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Case report

Partial craniofacial duplication: A review of the literature and case report

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ABSTRACT

Diprosopus (Greek; di-, "two" + prosopon, "face"), or craniofacial duplication, is a rare craniofacial anomaly referring to the complete duplication of facial structures. Partial craniofacial duplication describes a broad spectrum of congenital anomalies, including duplications of the oral cavity. This paper describes a 15 month-old female with a duplicated oral cavity, mandible, and maxilla. A Tessier type 7 cleft, midline meningocele, and duplicated hypophysis were also present. The preoperative evaluation, surgical approach, postoperative results, and a review of the literature are presented. The surgical approach was designed to preserve facial nerve innervation to the reconstructed cheek and mouth. The duplicated mandible and maxilla were excised and the remaining left maxilla was bone grafted. Soft tissue repair included closure of the Tessier type VII cleft. Craniofacial duplication remains a rare entity that is more common in females. The pathophysiology remains incompletely characterized, but is postulated to be due to duplication of the notochord, as well as duplication of mandibular growth centres. While diprosopus is a severe deformity often associated with anencephaly, patients with partial duplication typically benefit from surgical treatment. Managing craniofacial duplication requires a detailed preoperative evaluation as well as a comprehensive, staged treatment plan. Long-term follow up is needed appropriately to address ongoing craniofacial deformity.

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1. Introduction

Diprosopus (Greek; di-, "two" + prosopon, "face"), or craniofacial duplication, is a rare craniofacial anomaly. While true diprosopus refers to the complete duplication of facial structures with two faces on a single head, partial craniofacial duplication describes a broad spectrum of deformities and malformations. Diprosopus appears to have been first depicted between 1200 and 700 BC (Bendersky, 2000) (Fig. 1). Among the "Pretty Ladies of Tlatilco" ancient terracotta sculptures originating in the region of what is now Mexico City, are a few dozen bifacial and bicephalic figurines with various degrees of duplication. Later medical illustrations and descriptions in the 16th and 17th centuries (Ambroise, 1982) are followed by the first case report in the modern literature (Mclaughlin, 1949).

The duplication may involve the face, components of the cranium, or a combination of both. The mildest form of cerebral involvement is duplication of the pituitary gland (Shah et al., 1997; Burke et al., 2000; Shroff et al., 2003; De Penna et al., 2005; Akin et al., 2011), while nasal duplication represents the mildest form of facial involvement (Ghosh et al., 1971; Obwegeser et al., 1978; Barr, 1982; Maruotti et al., 2009). Most cases occur in isolation, although there are four reports among twins (Rai et al., 1998). In these cases, one twin is normal and the other manifests the duplicated state (Changaris and McGavran, 1976; Verdi et al., 1991; Rai et al., 1998; Al Muti Zaitoun et al., 1999). In partial craniofacial duplications, most of these cases involved maxilla or mandible (Wu et al., 2002) and concurrent duplication is very rare. Here, we

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Fig. 1. Female sculpture with the diprosopus anomaly illustrating synophthalmia. 1200–900 BC, Tlatilco (1). Photo: Copyright© 1986 Peabody Museum of Natural History, Yale Univ., New Haven, CT.

describe a patient with duplication of the maxilla, mandible, and the oral cavity.

2. Case Report

2.1. Birth history

A 15 month-old female was born via uncomplicated, full term spontaneous vaginal delivery in Cairo to a healthy 28-year-old mother and healthy 30-year-old father. She was noted to have a left sided duplication of the mouth, mandible, and maxilla, as well as the appearance of a Tessier 7 cleft. At age 15 months, she was referred to the Children's Hospital Los Angeles (CHLA) for further care by the teamwork of three nonprofit organizations: Mending Kids, Operation Smile, and Children of War.

She was born to a mother who had no exposures to medications, drugs, alcohol, or radiation therapy. No prenatal ultrasound screening had been performed, nor had serologic testing for alpha-fetoprotein. She was discharged home to her parents after 7 days due to difficulties feeding and introduction to a special nipple. Family history of both parents was negative for consanguinity or any similar congenital craniofacial anomalies. The patient had an older brother, who was non-dysmporphic, as were both parents.

