ORIGINAL RESEARCH PAPER

Radio-Diagnosis

KEY WORDS: Parry–Romberg Syndrome (PRS), progressive hemifacial atrophy, neurocutaneous disorder, facial hemiatrophy, fronto-temporal linear morphea, osseous thinning, cortical calcifications, white matter edema, phakomatosis.

CRANIOFACIAL PHACOMATOSIS IN EARLY CHILDHOOD: IMAGING SPECTRUM OF PARRY–ROMBERG SYNDROME IN A 3-YEAR-OLD CHILD.

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ABSTRACT

Parry–Romberg Syndrome (PRS), also known as progressive hemifacial atrophy, is an uncommon neurocutaneous disorder characteristically involving the skin and subcutaneous connective tissues and may later progress to affect the underlying musculature, cartilage, and osseous structures resulting into unilateral atrophy of the same with or without development of neurologic symptoms. PRS typically presents initially in children and young adults and slowly progresses over a highly variable course ranging from 2 to 20 years and radiologic evaluation plays a vital role in diagnosis and assessment of disease extent. We report a case of a 3-year-old female presenting with right-sided facial hemiatrophy, cutaneous tightening, alopecia, eyebrow hair loss and fronto-temporal linear morphea. CT and MRI demonstrated characteristic craniofacial involvement with soft-tissue atrophy, osseous thinning, and intracranial cortical calcifications along with white matter edema. Imaging features strongly favoured the diagnosis of craniofacial phakomatosis consistent with Parry-Romberg Syndrome.

INTRODUCTION

Parry–Romberg Syndrome (PRS) is a rare, progressive neurocutaneous disorder typically presenting during the first two decades of life.[1] Initially described independently by Caleb Hillier Parry in 1825 and Moritz Heinrich Romberg in 1846,[2] Parry–Romberg syndrome predominantly involves children and young adults, with a reported prevalence of approximately 1 per 700,000 individuals.[3] It manifests as progressive hemifacial wasting and atrophy of the skin, subcutaneous tissue, muscles and bone. Neurological associations—including white matter changes, calcifications, and seizures—have been well described. Linear scleroderma "en coup de sabre" may coexist, suggesting an overlapping spectrum of localized scleroderma and craniofacial phacomatoses.[1] Early-onset PRS, particularly in children below 5 years, is uncommon and poses challenges in diagnosis and differentiation from morphea and other neurocutaneous syndromes. Multimodality imaging is crucial for early recognition of PRS, documenting craniofacial and intracranial involvement, and guiding long-term management and follow-up.[4]

CASE REPORT

A 3-year-old female child presented with progressive right-sided facial hemiatrophy and skin tightness for one year. The child was developmentally normal with an uneventful full-term birth history and no antecedent trauma or neurological symptoms. Clinical examination revealed fronto-temporal linear morphea, eyebrow hair loss over the affected side with localized alopecia (Figure 1A & 1B). Past and recent clinical photographs demonstrated progression of facial contour asymmetry (Figure 2A & 2B).

Figure 1 (A) & (B). Clinical photograph (A) & (B) showing right-sided facial hemiatrophy, alopecia, eyebrow hair loss with skin tightening.

Past photograph - 1 year back. Recent photograph.

Figure 2 (A) & (B). Image (A) showing past photograph 1 year back and image (B) showing recent photograph demonstrating increased contour asymmetry, temporal alopecia and eyebrow hair loss compared to earlier image.

CT and MRI of the brain and face were performed for further evaluation.

IMAGING FINDINGS

Soft Tissue & Glandular Changes (Figure 3A - 3B & Figure 4A - 4B).

- Asymmetrical atrophy of the right parotid and submandibular glands.
- Atrophy of right-sided masticator muscles and facial muscles
- Asymmetric reduction of subcutaneous facial fat on affected side.
- Marginal reduction of retro-orbital fat on the right side.

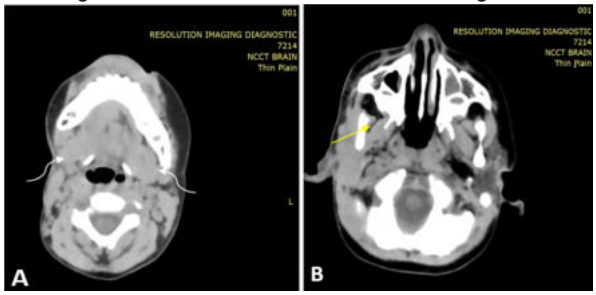

Figure 3 (A) & (B). Axial CT image (A) showing atrophy of

right submandibular gland as compared to other side (white curved arrows comparing both submandibular glands) and Axial CT image (B) showing atrophy of muscles of mastication (yellow arrow showing atrophy of right sided superficial temporalis and lateral pterygoid muscle), with reduced facial soft-tissue thickness and atrophy of right parotid gland as compared to opposite side.

