumour. Afterwards, he remained in clinical remission until age 23, when he had a recurrence of seizures. He reported no other symptoms. A magnetic resonance imaging (MRI) scan showed marked progression of the mass in the right frontal lobe, with extension to the left lobe. A PET-CT scan excluded peripheral disease, so he underwent bifrontal craniotomy and resection of the dura tumour, whose histological diagnosis was consistent with IgG4-RD. Imunossupression was started with steroids and a single cycle of rituximab 1+1g. A second MRI scan was compatible with residual IgG4-related hypertrophic pachymeningitis. He remained stable without immunosuppressive treatment until age 29 when he presented with sudden visual loss in his right eye (only count fingers acuity). Ophthalmology observation and MRI scan confirmed subacute right retrobulbar optic neuropathy due to recurrence of disease in the anterior cranial fossa with enhancing soft tissue completely surrounding the intracranial portion of the right optic nerve. Laborato-

ry tests were notable for an increase in serum IgG4 of 2.14 g/L (reference range <1.3), C-reactive protein (31.3 mg/L) and erythrocyte sedimentation rate (26 mm). He was subsequently treated with 3 pulses of intravenous methylprednisolone 1g, followed by prednisolone 60mg/day (with progressive tapering to 5mg), mycophenolate mofetil maximum 3g/day and rituximab 1+1g followed by 1g every 6 months. At 3 months, an orbit MRI showed decreased volume of enhancing disease. After 2 years, he has remained stable, although not in complete imaging remission, with a plan to continue current treatment for, at least, a total of 3 years.

Discussion/ Conclusion: CNS involvement in IgG4-RD is rare, and this case is further unusual due to its juvenile onset and lack of systemic involvement. Fewer than 60 cases of juvenile-onset IgG4-RD have been reported, typically showing no male predominance, more frequent single-organ and unilateral involvement, and a predominance of ophthalmic and/or "head and neck–limited" disease. While corticosteroids remain the first-line treatment due to the disease's typical responsiveness, recurrence is common. Long-term management often requires steroid-sparing agents such as rituximab, either alone or in combination with immunosuppressants like mycophenolate mofetil.

This case highlights the importance of considering IgG4-RD in paediatric patients with isolated CNS disease and supports the role of early immunomodulatory therapy to prevent relapse.

235 - ISOLATED RHEUMATIC MANIFESTATIONS IN WHIPPLE DISEASE: AN EARLY DIAGNOSIS?

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Introduction: Whipple's disease is a rare systemic infection caused by Tropheryma whipplei, mostly affecting middle-aged Caucasian men. Associated with poor hygiene and wastewater exposure, it classically evolves in three phases: an early phase (<6 years) with arthralgia and fever; a middle phase (6–8 years) with diarrhoea, abdominal pain and weight loss; and a late phase (>8 years) marked by neurological symptoms. Since seronegative arthritis and/or arthralgia are often early isolated manifestation, misdiagnosis as a rheumatic disease — particularly seronegative rheumatoid arthritis and spondylarthritis — is common, delaying correct diagnosis by an average of 6.7 years.

Case report: We present the case of a 50-year-old Caucasian male with a 1.5-year history of symmetric additive polyarthralgia involving the metacarpophalangeal joints, wrists, elbows, knees, ankles, and feet. The patient presented with episodes of worsening pain and swelling that significantly impaired his gait. Laboratory tests revealed elevated inflammatory markers, with a C-Reactive Protein (CRP) of 4mg/dL and an Erythrocyte Sedimentation Rate (ESR) of 53 mm/h. Musculoskeletal ultrasound showed mild-to-moderate synovitis in small joints of the hands and feet.

The patient underwent sequential treatment with methotrexate, deflazacort, colchicine, leflunomide over 1.5 years, without significant clinical improvement. He was then treated with adalimumab for a month, with clear worsening and immediate treatment suspension.

Extensive immunologic, infectious, and oncologic workup was negative. During this work-up, Helicobacter pylori was isolated in gastric biopsies and treated with bismuth, metronidazole and tetracyclines, with transitory improvement. Nearly two years after initial evaluation, synovial fluid from the extensor tendon sheath of the left hand was sent for analysis and tested positive for Tropheryma whipplei by PCR. Upper gastrointestinal endoscopy confirmed the diagnosis through positive duodenal PCR.

Immunosuppressive therapy was discontinued, and the patient received intravenous ceftriaxone (2g/day for 15 days), followed by oral sulfamethoxazole/trimethoprim (800/160 mg twice daily). Clinical improvement was rapid, with significant reduction in joint swelling and restoration of gait function. Laboratory markers also improved, with CRP decreasing from 6.2 to 0.75 mg/dL within five days of antibiotic therapy initiation.

