BILATERAL TRANSVERSE FACIAL CLEFTS (CONGENITAL MACROSTOMIA) -A CASE REPORT

Frances S. Okpokowuruk,[1] Charles Amanari[2]

Dept of Paediatrics,[1] University of Uyo Teaching Hospital, Uyo, Akwa Ibom State.
Burns and Plastics Unit,[2] Dept of Surgery, University of Uyo Teaching Hospital, Uyo

ABSTRACT:

We describe a very rare case of bilateral congenital macrostomia in a three month old female infant with an unusual association of brachycephaly and other dysmorphic features as a result of ingestion of an alcohol based herbal preparation, Sulphadoxine-Pyrimethamine and probably supplemental doses of Vitamin E in the early first trimester of pregnancy. This case report emphasizes the need for maternal education and support of maternal health care.

Key words: Congenital, Macrostomia, Z-plasty

INTRODUCTION:

Transverse facial cleft, otherwise known as congenital macrostomia is a rare congenital abnormality arising from a failure in the fusion of the mandibular and maxillary processes of the first branchial arch.[1] It has an incidence ranging from between 1:60,000 to 1:300,000 [2] with bilateral transverse clefts being even rarer.[3]. They tend to be associated with deformities of structures that arise from the first and second branchial arches such as ear anomalies, preauricular tags and mandibular dysplasia.[4] They may or may not be syndromic though isolated cases i.e. those not associated with a syndrome are also quite rare.[4]. Syndromes that have been associated with congenital macrostomia include the Ablepharon-Macrostomia syndrome, Barber-Say and Otomandibular syndromes. syndromes. [5]Out of 20 reported cases of isolated bilateral macrostomia, only 3 patients were found to be of African descent.[4] Locally, several authors have also reported cases of isolated bilateral macrostomia.[6,7,8] The index case is of African ancestry and not only has bilateral facial clefts but could also not be identified with any particular syndrome hence this report.

Correspondence: Dr F. S. Okpokowuruk
Dept of Paediatrics, University of Uyo Teaching Hospital, Uyo, Akwa Ibom State. Burns and Plastics Unit, Email add: zipsadoo@yahoo.com.

CASE REPORT

PB is a three month old infant who presented in the Unversity of Uyo Teaching hospital with bilateral defects involving the angles of the mouth observed at birth. Prior to the mother's discovery that she was pregnant; she had ingested three tablets of sulphadoxine -pyrimethamine (fansidar) followed by ten tablets of chloroquine being treatment for malaria at a peripheral hospital. She was also taking daily vitamin E capsules prior to the diagnosis of cyesis. She was treated during the third month of gestation with a course of Artemether-lumefantrine when the initial treatment for malaria failed. She also admitted to occasionally taking alcohol based herbal preparations during the index pregnancy. She had regular antenatal care at the same peripheral hospital and had no further problems.

Mother went into spontaneous labour at 30 weeks of gestation and delivery was by assisted breech. The baby cried immediately after birth and was nursed in the neonatal unit of the peripheral hospital on account of prematurity and low birth weight(weight (1300g) for four weeks. She developed hyperbilirubinaemia on the third day of life and was managed with phototherapy and exchange blood transfusion.

At the age of eleven weeks, she presented at the teaching hospital with complaints of fever and passage of watery stools of one week's duration and was subsequently managed for sepsis and discharged to the plastic surgery unit after one week of admission.

The patient was fed with breast milk and pre-term formula milk from birth until nine weeks of age when mature formula was introduced. She also suckles at the breast though breast milk escapes from the defects at the angle of her mouth. She is the first and only child of parents. The mother is a 33 year old house wife while the father is a 35 year old civil servant and both of them have tertiary level of education.

