Musculoskeletal tuberculosis presenting as polyarthritis: a diagnostic challenge

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Abstract

Background: In adults, unilateral facial thickening carries a wide differential including congenital, neoplastic, inflammatory, autoimmune, endocrine, and fibro-osseous causes. Early on, several of these can mimic an autoimmune connective tissue disease, making diagnosis a real challenge in rheumatology. We report a case highlighting the diagnostic overlap between Parry–Romberg syndrome and morphea.

Case description: A 28-year-old woman presented with left-sided facial changes progressing over approximately 1.5 years, beginning with edema and erythema of the left hemiface for roughly five months, followed by progressive volume loss of the nasolabial region and upper lip (Figure 1). She reported intermittent facial paresthesias but no Raynaud's phenomenon, digital ulcers, dysphagia, arthritis, seizures, focal deficits, or visual symptoms, and no contralateral facial/limb atrophy, dental/oral changes, prior trauma, or congenital asymmetry. History included asthma, prior smoking, a family history of psoriasis, and recent travel before onset. Examination showed clear left hemifacial asymmetry with soft tissue atrophy of the nasolabial fold and upper lip, no sclerodactyly or arthritis, and normal neurological exam. Antinuclear antibodies were low-titre (1:160, speckled) with negative ENA; inflammatory markers, muscle enzymes, and infectious workup were unremarkable. Brain MRI showed only an incidental, nonspecific white matter lesion. Skin biopsy (Figure 2) revealed epidermal atrophy, a perivascular lymphoplasmacytic infiltrate, and dermal/hypodermal fibrosis. Deep hypodermal involvement, upward displacement of adnexal structures, and absence of marked dermal sclerosis favoured Parry–Romberg syndrome over linear scleroderma. She started methotrexate 10 mg/week with folic acid; at follow-up, disease activity had stabilised, with reconstructive treatment (autologous fat grafting, Coleman technique) planned after further confirmed stability.

Discussion/Conclusions: Parry–Romberg syndrome follows a biphasic course—an inflammatory phase mimicking autoimmune disease, followed by progressive atrophy—making early misclassification common. Biopsy was key to distinguishing it from morphea, which typically shows dense dermal sclerosis with hyalinized collagen. Clinicians should consider Parry–Romberg syndrome in the differential of progressive unilateral facial changes and pursue early biopsy, since its atrophic phase can closely overlap with morphea both clinically and histologically.

Keywords: Parry–Romberg syndrome; Hemifacial atrophy; Morphea; Localized scleroderma; Skin biopsy.

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Key Messages:

- Recognize Parry–Romberg syndrome as a differential for progressive unilateral facial changes that can mimic autoimmune connective tissue disease.
- Know the key biopsy findings that distinguish Parry–Romberg syndrome from morphea.
- Understand the general treatment approach: immunosuppression to stabilise disease, followed by reconstructive surgery

Unilateral facial thickening in adults includes several differential diagnosis, ranging from congenital and neoplastic to inflammatory, autoimmune, endocrine, and fibro-osseous causes. In early stages, these presentations may resemble autoimmune connective tissue diseases, which can make diagnosis particularly challenging in rheumatology. Parry–Romberg syndrome (PRS), or progressive hemifacial atrophy, shares clinical and histopathological features with craniofacial morphea, and the two are increasingly regarded as part of an overlapping disease spectrum[1-3]. PRS is rare (0.3–2.5/100,000/year), predominantly affects females, and usually presents within the first two decades of life[3-4].

