Late progression of neurological symptoms and MRI T2 hyperintensities in Parry-Romberg syndrome

We describe a case of a 50-year-old woman who developed drug-resistant status epilepticus with complex partial and secondary generalised seizures. She had suffered from Parry-Romberg syndrome (PRS) for more than 40 years. Right-sided progressive hemifacial atrophy (PHA) had begun at the age of 7 (figure 1A), followed by epilepsy at the age of 14 years. In the past 2–3 years before the latest admission, the patient had developed a progressive left-sided hemiparesis concomitant with an increase of T2-hyperintensities in the white matter of the right hemisphere, ipsilateral to the PHA (B–D). Moreover, MRI scans illustrated ipsilateral cerebral atrophy (B–D). Blood-sensitive axial imaging revealed evidence of microhaemorrhages or microcalcifications in the right hemisphere (E). Cerebral atrophy was accompanied by contralateral cerebellar atrophy (F). Intrathecal IgG-synthesis in a brain biopsy (G) confirmed neuroinflammation as a potential pathophysiological correlate of PRS.1,2 Glucocorticoid treatment subsequently stabilised the late clinical progression.

Typically, PRS shows initial manifestation in the first 20 years of life and then progresses slowly over the following 2–20 years before reaching quiescence.3 However, late onset until the sixth and seventh decades of life has also been described.1,3 Our patient showed a comparatively late progression, more than 40 years after disease onset. As a neurocutaneous disorder, PRS is characterised by PHA, which shows a clinical overlap with localised scleroderma (morphoea en coup de sabre) as well as by central nervous system involvement, the most common extracutaneous finding.1,3 Epilepsy, often refractory to anticonvulsive medication, and headache, are the most common neurological manifestations. Other manifestations include cranial neuropathies as well as cerebrovascular abnormalities and vascular brain lesions, leading to weakness and atrophy of the contralateral extremities. An autoimmune-mediated pathophysiological mechanism of PRS has been postulated.1–4 Focal vascular dysfunction due to localised autoimmune-mediated vasculitis may lead to the clinical picture of PHA and diverse neurological symptoms. Evidence for inflammatory processes are given by the following findings in PRS: perivascular and diffuse lymphocytic infiltration in biopsy specimens, local immunoglobulin synthesis in cerebrospinal fluid, occasional coexistence with other autoimmune disorders and frequent improvement of the clinical course following immunosuppression. Three of these four inflammatory characteristics existed in our patient.

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Figure 1  Brain MRI follow-up study. Right hemifacial atrophy (A). Axial imaging revealed a distinct progression of T2 signal hyperintensity in the white matter of the right hemisphere, with corresponding atrophy demonstrated by serial MRI over 3 years (B–D). T2*-weighted axial imaging revealed microhaemorrhages or microcalcifications in the right hemisphere (arrows in E). Cerebral atrophy was accompanied by contralateral cerebellar atrophy (arrow in F, fluid-attenuated inversion recovery-weighted coronal imaging). Perivascular T-cell infiltration in two different regions (staining with CD3 and H&E; x200; scale bar 100 μm).
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