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"The Unkindest Cut of All": Parry-Romberg Syndrome - An Unwonted Affair

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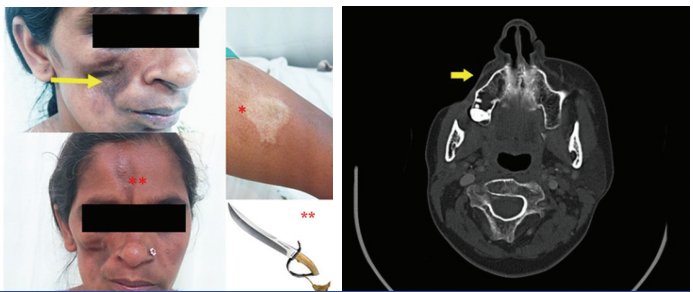
A 45-year-old middle-aged female was referred to our department as a clinically diagnosed case of Parry-Romberg Syndrome for assessment of the craniofacial manifestations. She presented with gradually worsening right facial asymmetry over the past 4-5 years with otherwise non-contributory medical and family history. A physical examination revealed marked right-sided hypoplasia primarily involving the maxillary region, with obvious and marked wasting of skin and loss of subcutaneous fat and resultant "sunken cheek appearance" and ipsilateral deviation of mouth and nose. The " en coup de sabre " lesion (resembling a wound as if "struck with a sword" as it follows the Blaschko's lines and is in fact the differentiation between the healthy and atrophic regions) with associated hyperpigmentation was seen over the right malar and forehead regions [Table/Fig-1]. History of associated migraine, nausea and vomiting was present. There was also seen hypo –to- depigmented lesions over the contralateral forearm and trunk. However there was no associated facial pain, focal seizures, cognitive impairment, hemiparesis, ocular or oral features. Neurological examination showed no deficits and limbs were symmetric.

On imaging our case demonstrated the typical findings of volume loss restricted to right side of the face attributable to significant generalized atrophy of mid-face soft tissues particularly the buccomasseteric region [Table/Fig-2] and poorly developed/ atrophic right buccal space with hypotrophic right buccinator and zygomaticus muscles and nearly deficient buccal space fat [Table/Fig-3a&b]. There were also seen hypotrophic right masseter and right temporalis muscles in the masticator space [Table/Fig-4a-d] as well as an atrophic right parotid gland and non-visualised ipsilateral parotid duct [Table/Fig-5a&b]. However the laryngeal architecture and cartilaginous framework was largely preserved as were bilateral submandibular glands. There were no associated atrophy of the right hemitongue [Table/Fig-4]. No accessory parotid tissue was seen in relation to masseter muscles bilaterally [Table/Fig-4]. On bone window settings; though the orbits, mandible and maxilla were symmetrical bilaterally there was evidence of thinning of the zygoma and the zygomatic arch on the right side [Table/Fig-6]. There was also evidence of extension of these atrophic changes onto the forehead region [Table/Fig-7].



[Table/Fig-1]: Marked Right-sided Mid-Face Hypoplasia • 'Sunken Cheek Appearance' (yellow arrow) • Hypo-to-depigmented lesions over left forearm (red single asterisk*) • 'en coup de sabre' lesion (red double asterisk**).

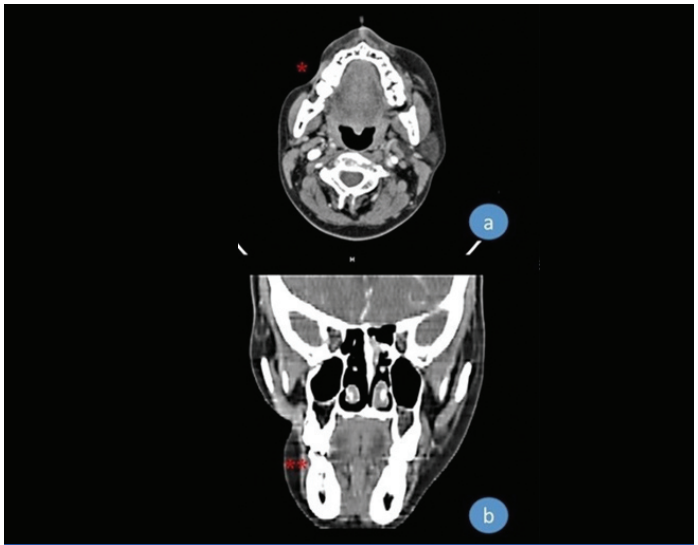
[Table/Fig-2]: The Image at this level mirrors the clinical profile of the patient. • Significant generalized atrophy of soft tissues of right mid-face especially in the buccomasseteric region (yellow arrow) • Gross asymmetry of facial soft tissues with volume loss restricted to the right side with resultant "sunken cheek appearance"(yellow arrow) However there is no extension across the midline.