A 28-year-old woman was evaluated for progressive unilateral facial changes over approximately 1.5 years, initially presenting with inflammatory swelling and later evolving to localized atrophy (Figure 1, Panels A and B). The condition began with edema and erythema lasting around five months, followed by gradual atrophy of the nasolabial region and upper lip. She also described intermittent facial paresthesias, but denied Raynaud's phenomenon, digital ulcers, dysphagia, inflammatory arthritis, skin involvement beyond the face, seizures, focal neurological deficits, or visual complaints. No contralateral limb atrophy, dental/oral abnormalities, constitutional symptoms, prior trauma, or congenital facial asymmetry were identified. Her past history included asthma; she was an ex-smoker with a family history of psoriasis and recent travel prior to onset, which initially raised concern for an infectious cause.
On examination, there was clear left hemifacial asymmetry, with soft tissue atrophy involving the nasolabial fold and upper lip. No sclerodactyly, digital ulcers, or inflammatory arthritis were observed, and neurological examination was unremarkable, with no evidence of trigeminal involvement. She was referred to Infectious Diseases, Neurology, and Dermatology. Laboratory studies showed low-titre antinuclear antibodies (1:160, speckled), with negative extractable nuclear antigens (including systemic sclerosis- and myositis-specific antibodies); inflammatory markers and muscle enzymes were normal, and an extensive infectious workup was negative.

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Brain MRI revealed only a small nonspecific white-matter lesion, considered incidental — white-matter abnormalities being among the most frequently reported neuroimaging findings in PRS and often clinically silent[6]. Skin biopsy (Figure 2) showed epidermal atrophy, a perivascular lymphoplasmacytic infiltrate, and fibrosis involving the dermis and hypodermis, arguing against panniculitis. The predominance of hypodermal involvement, with upward displacement of adnexal structures and without marked dermal sclerosis or dense collagen hyalinization, favored PRS over linear scleroderma[2]. A diagnosis of PRS was established, and methotrexate 10 mg weekly with folic acid was started, aiming for disease stabilization over 12–24 months before reconstructive procedures (e.g., Coleman technique)[7].

PRS typically follows a biphasic course, with an initial inflammatory phase resembling autoimmune connective tissue disease, followed by progressive atrophy, which can lead to misclassification early on[1]. Here, the initial presentation raised several differentials, including post-infectious panniculitis, localized scleroderma/morphea, autoimmune connective tissue disease, and trigeminal nerve disorders. The absence of systemic involvement, negative disease-specific autoantibodies, normal markers, negative infectious workup, and unremarkable neurological/MRI findings argued against these alternatives. Histopathology, with predominant deep dermal and hypodermal fibrosis rather than the prominent dermal sclerosis and hyalinized collagen typical of morphea, further supported PRS[2]. This case illustrates the importance of considering PRS in progressive unilateral facial changes.

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Tables and Figures

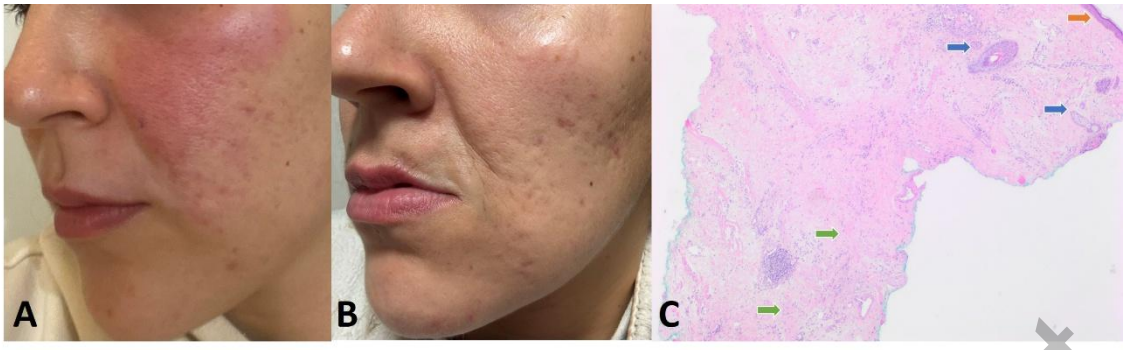


Figure 1. Clinical and histopathological features of Parry–Romberg syndrome. (A) Early inflammatory phase showing unilateral erythema and edema involving the left hemiface. (B) Progression to hemifacial atrophy over approximately 1.5 years, with marked volume loss of the nasolabial region and upper lip, consistent with Parry–Romberg syndrome. (C) Skin biopsy demonstrating epidermal atrophy (orange arrow), upward displacement of adnexal structures within the dermis (blue arrows), and fibrosis involving the hypodermis (green arrows), findings supporting the diagnosis of Parry–Romberg syndrome (hematoxylin and eosin stain, ×20).

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