Case Report

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Transverse Facial Cleft, Tessier number 7: Case Report of a Rare Congenital Anomaly

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KEYWORDS

Orofacial clefts, congenital macrostomia, syndromic, newborn

ABSTRACT

Transverse facial cleft (TFC), Tessier number 7, is a rare congenital anomaly commonly associated with other anomalies of the first and second branchial arch with varying severity. Risk factors such as alcohol abuse, folic acid deficiency, and ingestion of herbal concoctions in pregnancy have been implicated in this deformity. We report a case of a bilateral transverse facial cleft in a newborn and review the existing literature on the condition to increase awareness and knowledge.

INTRODUCTION

Orofacial clefts are the most common congenital malformations of the head and neck [1]. Transverse facial cleft, Tessier number 7, also referred to as congenital macrostomia, is a very rare form of facial cleft [2]. Transverse facial cleft (TFC) results from failure of fusion of mandibular and maxillary processes, due to the interruption of mesoderm migration of the first branchial arch, during the 4th - 5th week of embryonic development [3]. It seldom occurs alone and is associated with other anomalies arising from the first and second branchial arches [4]. The facial structures that form the embryonic lip fuse between the 5th - 6th week of life and the palatal shelves fuse during weeks 7-10. Various classification systems have been proposed for TFC, but the Tessier classification devised by Paul Tessier, a numerical classification founded on clinical observation, is widely accepted [2,5]. There are 15 clefts locations (numbered 0-14), with the orbit as the reference point, and these clefts were distributed according to 8 time zones. The clefts can be divided into the midline, paramedian, orbital, and lateral clefts based on their position. Clefts running through the lower eyelid are facial, and those running upwards through the upper eyelids are cranial [2]. Tessier number 7 is the most lateral facial cleft in the Tessier Classification [5]. TFC can present with various degrees of severity from slight widening of the mouth to a cleft extending up to the ears, macrostomia, lateral facial muscular diastasis, and bony anomalies of the maxilla and zygoma [6]. It accounts for less than 0.5% of all clefts [3], with a frequency estimated at 1 in 50,000 to 175,000 live births [7]. TFC is more common in males with a predilection for the left [3,8,9]. Bilateral facial clefts are rarer [10]. They can occur as part of a syndrome (Goldenhar, Dandy-Walker, PHACE, Treacher Collins, Peter Plus, Amniotic band syndromes) and rarely as an isolated entity [2].

The etiopathogenic mechanism of bilateral TFC is unknown, but it is postulated to be associated with genetic factors, uncontrolled apoptosis, damage or

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interruption of stapedius artery, the inadequate blood supply to the branchial arch during the period of facial development, usually at about the first 4-10-weeks gestational age, and the restraining or physical restrictive forces of amniotic bands which may lead to the formation of unusual clefts. Amniotic bands can result in the incomplete formation of facial structures in humans [2,11]. Genetic factors contributing to syndromic facial cleft include cleft-lip-and-palate transmembrane protein 1, glutamic acid decarboxylase1 (GAD1), and variation in Interferon regulatory factor 6 (IRF-6) gene seen in Van Der-Woude syndrome, which increases the occurrence of clefts by threefold. These factors have been implicated in protein binding, palatal development, cellular differentiation, cellular development, and the regulation of these activities [12,13].

Identifiable risk factors for orofacial clefts include increased maternal age, maternal stress, poor maternal nutrition with vitamin A and folic acid deficiency, maternal hypoxia from cigarette smoking, abuse of alcohol, cocaine, and heroin, anticonvulsant drugs like phenytoin and phenobarbitone, exposure to lead and pesticides, low socioeconomic status, consumption of herbal concoctions in pregnancy and genetic history [2, 14]. In addition to facial findings, other associated anomalies include hypoplasia of innervation of fifth and seventh cranial nerves, palate and tongue, preauricular skin tag, polydactyly, cardiac and renal anomalies [15]. Prenatal diagnosis of TFC is possible with a 3-dimensional ultrasound scan [6]. Although anomalies can be detected as early as ten weeks of gestation, sensitivity for detecting structural anomalies improves with advancing gestational age in the second trimester [16]. Current international guidelines have recommended routine mid-trimester ultrasound scans for evaluation of fetal face and other congenital anomalies. This congenital deformity is disfiguring but can be surgically corrected. We present a case of TFC, Tessier number 7 in a newborn, and highlight the challenges associated with its management. This report aims to create awareness of the existence of this entity to clinicians and reinforce the need to avoid modifiable risk factors, and encourage routine pre-conception folic acid for all women of child-bearing age.

CASE REPORT

Baby R2 was the 2nd of a set of twins delivered via spontaneous vaginal delivery to a 26-year-old para-2^{+0} mother at 28-weeks gestation. Pregnancy was managed from 6-weeks gestational age by a traditional birth attendant (TBA) who gave the mother weekly herbal concoctions. She did not receive any orthodox medication or antenatal care throughout pregnancy. She was neither hypertensive nor diabetic and had no history to suggest any other chronic illnesses. She did not smoke or take alcohol. The young mother had a pre-term premature rupture of membranes at 28-weeks of gestation and delivered a pair of male twins at the home of the TBA. The twins were referred to our facility shortly after birth. Baby R2, who had gross facial deformities, appeared dyspnoeic and icteric at presentation. Upon examination, Baby R2 had bilateral corneal opacity, bilateral wide transverse facial cleft extending from ear to ear with left unilateral complete cleft lip and palate, right incomplete cleft lip, and anomalies of the maxilla and zygoma (Fig.1). No other deformities were detected, except for a very low birth weight of 1400g. On the other hand, twin R1 who had no obvious congenital anomalies only weighed 1200g. A provisional diagnosis of Treacher Collins and Peter Plus syndromes was made based on clinical findings.