Physical examination revealed a well nourished and alert infant with brachycephaly, lowbrachycephaly, low set ears, flattened nasal bridge, hypertelorism, high arched palate and bilateral commisural clefts of the lips. The right transverse facial cleft measured measured 2cm in length while the left measured 1cm. She had no other significant abnormality on physical examination. 2-D echocardiography carried out was also normal though other possible in apparent congenital anomalies could not be ruled out because the patient did not have a total body Ct-scan done. She was subsequently referred to the plastic surgery unit where she had a surgical repair incorporating a Z-plasty with a very good functional and aesthetic outcome.

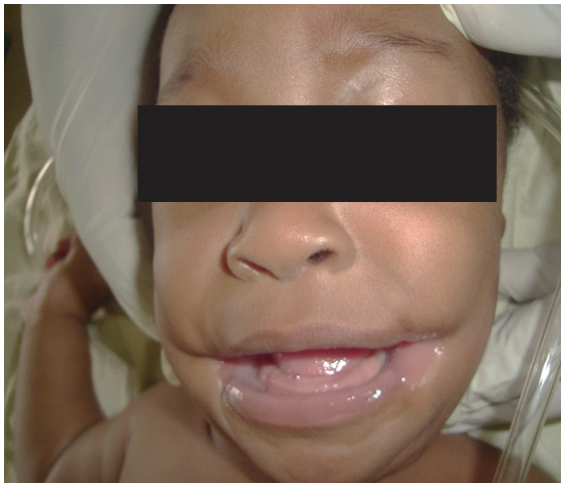

Fig 1: Preoperative photograph of the patient

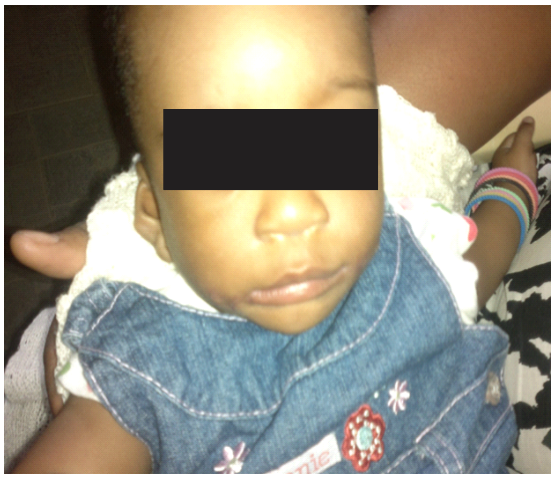

Fig 2: Postoperative photograph of the patient (2 weeks after surgery)

DISCUSSION

Congenital macrostomia, a Tessier cleft no. 7, is one of the rarest orofacial clefts.[9] It results from failure of fusion of the mandibular and maxillary processes that form that form the mouth during the fourth and fifth weeks of gestation (early first trimester).[10] The anomaly is believed to result from a combination of genetic and environmental factors.[11] Several mechanisms for the anomaly have been proposed including mutations of the surface proteins,[12]and disruption of the stapedius artery.[13]. Based on human data on toxicity in pregnancy, choroquine is considered safe in pregnancy[14]. Sulphadoxine-pyrimethamine which the mother ingested early in the first trimester can be teratogenic, has been associated with orofacial clefts in experimental studies in rats.[15]A case control study showed a higher exposure to sulphonamides among 134 mothers whose babies had cleft palate than among controls.[16]. Artemisinin derivatives, though embryotoxic in rats, has not been shown to be so in humans.[17,18]. Lumefantrine did not show any embryotoxicity in animal studies, and there is no human data on toxicity in pregnancy.[19]Other possible environmental factors that were noticed in her pregnancy history include the regular ingestion of vitamin E possibly into her pregnancy, and ingestion of alcohol through herbal medications. While alcohol is a known

teratogen,[120] there are conflicting reports on whether high vitamin E intake is teratogenic or not. While some report an increased incidence of various congenital anomalies like congenital heart diseases (syndromic and non-syndromic), hip dysplasia, hydronephrosis, deafness and hydrocephalus with high doses of vitamin E, others report no significant association with fetal anomalies.[21,22]Orofacial clefts have also resulted from several chromosomal anomalies involving specific deletions and duplications.[23].OtherOtherassociated environmental factors include nutritional deficiencies especially folic acid deficiency, tobacco use, ingestion of certain anticonvulsants and sedatives, and pesticides like dioxin.[20,24,25, 24,25]

