Congenital haemifacial hyperplasia

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DESCRIPTION

A 19-year-old man presented with gradually progressive facial asymmetry since birth with difficulty in chewing food and the right-sided hearing loss. On examination, there was asymmetric enlargement of the right side of the face, including the right lower lip and pinna with multiple nodules over dorsum of the right side of the tongue. The overlying skin on the right side of the face showed the areas of dark pigmentation with no ulceration or palpable thrill. The left side of the face, vitals and rest of physical examination were normal. There was no family history of similar complaints.

MRI showed increased bulk with fatty change in muscles of mastication on the right side with increased subcutaneous fat. Fatty enlargement of the right side of the tongue and increased bulk of the right parotid gland were also seen in addition to asymmetric dentition (figure 1A–C). Bony hypertrophy was seen in underlying hemimandible, maxilla and frontal bone on the right side (figure 1C,D). No underlying vascular malformation or cerebral asymmetry was seen. A diagnosis of congenital haemifacial hyperplasia with lipomatosis was made. The patient is now scheduled for a multistep surgery (planned first phase is Le fort I and sagittal split ramus osteotomy with possible inferior body osteotomy followed by recontouring of malar prominence and facial debulking).

Congenital haemifacial hyperplasia is a rare developmental disorder characterised by unilateral enlargement of both hard (bones and teeth) and soft tissues of face since birth.1 It is commoner in women and usually affects the right side of the face.2 3 Histologically, the increased number of cells rather than the increased cell size is seen in all the involved tissues.1 It can be further classified into true haemifacial hyperplasia (having unilateral enlargement of viscerocranium, bounded superiorly by frontal bone (not including the eye), inferiorly by inferior border of mandible, medially by the midline and laterally by the ear, including pinna) with enlargement of all tissues (teeth, muscle, bone and soft tissues) and partial haemifacial hyperplasia when all structures are not enlarged to the same degree or abnormal growth is limited to a single structure.3 4 Proposed aetiological factors include hormonal imbalances, neural disorders, incomplete twinning, abnormal intrauterine environment, somatic mutations, mechanical influences, vascular conditions and congenital syphilis.3

Figure 1  (A) axial T2-weighted and (B) T2 fat-suppressed images show increased bulk with fatty change in muscles of mastication on the right side with increased subcutaneous fat. Fatty enlargement of the right side of the tongue (asterisk) and increased bulk of the right parotid gland are also seen. Teeth are also bigger on the right side. (C, D) Coronal T1-weighted images show bony hypertrophy of the frontal bone (white arrow in C) and mandible (white arrow in D) on the right side. No cerebral asymmetry is seen.


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Associated abnormalities include thickened skin and hair, excessive secretion of sebaceous and sweat glands, telangiectasias and pigmentary defects on the affected side.4

Isolated haemifacial hyperplasia is a diagnosis of exclusion. Differentials include other causes of facial asymmetry which include fibro-osseous lesions such as Paget’s disease, fibrous dysplasia, dyschondroplasias, vascular malformations and malignant conditions (osteosarcomas, chondrosarcomas). These can be differentiated by the presence of foraminal enlargement in haemifacial hyperplasia and characteristic clinic-radiological features in rest of the entities.4 Other malformation syndromes, such as neurofibromatosis, Proteus syndrome, Beckwith-Wiedemann syndrome, Epidermal nevus syndrome, Russell-Silver syndrome and Klippel-Trenaunay-Weber syndrome are usually bilateral affecting multiple body parts. Apart from that, unilateral distribution of dental anomalies and concurrent ipsilateral tongue enlargement are unique to haemifacial hyperplasia.4 5

Treatment involves integrated multidisciplinary approach and includes soft tissue debulking, reconstructive bony procedures and orthodontic treatment.2 It is indicated mainly for cosmetic purposes and ideally deferred until physiological growth ceases.1

Learning points

► True haemifacial hyperplasia is a rare developmental disorder having unilateral enlargement of hard and soft tissues of the face due to the increased number of cells in the involved tissues.
► It is a diagnosis of exclusion and can be differentiated from other malformation syndromes (such as neurofibromatosis, Proteus syndrome, Beckwith-Wiedemann syndrome, Epidermal nevus syndrome, Russell-Silver syndrome and Klippel-Trenaunay-Weber syndrome) by unilateral distribution of dental anomalies and concurrent ipsilateral tongue enlargement which are unique to haemifacial hyperplasia.

REFERENCES