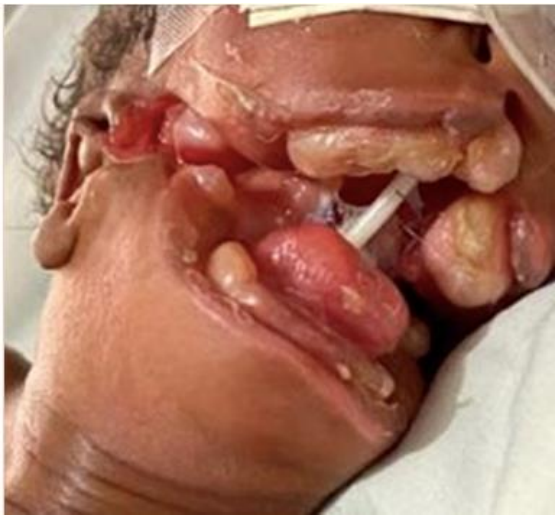

Figure 1 The bilateral transverse facial cleft extending from ear to ear

The patient was admitted and commenced on intravenous fluids, oxygen via face mask, and phototherapy with regular ocular care. A multidisciplinary team involving the neonatologists, oral and maxillo-facial cleft surgeons, ophthalmologists, and psychologists were involved in his management, and the line of management was agreed by all. The complete blood count was suggestive of sepsis, for which empirical antibiotics were commenced. Blood culture yielded no growth. An echocardiogram showed no structural heart defect. Chest X-ray could not be done as the child was too ill to be moved to the radiology department. Parents were appropriately counseled on the patient's condition and the required surgical

interventions. They were very anxious about the facial deformity and were eager for any intervention that could be offered. The child's low weight and severe respiratory distress precluded immediate surgical intervention. The respiratory distress progressed to recurrent apnoeic episodes, and the baby died two days after admission before any surgical intervention could be offered. Parents declined an autopsy.

DISCUSSION

Bilateral TFC is very rare, and this is the first documented case in our hospital. Our patient was a male child, and the study done by Oghale et al. showed a male preponderance [2]. We could not determine the cause of TFC in our patient; however, he was exposed to several risk factors such as maternal liberal use of herbal concoctions and lack of folic acid intake throughout pregnancy. Omo-Aghoja et al. [14] noted indulgence in the intake of herbal medications in pregnancy as an important risk factor. The use of herbal medications was also suggested in the study by Oghale et al. [2]. Many of the implicated herbal concoctions are alcohol-based. Therefore, it is possible that our patient's mother was exposed to chronic alcohol ingestion, which started at the critical period of embryonic development. It is also possible that the maternal periconceptional folic acid level was low, and the use of the alcohol-based concoctions could have further reduced this. Folic acid deficiency and alcohol abuse were both important risk factors for facial clefts. Previous studies found a reduction in orofacial cleft with folic acid intake. The study by Wilcox et al. [17] noted that daily folic acid supplementation reduces the risk of cleft lip, with the lowest risk seen in those who took daily folic acid in combination with multivitamins or a good diet. A similar study by Yazdy et al. [18] reported a 6% reduction in the prevalence of orofacial clefts with folic acid supplementation.

Our index case presented with the syndromic form of the TFC with bilateral corneal opacity and defects of the lip, palate, and alveolus. These features may also be found in both Treacher Collins and Peter-Plus syndrome [19,20]. While Peter-Plus syndrome has prominent limb deformities and developmental delay [20], our patient did not have limb deformities. In Treacher Collins syndrome, mandibulofacial dysostosis is prominent with coloboma but with normal intelligence [19]. Our patient had no coloboma but did not live long enough to assess his cognitive capacity. There were no other features to suggest amniotic band syndrome.

As with any other facial congenital anomalies, orofacial clefts should be repaired early to relieve parental anxiety. Multiple counseling sessions are necessary to allay parental fears and anxieties. This will also improve their co-operation and participation in any intervention being given. Our index parents were very anxious, but with counseling and encouragement, they were willing and able to provide the resources needed for their child's treatment until the time of demise. Adequate counseling and communication may prevent catastrophic reactions such as the suspected infanticide reported by another study [21]. Cosmetic distortion of the face threatens the quality of life, leading to reduced self-esteem and confidence [19]. This condition is amenable to surgery, and in Nigeria, "Smile Train" – a non-governmental organization (NGO), has been involved in helping children born with facial clefts to finance their repair. This NGO, through a partnership with our institution, was actively involved in the management of the index case until his demise. Though many operative techniques have been described, the repair goals are symmetry of lip, commissural shape, and restoration of oral sphincter with good and minimal scarring quality. Our patient, however, died from other complications before any of these could be done.

CONCLUSION

Transverse facial cleft is a disfiguring but treatable condition requiring a multidisciplinary approach. Adequate attention must be paid to the systemic stabilization of the patient while awaiting reconstruction of soft tissues and the bony frame.

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DECLARATION OF INTEREST

The authors report no conflicts of interest in this work.

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