After one month, a slight worsening of ankle arthralgia was noted, accompanied by a mild elevation of CRP (3.29 mg/dL). An immune reconstitution inflammatory syndrome (IRIS) was assumed and managed successfully with a low dose of prednisolone (5 mg/day). At three-month follow-up, the patient remained asymptomatic, and corticosteroid tapering was initiated.

Conclusion: Timely recognition of Whipple's disease is crucial in patients presenting with atypical rheumatologic symptoms, particularly when subtle systemic signs or elevated inflammatory markers are also present. Worsening arthritis or new systemic manifestations following immunosuppressive treatment should raise clinical suspicion. Including Whipple's disease in the differential diagnosis of persistent seronegative arthritis may enable earlier identification and timely

initiation of antibiotic therapy, thus reducing the risk of disease progression and complications. Given its ability to mimic other rheumatic conditions, and the potential harm of delayed diagnosis, rheumatologists should be watchful of this rare but treatable disease.

236 - SHOULDER PAD SIGN: THE PATH TO AMYLOIDOSIS

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Introduction: Bilateral shoulder swelling is an uncommon clinical finding and should prompt a comprehensive diagnostic workup. The differential diagnosis includes articular and periarticular conditions of inflammatory, infectious, traumatic, degenerative, or neoplastic origin. We describe a case of bilateral subacromial bursitis refractory to standard therapy, in which the shoulder pad sign served as a key diagnostic clue to systemic AL amyloidosis.

Case Description: A 73-year-old woman with a history of renal transplantation and multiple myeloma was referred from physiatry for persistent bilateral shoulder pain and swelling. She had been treated with local corticosteroid injections and high-dose systemic prednisolone, initially prescribed for unexplained proteinuria (inconclusive renal biopsy), without clinical improvement.

She reported mechanical-type shoulder pain, exacerbated by abduction and lateral decubitus, with a duration of approximately two years. There was no history of trauma, systemic symptoms, or involvement of other joints. On physical examination, she presented with symmetrical bilateral shoulder swelling and pain on active mobilization. Ultrasound imaging revealed marked bilateral subacromial bursitis with power Doppler signal and concomitant extensive glenohumeral synovitis. Joint aspiration yielded turbid synovial fluid with low cellularity, no crystals, and negative cultures, consistent with a non-inflammatory effusion.

Laboratory investigations showed normal inflammatory markers but revealed an elevated serum free light chain ratio and monoclonal IgA lambda on serum protein electrophoresis. Given the high clinical suspicion for AL amyloidosis, a re-evaluation of the renal graft biopsy was performed, which was negative for amyloid deposits. While awaiting a scheduled subacromial bursa biopsy, a second renal biopsy was performed. Both specimens demonstrated Congo red-positive amyloid deposition, establishing the diagnosis of systemic AL amyloidosis with articular involvement. The patient was referred to Haematology, where she initiated targeted chemotherapy with subsequent clinical improvement.

Conclusion: This case illustrates the diagnostic value of the shoulder pad sign and highlights amyloid arthropathy as a rare but important cause of chronic bilateral subacromial bursitis. In patients with underlying plasma cell dyscrasias, the presence of symmetrical shoulder swelling should prompt consideration of AL amyloidosis. Early recognition and histopathological confirmation are critical to initiate disease-modifying treatment and improve prognosis.

238 - THE VIRUS ON THE TIP OF THE TONGUE: CMV IN A VASCULITIS PATIENT

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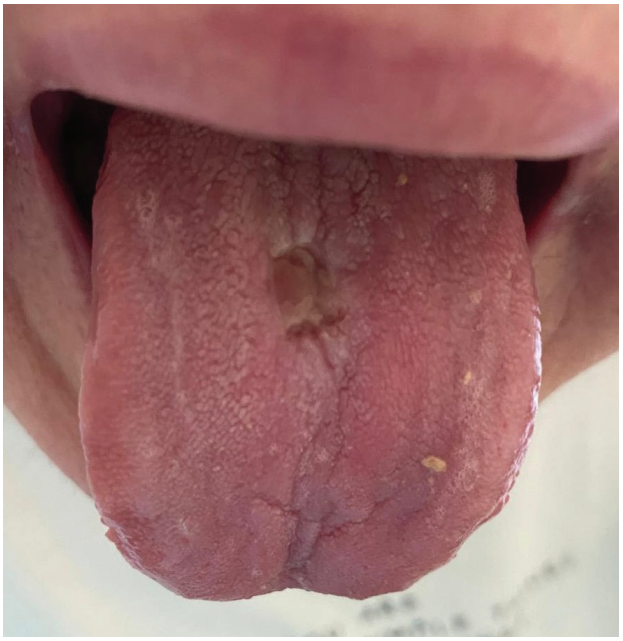
Introduction: Cytomegalovirus (CMV), a member of the Herpesviridae family, typically causes asymptomatic or mild self-limiting illness in immunocompetent individuals. In contrast, in immunocompromised hosts, CMV may lead to severe systemic disease with multiorgan involvement, including less commonly the skin and mucous membranes. Oral ulcers due to CMV are rare, often painful, and have been primarily described in patients with HIV, those undergoing chemotherapy, or organ transplant recipients receiving immunosuppressive therapy.