DISCUSSION

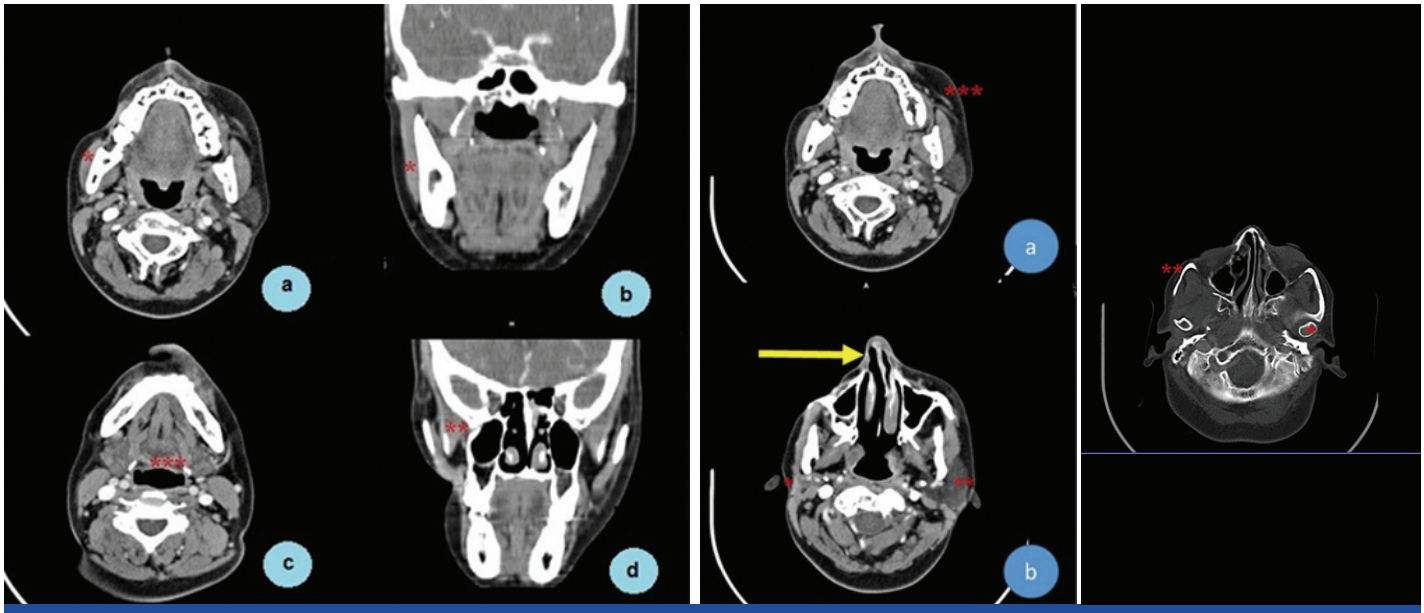
Despite riveting advances in molecular analysis and genetics, Parry-Romberg Syndrome remains the most perplexing of all craniofacial afflictions. PRS aka hemifacial progressive atrophy is a rare degenerative neurocutaneous syndrome of ambiguous etiopathogenesis. An estimated 1 in every 700,000 births present with this syndrome, as per an internet- based research carried out by Romberg's Connection [1,2].

Sporadic in occurrence it has a peculiar indolent albeit self-limiting course with no known Mendelian inheritance. Characterised by slowly progressive, usually unilateral atrophy, it most commonly affects the facial tissues involving the cutaneous, subcutaneous and skeletal components. It has been reported to be more common in females [2-5] without apparent geographic or ethnic predilection [3,4].