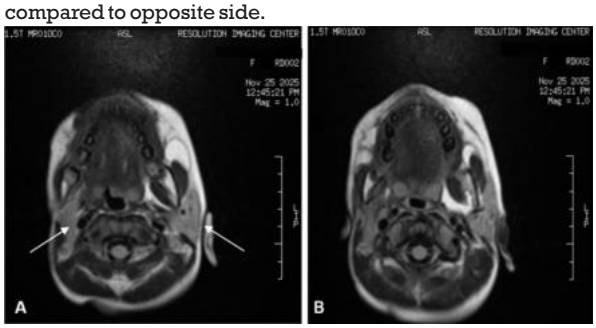

Figure 4 (A) & (B). MRI T2 axial images (A) & (B) showing reduced bulk of right parotid gland (white arrow in image A), right masticator muscles, skin-subcutaneous tissue as compared to opposite side.

Osseous Findings (Figure 5A & 5B).
Mild thinning of:

- Right frontal bone,
- Lower right parietal bone,
- Right pterygoid plates
- Right zygomatic bone,
- Right half of middle cranial fossa
- Right ramus and condylar process of the mandible

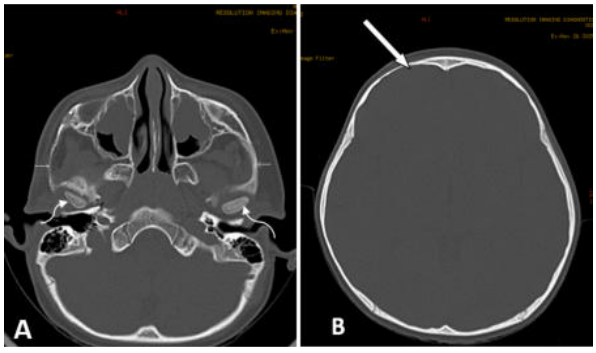

Figure 5 (A) & (B). Axial bone window CT image (A) showing thinning of right zygomatic arch (white arrows) and condylar process of mandible on right side (curved white arrow) and Axial bone window CT image (B) showing thinning of right frontal bone (white arrow head).

STIR hyperintense marrow edema in the lateral wall of the right maxillary sinus and right zygoma with minimal adjacent peri-articular soft-tissue edema. (Figure 6).

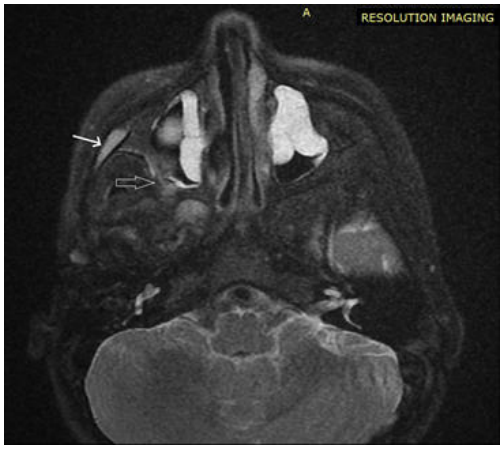

Figure 6. STIR hyperintense marrow edema in the right maxillary sinus wall (white arrow head) and zygoma (white arrow) with minimal surrounding edema.

Intracranial Findings

- **Multiple tiny cortical calcifications** in **right supra-sylvian frontal lobe** (Figure 7).
- **Adjacent FLAIR hyperintense white matter edema**, without mass effect (Figure 8).

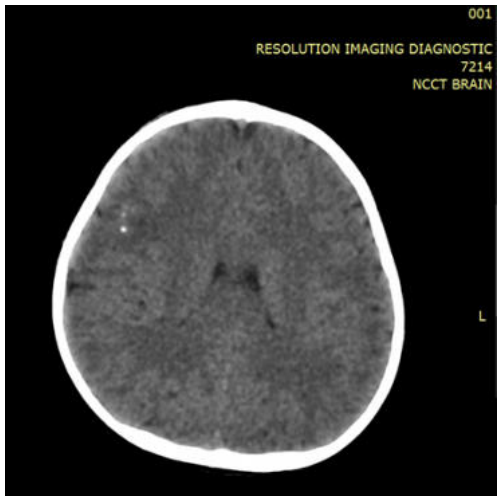

Figure 7. Tiny cortical calcifications in the right supra-sylvian frontal lobe.

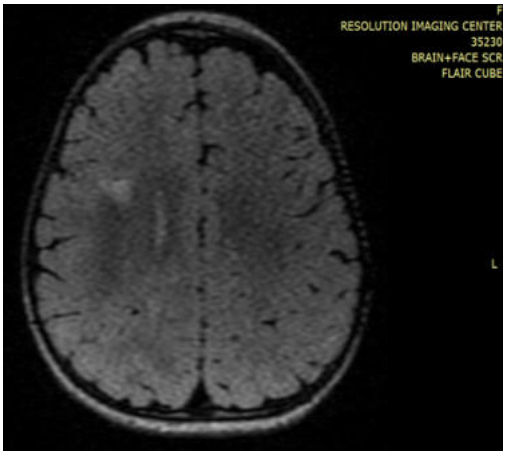

Figure 8. Adjacent FLAIR hyperintense white-matter edema in the right frontal lobe.