Case Report: We present the case of a 54-year-old woman with a longstanding diagnosis of polyarteritis nodosa, with prior ocular, cutaneous, and neurological manifestations. Due to new-onset mononeuropathy of the right sural nerve, she was initiated on oral prednisolone 60 mg/day and oral cyclophosphamide 150 mg/day. Two weeks into treatment, the patient developed a painful ulcer on the dorsal surface of the tongue, in the absence of systemic symptoms, initially attributed to underlying vasculitis.

Due to worsening pain and neurological deterioration, with new involvement of the ulnar nerve consistent with mononeuritis multiplex, the patient was admitted to the rheumatology ward. During hospitalization, a biopsy of the tongue ulcer was performed, and



238 – Figure 1. Tongue ulcer previous to biopsy

her immunosuppressive regimen was subsequently switched to intravenous cyclophosphamide according to the CYCLOPS protocol, along with prednisolone 30 mg/day.

Histopathological examination revealed eosinophilic nuclear and cytoplasmic inclusions, with immunohistochemical staining positive for CMV. Serological testing was positive for both CMV IgG and IgM, and blood PCR demonstrated a viral load of 4790 IU/mL. In light of active CMV infection and the associated risk of reactivation, cyclophosphamide was temporarily discontinued, and prednisolone was tapered to 20 mg/day. Antiviral therapy with intravenous ganciclovir 500 mg twice daily was initiated and maintained for 21 days, resulting in a reduction of viral load to <34.5 IU/mL. The patient was then transitioned to oral valganciclovir 900 mg/day for maintenance during continued immunosuppression. Cyclophosphamide was reintroduced after 21 days of antiviral therapy, with no subsequent evidence of viral reactivation.

Conclusion: This case illustrates an uncommon presentation of CMV infection manifesting as a solitary tongue ulcer in an immunocompromised patient. The absence of systemic symptoms posed a diagnostic challenge, particularly in the context of underlying autoimmune disease. CMV-related oral ulcers may mimic manifestations of rheumatic disease, malignancy, or traumatic lesions. Histopathological confirmation is essential for accurate diagnosis and timely initiation of targeted antiviral therapy, preventing potentially severe complications in immunosuppressed individuals.

240 - BEYOND THE USUAL SUSPECTS: CRYPTOSPORIDIUM AS A RARE CAUSE OF DIARRHOEA IN SLE

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Introduction: Systemic lupus erythematosus (SLE) is a complex autoimmune disease characterized by multisystem involvement and unpredictable flares. While disease activity is often idiopathic, infectious triggers — particularly in immunosuppressed individuals — may precipitate flares and complicate management. Cryptosporidium spp. is a protozoan parasite known to cause gastrointestinal infections, especially in immunosuppressed hosts. However, its potential to trigger diarrhoea and a lupus flare has not, to our knowledge, been previously reported in the literature.

Case report: We present the case of a 21-year-old female with a recent diagnosis of systemic lupus erythematosus (SLE), with cutaneous, articular, hematologic, and renal involvement (class II lupus nephritis). She was under treatment with hydroxychloroquine 400 mg, mycophenolate mofetil 1000 mg BID, and prednisolone 60 mg.

Shortly after an increase in the dose of mycophenolate, she developed profuse watery diarrhoea, initially presumed to be drug related. Despite dose reduction and oral rehydration, symptoms worsened, prompting emergency department admission. Laboratory results revealed acute kidney injury (AKIN stage 2), leucocytosis with neutrophilia, hyponatremia, and hyperkalaemia, raising concerns for an infectious trigger and a possible lupus flare, further supported by the presence of a malar rash.