It typically presents initially in children and young adults and slowly progresses over a highly variable course ranging from 2 to 20 years, eventually reaching a "burned-out phase" and stabilizing abruptly for no apparent reason [3,4]. This peculiar disease course, along with multifaceted signs and symptoms viz. ocular, oto-rhino-laryngological, orthodontic, neurologic etc., impedes consistent understanding of the underlying pathophysiology of PRS. Of the assorted rationale hypothesized over the years; such as infection, trauma, radiation exposure, embryonic developmental dysfunction, sympathetic cervical ganglion dysfunction, vascular abnormalities,



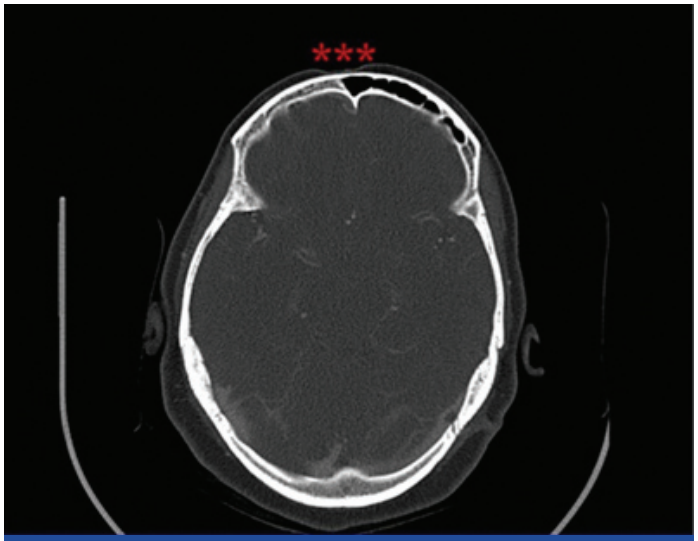
[Table/Fig-3a & b]: Buccal Space Level. • The right side is poorly developed as compared to the left side with nearly deficient skin, subcutaneous fat & buccal space fat(* red single asterisk) • Hypotrophied right buccinator muscle with nearly atrophic right zygomaticus muscle (** red double asterisk).



[Table/Fig-4(a-d)]: Masticator space level. • Hypotrophic right masseter (* red single asterisk) and right temporalis muscles (** red double asterisk) as compared to their counterparts on the left. • No Evidence of accessory parotid gland / tissue is seen in relation to the masseter muscles bilaterally • Bilaterally symmetrical genioglossus. geniohyoid, mylohyoid & hyoglossus muscles (***red triple asterisk)

[Table/Fig-5(a-b)]: Parotid glands & ducts assessment. • The parotid duct is seen piercing the left buccinator muscle (***red triple asterisk) whereas the same is not visualised on the right side • The right parotid gland is not visualised in its expected anatomic location (*red single asterisk) whereas the left parotid gland is normally visualised (**red double asterisk) Note is also made of the deviation of the nose towards the right side in this image (Yellow Arrow).

[Table/Fig-6]: Findings on bone window settings. • There is not seen any significant asymmetry between the bilateral mandibular condyles(*red single asterisk) • However there is seen thinning of the zygoma & the zygomatic arch on the right side (**red double asterisk).



[Table/Fig-7]: Findings on bone window settings.
There is seen evidence of cranial extension of the skin & subcutaneous tissues deficiency onto the right forehead region as well (***red triple asterisk).

inflammatory conditions; metabolic, endocrine and autoimmune disorders; none satisfactorily characterises and predicts PRS in isolation [2,3].

As regards its differentials; restricted lipodystrophy besides being bilateral affects only the fatty layer while generalized lipodystrophy compromises all fatty tissues of the mesentry, omentum etc [1]. Hemifacial microsomia being a more complex craniofacial syndrome presents with varying degrees of regional hypoplasia besides having a male predilection [1]. Seckel syndrome, chromosome 13q syndrome, hemifacial hypertrophy etc. are some others which need exclusion [1].

With no definitive curative algorithms; aesthetics and functionality entailing restoration of facial morphometric symmetry and dynamics are the sine qua non of management. Imaging evaluation aids by assessing extent and progression of disease, excluding differentials and monitoring post treatment response.

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