Congenital macrostomia can be partial or complete. Most congenital macrostomia are of the partial variety, terminating before the anterior border of the masseter, [26], as in this patient. Congenital macrostomia are more commonly syndromic, but they can be non-syndromic occurring either alone, or in association with other anomalies.[27]More often the associated anomalies involve derivatives of the first and second branchial arches,[28], [28]like in this case where it is associated with brachycephaly and other dysmorphic features previously outlined. Various methods of repair has been described for macrostomia including the straight line closure, Z-plasty, and W-plasty however, Z-plasty offers a good aesthetic and functional outcome as was seen in this patient while limiting the possibility of contracture formation.

The circumstances surrounding the pregnancy leading to the delivery of this patient with congenital macrostomia points to need for adequate health education of mothers in our locality on the teratogenic effects of alcohol and drugs-, orthodox or herbal, in pregnancy, and the need to be mindful of what they ingest during their reproductive years. Adequate funding for maternal health will also help reduce this problem as poverty contributes to the continued patronage of herbal practitioners by these mothers to reduce cost.

CONCLUSION

We described a very rare case of bilateral congenital macrostomia in a 3 month old female infant with an unusual association of brachycephaly and other dysmorphic facial features as a result of ingestion of alcohol based herbal medication, sulphadoxine-pyrimethamine and probably supplemental doses of Vitamin E in the early first trimester. This case report emphasizes the need for maternal education and support of maternal health care. The repair by the Z-plasty technique offers an excellent aesthetic and functional outcome.

REFERENCES

1. Tessier P. Anatomic classification of facial, craniofacial and laterofacial clefts. J Maxillofac Surg. 1976; 4 :4: 69-929-92.

2. Chen K.T, Noordholff S.M. Congenital macrostomia-transverse facial cleft. Chang Yi XueZaZh. I 1994; 17: 236-249.

3. Hawkins D.B, Miles J.W, Seltzer D.C. Bilateral macrostomia as an isolated deformity. J Laryngotol.1973; 87 :87: 309-313.

4. Arnaud Gleizal, Derrick C. Wan, Arnaud Picard, Jean-Franc¸oiseLLavis, Marie-Paule Vazquez, Jean-Luc Beziat. Bilateral Macrostomia as an Isolated Pathology. Cleft Palate–Craniofacial Journal., January 2007; Vol. 44(No. 1):; 58-61.

5. Haensel J, Kohlschmidt N, Ppitz S et al. Case report supporting that the Bbarber Say and Aablepharonmacorstomia syndromes could represent one disorder. Am J Med Genet A. 2009;Oct;149A(10):2236-2240.

6. Akinmoladun V.I, Derrick C.W, Afolabi A.O. Bilateral transverse facial cleft as an isolated deformity: case report. Ann Afr Med 2007;6:39; 6:39-40.

7. Fadeyibi I.O, Ugburo A.O, Fasawe A.A, Idris O, Ademiluyi S.A. Macrostomia: a study of 15 patients seen in Lagos, Nigeria and proposal for a classification of severity. J Plast. Surg 2010 Dec;44; 44(6):289-295.

8. Oghale OP, Chris- Ozoko LE. Asyndromic bilateral transverse facial cleft. Ann Med Health Sci Res. Case report. 2013;3; 3(1):122-124.

9. Shima Y, Ogawa K, Kuwabara Y, et al. Newborn with transverse facial cleft associated with polyhydramnios. JPerinatol2002;22; 22(1):91-2.