Due to persistent symptoms, low-grade fever, and worsening cutaneous lesions, hospital admission was warranted. Mycophenolate was suspended, empiric antibiotic therapy was initiated, and stool studies were performed, revealing a positive Cryptosporidium spp. antigen.

Following discussion with Infectious Diseases, tentative treatment with nitazoxanide 500 mg BID was started, along with intravenous immunoglobulin (2 g/kg over 3 days). The patient showed marked clinical improvement within 48 hours, including reduced bowel frequency, improved fatigue, and resolution of the cutaneous flare. Renal function also normalized

rapidly, suggesting that the acute kidney injury was more likely due to dehydration and infection rather than a lupus nephritis flare — the latter of which could only be confirmed by renal biopsy.

Conclusion: This case represents a rare — and possibly first-reported — instance of an SLE flare precipitated by Cryptosporidium-associated gastroenteritis. It highlights the importance of considering Cryptosporidium spp. as a potential trigger for both diarrhoea and disease activity in patients with lupus. Moreover, it supports the off-label use of nitazoxanide as a therapeutic intervention in selected cases involving immunocompromised patients. Recognizing such associations is crucial for guiding timely diagnosis, targeted infectious screening, and personalized therapeutic interventions in autoimmune patients presenting with gastrointestinal symptoms.

248 - A NEUROLOGICAL TWIST IN A PATIENT WITH INFLAMMATORY MYOPATHY: A RARE CASE REPORT

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Inflammatory myopathies can pose a diagnostic challenge due to the overlap of their cardinal symptom – muscle weakness – with various other syndromes, namely of metabolic and neurological origin. When a disease flare in a patient with known myositis is suspected, identifying such mimickers becomes even more complex, requiring careful clinical assessment and often further complementary workup.

We present the case of a 76-year-old female patient followed in our Rheumatology department due to a recent diagnosis of anti-Mi2 antibody dermatomyositis with musculoskeletal (including oesophageal), cutaneous and systemic involvement. At this time, she was under treatment with monthly intravenous immunoglobulin (IvIg) (2g/kg/month) and low-dose prednisolone (5mg/kg/day) - with no additional immunosuppression due to an infectious intercurrence – with ongoing improvement. Her remaining medical history was positive for asthma, hypertension and a 3rd degree atrioventricular block.

Two days prior to the scheduled 3rd cycle of IvIg, the patient presented to the Emergency Department (ED) with a 1-week history of major relapse of muscle weakness and left ptosis. On ED observation, severe tetraparesis was recognized and a dermatomyositis flare was assumed. Hence, prednisolone was titrated to 20mg/day, and the patient was admitted at our care.

Laboratory analysis with complete blood count, inflammatory markers, and muscle, hepatobiliary, thyroid and renal parameters were within normal range. Thoracic radiograph, electrocardiography and urinalysis were uneventful.

Upon joint observation by Rheumatology and Neurology, not only was a proximal-predominant quadriparesis evident but the patient also exhibited severe bilateral facial paresis, ophthalmoparesis and diplopia, severe hypoesthesia of the trunk and limbs, ataxia, pseudoathetosis and absent deep-tendon reflexes. So, what was initially integrated as a flare of dermatomyositis, had otherwise unexplained neurological features, to be specific, profound sensory impairment, ataxia, and facial and extraocular musculature involvement.

Investigation of the cerebrospinal fluid (CSF) revealed elevated protein (63.8 (NR:15-45mg/dL), with albuminocytologic dissociation (leucocytes<2/µL) but culture and search for autoantibodies (namely, anti-gangliosides) and neurotropic viruses were unrevealing. Electromyography revealed loss of F waves and low amplitude action potentials with normal velocity, findings suggestive of axonal neuropathy. Encephalic and radicular lumbosacral magnetic resonance imaging were unremarkable.

Hence, the patient was instead diagnosed with Miller Fisher syndrome: an immune-mediated neuropathy classically represented by the triad of ataxia, areflexia, and ophthalmoparesis, along with ptosis, facial nerve palsy, hypoesthesia and paresis.

Ongoing IvIg therapy was continued, as it is a recommended treatment for such cases, along with corticosteroids. In less than a month, the patient displayed a major recovery of her deficits, fully regaining sensation and the ability to walk, in line with the favourable response to therapy reported for this syndrome.

We thus describe two rare immune-mediated entities with overlapping presentations coexisting in the same patient, a situation which is seldom described in the literature. This case underlines the need for a broad clinical reasoning at every patient encounter and emphasizes the value of the clinician's observation and their role in accurately interpreting signs and symptoms, not only when they align with the diagnosis but also when they do not.