DISCUSSION

Parry Romberg syndrome (PRS) is an uncommon sporadic condition more common in females with no ethnic or geographic predilection.[1] PRS represents a spectrum of neurocutaneous disorders involving progressive hemifacial atrophy, often overlapping clinically and radiologically with linear scleroderma. Evidence from laboratory and histologic studies most strongly favors an inflammatory autoimmune etiology, which may occur with associated vasculopathy.[5] The effectiveness of immunosuppressive treatment during periods of active disease provides additional confirmation of an underlying immunologic mechanism.[6] The disease is typically slowly progressive and may plateau after several years. Diagnosis of PRS mainly relies on the clinical history and examination and exclusion of other possibilities, supported by imaging findings.[7] Neurologic symptoms may occur in 15%–20% of patients commonest being ipsilateral headaches, facial pain, and seizures, which may be refractory to treatment.[8]

Intracranial involvement in PRS has been variably reported, with cortical calcifications, white matter hyperintensities / edema, vascular abnormalities, and ipsilateral

leptomeningeal enhancement described in a subset of patients.[9,10] These neurological features correlate with proposed autoimmune, inflammatory, and neuro-vasculitic etiologies.[6]

In our case, the diagnosis of Parry–Romberg Syndrome was supported by a strong correlation between clinical and imaging findings. Clinically, the child presented with progressive unilateral facial hemiatrophy, skin tightening, and fronto-temporal linear morphea with ipsilateral eyebrow hair loss and alopecia, all of which are well-recognized features within the PRS spectrum. Serial clinical photographs demonstrated progressive facial asymmetry, reinforcing the evolving nature of the disease.

Imaging findings closely paralleled the clinical presentation and revealed more extensive involvement than was clinically evident. CT and MRI demonstrated asymmetric atrophy of the right parotid and submandibular glands, along with reduced bulk of the right facial, masticator, and intrinsic tongue muscles, correlating with visible soft-tissue volume loss. Loss of subcutaneous fat and marginal reduction of retro-orbital fat further explained the facial contour deformity.

Osseous changes, including thinning of the right frontal, parietal, zygomatic, maxillary, and mandibular bones, reflected chronic involvement and are characteristic of PRS. The presence of STIR hyperintense marrow oedema in the right maxillary sinus wall and zygoma suggested ongoing inflammatory activity, supporting active disease. Additionally, intracranial involvement in the form of tiny cortical calcifications with adjacent T2/FLAIR white-matter oedema highlighted subclinical neurological involvement, a recognized but variable association in PRS.

The combined clinical and radiological features strongly favored a diagnosis of craniofacial phacomatosis consistent with Parry–Romberg Syndrome.

PRS shares overlapping clinical features with *en coup de sabre*, including similar age of onset, neurologic involvement, and cutaneous findings. *En coup de sabre* (ECDS) demonstrates localized linear cutaneous and subcutaneous sclerosis along the frontoparietal scalp, with imaging findings typically limited to superficial soft-tissue thickening and focal calvarial changes.[11,12] Diffuse facial atrophy and significant intracranial involvement are more characteristic of PRS, whereas ECDS remains primarily superficial and localized.[13,14]

Parry–Romberg syndrome and Rasmussen encephalitis share overlapping clinical and imaging features, including unilateral hemispheric T2 hyperintensity and cerebral atrophy.[15] Reported cases of their coexistence suggest a possible common pathophysiologic mechanism. Differentiation relies on clinical features, particularly epilepsia partialis continua in Rasmussen encephalitis and cutaneous involvement in PRS.[15,16]

Facial asymmetry may also be seen in hemifacial microsomia and Goldenhar syndrome, which are congenital and non-progressive, unlike PRS.[13,17] Hemifacial hyperplasia results from tissue overgrowth rather than atrophy.[13,18] Partial lipodystrophy may mimic PRS but is typically bilateral.[17] Silent sinus syndrome usually presents later in life with characteristic maxillary sinus opacification, atelectasis, and ipsilateral ostiomeatal obstruction.[19]

Key imaging findings in our case-including unilateral glandular atrophy, muscle wasting, atrophy of skin-subcutaneous tissue, osseous thinning, and intracranial cortical calcifications with white matter signal abnormality-are characteristic of PRS and support the diagnosis of craniofacial phakomatosis. Recognition of early osseous changes is crucial, as they are frequently under-appreciated in pediatric patients. Imaging provides essential guidance for disease staging, therapeutic planning, and monitoring progression.

Limitation

Limitations include the single-case design with limited generalizability, restricted follow-up duration, absence of histopathological confirmation, and lack of advanced functional or genetic evaluation. The retrospective literature review may also be subject to selection bias.

CONCLUSION

This case underscores the role of clinical–radiological correlation in the diagnosis of Parry–Romberg syndrome in a very young child. Multisystem radiologic involvement-including soft tissue atrophy, osseous thinning, and intracranial calcifications with white matter edema-confirms the characteristic pattern of craniofacial phakomatosis. Early, accurate identification supports timely initiation of immunomodulatory therapy and long-term follow-up to monitor progression.

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