

Unilateral Cranio-Facial Cleft with an Open Encephalocele

MANTRARATAM PRAMILA PADMINI, BHATTAM NARASINGA RAO

ABSTRACT

Foetal craniofacial development is a complex series of events that occurs between the third and eighth weeks of gestation. Facial clefts are deformations of the face and/or cranium caused by relative excesses or deficits of tissue along linear anatomic planes. The present case was a stillborn female child of a 31-year-old mother by caesarian route at 36 weeks' gestation. It is a very rare case with an open encephalocele in the squamous part

of the frontal bone with a unilateral craniofacial cleft extending up to the encephalocele and probably would be the first case to be reported. Although the precise aetiology is unknown, multiple genetic and environmental factors may be involved. The environmental risk factors implicated include antenatal exposure to radiation, viral infections, metabolic abnormalities, and teratogenic compounds.

Key Words: Craniofacial, Club hand, Encephalocele, Oro-ocular, Squamous

INTRODUCTION

The development of the face and the cranium during embryogenesis is a complex and orchestrated process that involves cellular proliferation, differentiation, migration, and selective apoptosis. Facial clefts are deformations of the face and/or the cranium which are caused by relative excesses or deficits of tissue along the linear anatomic planes (Tessier P. 1976) [1], (Bradley, JP et al 2006) [2]. Although the exact incidence of facial clefts is unknown, they are estimated to affect 1.4 to 4.9 of the newborns per 100,000 live births (Tessier P. 1976) [1], (Kawamoto HK Jr 1976) [3], (Kawamoto HK Jr 1990) [4]. Although their precise aetiology is unknown, multiple genetic and environmental factors may be involved. The environmental risk factors which are implicated include the antenatal exposure to radiation, viral infections, metabolic abnormalities, and teratogenic compounds (Bradley, JP et al 2006) [2].

CASE HISTORY

The present case was a stillborn, female child of a 31-year-old mother who was delivered by the caesarean route at 36 weeks of gestation. The pre-natal history showed that the mother was not exposed to any teratogens such as X-rays, infectious diseases or drug usage. On physical examination, a facial defect, skeletal defects of the skull and limb defects were observed.

The facial defects which were observed were a medial, oro-ocular, facial cleft, extending from the right angle of the mouth to the medial cantus of the right eye. There was hypertelorism with small palpebral fissures. There were low set ears on the right side as compared to the left one [Table/Fig-1]. The left ear was malformed and it was positioned close to the left lateral occipital region [Table/Fig-2]. A large open encephalocele was present in the squamous part of the frontal bone on the right side. The parenchyma of the brain tissue which was lying outside the cranial cavity was jelly in form and skeletal defects of the upper right extremity such as radial hypoplasia and club hand were observed [Table/Fig-3].



[Table/Fig-1 & 2]



[Table/Fig-3 & 4]

RADIOLOGICAL EXAMINATION

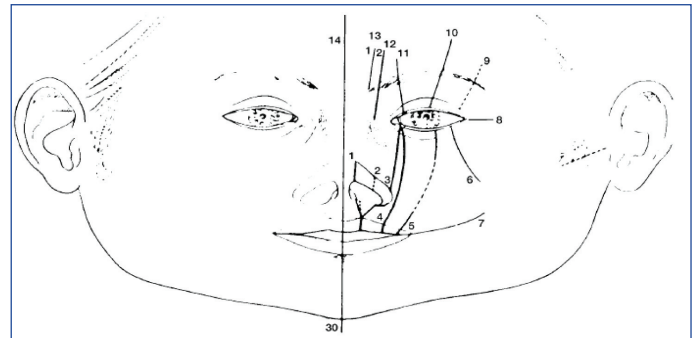
The skeletal defect was clinically and radiologically apparent in the right, lateral, maxillary alveolus which traversed the maxilla to the medial side of orbital rim, just medial to the [Table/Fig-4].

