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The importance of a differential diagnosis between true hemifacial microsomia and pseudo-hemifacial microsomia in the post-surgical long-term prognosis

Maria C. Meazzini [a,*], Roberto Brusati [a], Patrik Diner [b], Ennio Giannì [c], Faustina Lalatta [d], Alice S. Magri [e], Arnaud Picard [b], Enrico Sesenna [e]

[a] Department of Maxillofacial Surgery, Cleft lip and palate Center, University of Milano, Italy
[b] Department of Plastic Surgery, Trousseau Hospital, Paris, France
[c] Department of Orthodontics, Dental School, University of Milano, Italy
[d] Medical Genetic Unit, Fondazione IRCCS Foundation, Cà Granda Hospital Maggiore Policlinico, Italy
[e] Department of ENT–Dental–Ophtalmologic and Cervico-Facial Sciences, Maxillofacial Surgery Unit, University of Parma, Italy

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ABSTRACT

Long-term results after surgical treatment of the mandibular asymmetry in growing children with hemifacial microsomia (HFM), whether with osteotomies or distraction osteogenesis, have mostly shown a tendency towards the recurrence of the asymmetry. In contrast, in the literature we find sporadic case reports where the long-term post-surgical follow-up of patients diagnosed as HFM, are surprisingly stable. All these reports refer to patients who have substantially no soft tissue involvement, but only severe mandibular ramus and condyle deformities. The phenotypes of these cases are unexpectedly similar. The authors suggest, that it is possible that all of these cases might be isolated hemimandibular hypoplasias, misdiagnosed as HFM, which present a normal functional matrix and, therefore, tend to grow towards the original symmetry. Differential diagnosis between true HFM and this HFM-like isolated hemimandibular hypoplasia (pseudo-HFM) is of great importance given the very different prognosis and it is possible through the collaboration between not only surgeons and orthodontists, but also of geneticists and dysmorphologists.

1. Introduction

Aetiologic diagnosis is possibly the most difficult, but also the most important step in the treatment of facial deformities. Hemifacial microsomia (HFM) is a relatively common craniofacial anomaly with a birth prevalence of at least 1/5600, characterized by the asymmetric underdevelopment of structures originating from the Ist & IInd branchial arches. Deformities may involve the ear and the mandible, as well as the maxilla, the zygomatic arch, the temporal bone, the V & VII cranial nerves, the cervical spine, and facial muscles. In patients affected by HFM the face may be strikingly asymmetrical because of the hypoplastic changes in the mandible and the dysplastic changes and displacement of the ear. The degree of ear involvement is markedly variable and cleft lip or cleft palate may be associated (Jones, 2005).

A condition analogous to HFM has been induced in the mouse by causing a local haemorrhage from the embryonic stapedial artery during the 30th–40th day of foetal development, a critical period of neural crest cell migration (Poswillo, 1974). Although the haemorrhage model could account for much of the observed variability, embryological research has increased significantly our knowledge on HFM.

It is known that HFM is aetiologically heterogeneous. Many chromosome abnormalities have been recorded, but also environmental causes which include thalidomide, primidione and retinoic acid administered during the organogenesis. The phenotype has also been noted in infants born to diabetic mothers. A recent model, based on a mutation of a locus on chromosome 10, appears to support the hypothesis that HFM anomalies have partly a genetic causation (Cousley et al., 2002; Dabir and Morrison, 2006).

So far, evidence for genetic involvement, include family history of HFM phenotype and rare familial cases that exhibit autosomal dominant inheritance (Robinow et al., 1986; Brady et al., 2002).

From the embryological point of view, HFM is one of four conditions defined as otofacial malformations, which are Neurocristopathies, which share a major involvement of neural crest cells,

* Corresponding author. Via Appiani n.7 20121 Milano, Italy. Tel./fax: +39 02 29010303.
E-mail address: cmeazzini@yahoo.it (M.C. Meazzini).

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Table 1
Differential diagnosis chart to help distinguish between true hemifacial microsomia and pseudo-HFM (misdiagnosed as HFM).

	HFM	Pseudo-HFM
Clinical history	Mostly diagnosed at birth	Not diagnosed at birth Seldom history of trauma
Clinical examination	Soft tissue defects (may be very mild): Ear defects, pre-auricular tags Possible facial nerve involvement Masseter muscle hypoplasia Deviation of the chin on the affected side, associated with flatness on the affected cheek Mild deviation to the affected side during opening	No soft tissue defects: Normal ears, no pre-auricular tags No nerve deficit Well-developed masseter Deviation of the chin on the affected side, associated with fullness on the affected cheek Significant deviation to the affected side during opening
Panoramic X-ray (or CT)	Hypoplasia of the ramus and condyle and coronoid processes up to absence of the condyle and temporal fossa	Hypoplasia of the ramus and condyle and coronoid processes which are typically collapsed one on the other. There is a typically V-shaped sigmoid notch. The temporal fossa is always present