2.2. Physical examination

Physical examination revealed significant facial asymmetry (Fig. 2). The upper third of the face was notable for vertical orbital dystopia, with the left orbit apparently cephalically displaced by the accessory oral structures. There was strabismus but no proptosis. The left middle third demonstrated increased fullness and a second accessory oral cavity, which was located at the cheek lateral to the alar base, causing supero-lateral displacement of the ala. The accessory cavity contained teeth and produced saliva but did not contain a tongue.

The lower third of the face demonstrated an abnormal midline mouth. There was the near complete obliteration of the normal architecture on the left side, with no oral commissure or modiolus, and distorted, hypoplastic upper and lower lips lacking vermillion. There was an excess of soft tissue corresponding to a Tessier type clefting pattern, extending laterally from the midline oral cavity towards the ear, with invagination of the soft-tissues into the oral cavity. Tufts of hair were located within the segment of soft tissue excess. Intraoral examination revealed the presence of the maxillary and mandibular teeth, a hypoplastic left buccal sulcus, and a cleft of the soft palate extending into the hard palate. There was no alveolar cleft, though significant hypoplasia of the left maxilla was seen as it met the anomalous maxillary segment. The presence of the maxillary and mandibular dentition was suggested by the orientation of the teeth.

Near normal facial innervation was seen on the left side. The duplicated mouth demonstrated movements synchronous with those of the midline mouth. When smiling, a strong nasolabial fold was present adjacent to the anomalous oral cavity. Depression in the area of the commissure was lacking, however. Overall, she could animate well from the displaced segment, though with asymmetry compared to the right side, which had normal motor innervation. Cranial examination revealed a 3×2 cm soft tissue swelling at the midline vertex. The mass appeared fluid filled on palpation, and there was an associated bony defect, consistent with a meningocele.

At age 15 months, she had delayed developmental milestones, with difficulty on attempted sitting without support and using two single words in her speech. She was able to smile towards her parents and track objects. She was bottle-fed through the midline oral cavity with occasional nasal regurgitation from the midline but not the accessory mouth.

She was able to tolerate both solids and liquids. She appeared moderately malnourished, with a low weight for length at 8.1 kg for 82 cm, the 50th percentile for an 8-month-old female. Her head circumference was in the 25th percentile at 44.7 cm. She was at the 95^{th} percentile for height and less than 5th percentile for weight.

2.3. Laboratory testing and imaging

Laboratory evaluation showed normal values for haemoglobin, platelets, white blood cell counts, coagulation parameters, and hepatic and renal function tests. Adrenal function testing was normal. Chromosomal evaluation from Egypt revealed a normal 46XX karyotype. Chromosomal microarray analysis (CMA; Affymetrix SNP 6.0 array) of DNA isolates from venous blood, saliva and extracted cheek tissue was used to screen for copy number variations or subtle signs of somatic mosaicism. No genetic differences were detected. Echocardiogram at 12 days of age revealed an *ostium secundum* atrioseptal defect at 0.3, partially closing by atrial septal aneurysm with a restrictive left to right flow.

Computed tomography (CT) scanning performed at 3 days of age in Cairo revealed multiple bony deformities. The left orbital floor was superiorly displaced; the orbit was shorter and wider compared to the right side. There appeared to be a duplicated maxilla and mandible with a significant disruption of adjacent structures. There were accessory upper and lower alveoli with cephalic displacement of the maxillary alveolar ridge. Sinonasal abnormalities included absence of the left maxillary sinus, turbinates, choanal atresia, and near absence of ethmoid air cells. The left nasal bone was absent, the septum distorted, and the nasal cavity and left hemi-maxilla were nearly obliterated by the duplicated segments.

The rudimentary accessory mandible and temporo-mandibular joint possessed a Pruzansky type IIB hypoplasia with a small ramus, coronoid process, and body fused to the maxillary alveolar