10. Powell W.J, Jenkins H.P. Transverse facial clefts. Plast. Reconstr. Surg. 1968;42:454; 42:454–459.

11. May H. Transverse facial clefts and their repair. Plast. Reconstr. Surg. 1962;29:240; 29:240–249.

12. Askar A, Gurlek A, Sevin K. Lateral facial clefts(macrostomia). AnnPlasticSurg2001;47; 47(3):355-6

.13. Ahmad Khaleghnejad-Tabari, Katayoun Salem, Masoud F. Allahinejad Ghajar. Treatment of Bilateral Macrostomia (Lateral Lip Cleft): Case Report. Iran J Pediatr. 2012; 22(3):425-427.

14. Wolfe M S, Cordero JF. Safety of Chloroquine in chemosuppression of malaria during pregnancy. Br Med J. 1985;290(6480):1466-1467.Wolfe M.S, Cordero J.F. Safety of chloroquine in chemosuppression of malaria during pregnancy. Br Med J 1985; 290(6480): 1466-7

15. Phillips-Howard P.A, Wood D. The safety of antimalarial drugs in pregnancy. Drug Saf 1996; 14(3): 131-145.

16. ..Nelson M.M, Forfar J.O. Associations between drugs administered during pregnancy and congenital abnormalities of the fetus. Br MedJ 1971; 1(5748): 523-7.

17. Studies on the toxicity of qinghaosu and its derivatives. China Cooperative Research Group on qinghaosu and its derivatives asantimalarials. J Tradit Chin Med 1982; 2(1): 31-38

18DeenJJ.L, von Seidlein L, Pinder M, Walraven G.E, Greenwood B.M. The safety of the combination artesunate and pyrimethamine sulfadoxine given during pregnancy. Trans R Soc Trop Med Hyg2001; 95(4): 424-8.

19. François Nosten, Rose McGready R, Umberto d'Alessandro, Ana Bonell, Francine Verhoeff, Clara Menendez, Thenonest Mutabingwa, and Bernard Brabin. Antimalarial Drugs in Pregnancy: A Review. Current Drug Safety., 2006;,*1:1*,1-15.

20.Wyszynski, D.F, . and Beaty, T.H. (1996) Review of the role of potential teratogens in the origin of human nonsyndromic oral clefts. Birth defects Res a Clin Mol Teratology. 1996; ,53: 309–317.

21.Smedts H, de Vries J, Rakhshandehroo M, Wildhagen M, Verkleij-Hagoort A, Steegers E, Steegers-Theunissen R. High maternal vitamin E intake by diet or supplements is associated with congenital heart defects in the offspring. BJOG 2009;116:416; 116:416–423.

22. Szilasi M, Bártfai L, Bártfai Z, Bánhidy F, Czeizel A.E. No association of

maternal vitamin E intake with higher risk of cardiovascular malformations in children: a population-based case–control study. Therapeutic Advances in Drug Safety. 2011; vol. 2: no. 377-386.

23. Schutte BC, Murray JC. The many faces and factors of orofacial clefts. Human Molecular Genetics.,1999;, Vol. 8:,No. 10 Review 1853–1859.

24. Warkany, J., Nelson, R.C. and Schraffenberger, E. (1943) Congenital malformations induced in rats by maternal nutritional deficiency. Am. J. Dis. Child.1943; 65:, 882–894.

25. Garcia, A.M., Fletcher, T., Benavides, F.G,. and Orts, E. (1999) Parental agricultural work and selected congenital malformations. Am. J. Epidemiol. 1999; 149:, 64–74.

26. May H. Transverse facial clefts and their repair. *Plast*. Reconstr. Surg. 1962;29:240–249.

27. Darlene M, Guse B.S, Vincent A. Bilateral macrostomia. *Eplasty*2010:10. Available at: http://www.eplasty.com/images/PDF/eplastyd-10-00011.pdf. Access date: April 1, 2010.

28. Eguchi T, Asato P.H, Takushima A, Takato T, Harri P. Surgical repair for congenital macrostomia: Vermillion square flap method. Ann Plast. Surg. 2002;48s:328; 48:328-9.