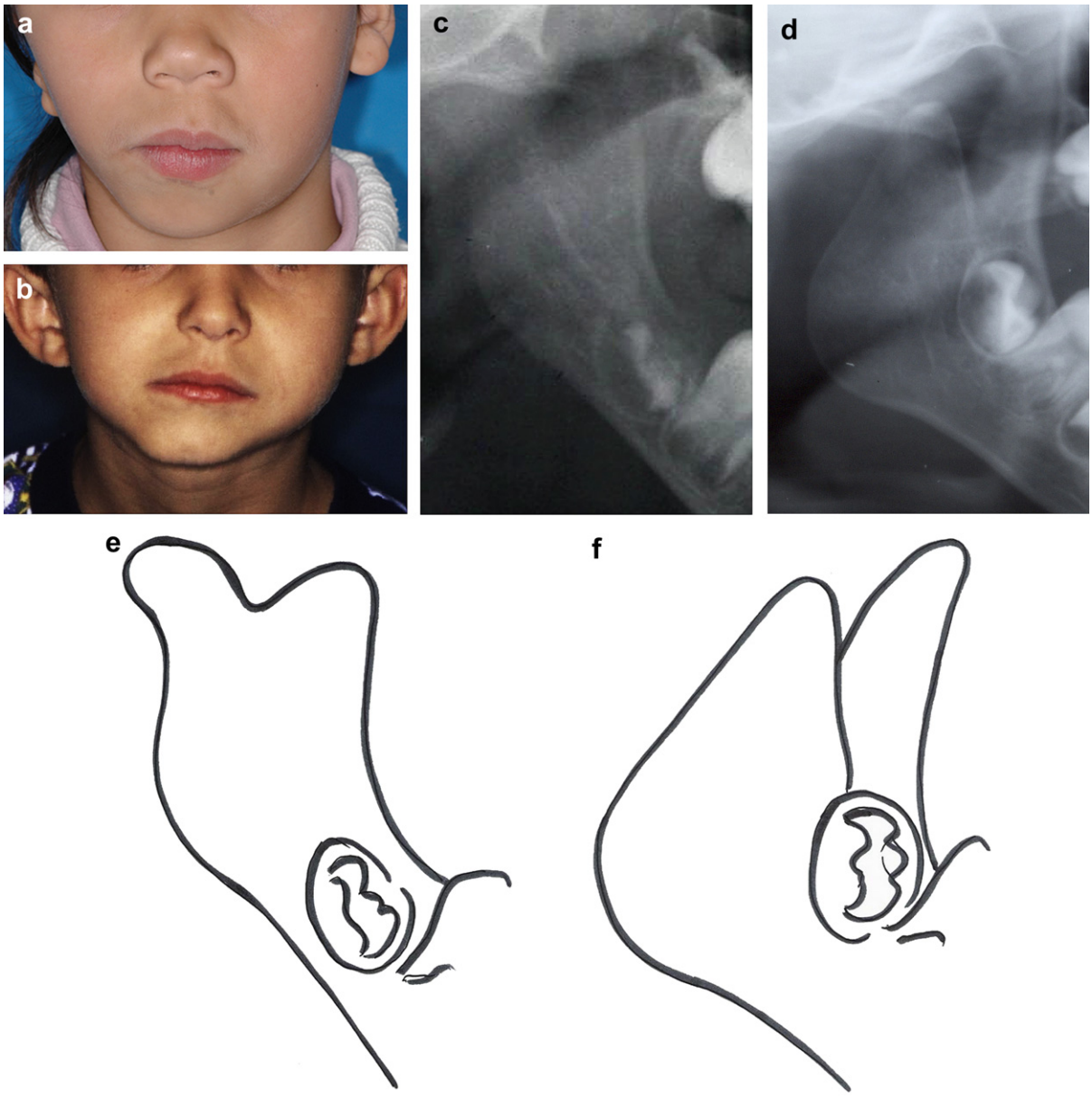

Fig. 1. a: Type II Pruzansky HFM ramal deformity. b: Misdiagnosed ramal deformity. c: Tracing of a Type II Pruzansky HFM ramal deformity. d: Tracing of a misdiagnosed Type II Pruzansky HFM ramal deformity. Note the typical shape, visible in all cases, where the condyle is short and collapsed against the coronoid process. The sigmoid notch is very deep. e: Mandibular facial contour in a Type II Pruzansky HFM ramal deformity. Note the transverse hypoplasia of the gonial area as well as vertical on the affected side. f: Mandibular facial contour in misdiagnosed Type II Pruzansky HFM ramal deformity. Note the fullness of the cheek on the affected side.