DISCUSSION

Embryology: More than one theory exists regarding the embryological pathogenesis of facial clefts. The foetal craniofacial development is a complex series of events that occur between the third and eighth week of gestation. During the initial stages, five facial processes (one frontal, two maxillary and two mandibular) form and they subsequently fuse (by the sixth week of gestation) to form the human face. The classic theory states that facial clefting occurs when the fusion process is disrupted. However, other theories propose that the pathogenesis is related to the infraction of the primordial blood vessels, amniotic bands, the failure of certain developmental zones of the face to develop completely, or to the errors in the cellular migration, penetration, and differentiation (Bradley, JP et al 2006) [2].

Although it is possible that both the oblique facial clefts and the anomalies of the extremities may have occurred spontaneously and without any association, the combination of these anomalies has importance in the explanation of their aetiological mechanisms. The morphology of these clefts can be explained on the basis of the early embryonic development of the face. If the arrest occurs before the different processes have merged at the 17-mm crown-rump length (CRL) stage, the lacrimal canal will fail to form, and a primary or transformation defect will be formed, such as primary clefts, Tessier numbers 3 and 7. If the disturbance takes place after the closure of the ectoderm of the face has been completed and a canal has been produced, before the end of the differentiation phase at the 60-mm CRL stage, a secondary or differentiation defect will result, such as secondary clefts, Tessier numbers 1, 4, 5, 6, 8, 9, 10, 13, and 14 (Van der Meulen, 1985) [5]. There are several theories on the aetiopathogenesis of the oblique facial clefts. One of these theories is that the oblique facial clefts are caused by a developmental arrest such as focal foetal dysplasia and a variety of mechanisms has been suggested, such as linear necrosis, diminished arterial supply, disturbance in the migration of the neural crest cells, or the failure of mesodermal penetration and coalescence between the facial processes (Van der Meulen, 1985) [5]. This theory seems to explain the formation of the primary clefts.

The clefts are numbered from 0 to 14 and they follow constant lines or axes through the eyebrows or the eyelid, the maxilla, the nose, and the lip. The orbit is regarded as the reference landmark, since it is common to both the cranium and the face. Clefts that are located cephalad to the palpebral fissure are directed "northbound" and are considered to be mainly "cranial" in nature. The "southbound" clefts pass caudally from the palpebral fissure to become "facial". The "craniofacial" clefts are formed by the combination of the northbound and the southbound clefts. The craniofacial clefts usually follow the same well-defined "time zones". Thus, the following combinations can be clinically observed: 1-14, 1-13, 2-12, 3-11, and 4-10. Although the craniofacial clefts tend to coincide with these "time zones" across the orbit, the vascular supply and the embryonic processes do not necessarily follow the same north-south pathways. Cleft 5 is the rarest of all the oblique facial clefts. There is a cleft lip with the fissure



[Table/Fig-5]: Tessier's classification of craniofacial clefts

just medial to the angle of the mouth, but not in the commissure (would be another type of cleft). It courses upward across the lateral cheek to reach the lower eyelid between the medial and the lateral third (http://www.cpmundi.org/adjuntos/manuales/es/rare_cranio-facial_clefts-5.pdf [6]). A unilateral upper lateral orbital cleft which fits the description of the extremely rare Tessier cleft number 9 has been reported by Al-Ani et al [7]. Consequently, the embryo pathogenesis of some of the craniofacial clefts is difficult to explain. (Henry K. Kawamoto et al [8]). This system has been internationally accepted and it allows a concise, effective communication among the clinicians [Table/Fig-5]. (Jeremy A. et al. 2003 [9])

CONCLUSION

The present case is a very rare case with an open encephalocele in the squamous part of the frontal bone, with a unilateral craniofacial cleft extending up to the encephalocele. Many of the above authors have reported craniofacial clefts, but without an encephalocele. Hence, this type of the cleft with an open encephalocele is a very rare abnormality and probably would be the first case to be reported.

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