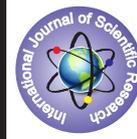


Lower jaw defect anomaly-rare first arch syndrome



Medical Science

KEYWORDS:

ANSHU SHARMA

ASSOCIATE PROFESSOR,GMCH,SECTOR 32,CHANDIGARH,INDIA160030.

JOSEPH ABRAHAM

KAITHARAN HOUSE,MANALIKADU,MADATHUMPADY P O,THRISSUR,INDIA

AMRUTHA K V

KUDIL,TRICHAMBARAM,TALIPARAMBA,KANNUR,KERALA,INDIA,670141.

Introduction

Maxillofacial defects are common defects occurring congenitally. First pharyngeal arch, which are composed of mesenchymal cells derived from mesodermal and cranial neural crest cells, give rise to a wide variety of facial structures including skeletal, muscular and neural elements through a complex signaling network still poorly characterized. Cleft of mandible and lower lip are comparatively rare and form severe anomaly amongst this group. Severe and complete division of the mandible is also called as inferior gnathochisis. These defects may be found alone or they may form a part of some syndrome¹

Case report

A male fetus of 19+2 wks with congenital malformation was sent for routine autopsy by department of OBG, GMCH, Chandigarh. Congenital anomaly was diagnosed on u/s as microcephaly and 2 heterogenous foci posterior to skull and neck. Relevant history from parents were taken. Fetus was examined externally and was photographed, radiographed. Routine autopsy procedures were followed for observing other defects associated with first arch anomaly, vertical incision was extended into neck. Findings were noted and described in light of available literature. Mother was 22 years old 2nd gravida. First pregnancy resulted in abortion due to fall in 1st trimester. Family history of parents, past history, medical history was found to be not suggestive of any contributing factor. There was no history of drug intake, overdose of vitamin A, steroid or alcohol and tobacco abuse during pregnancy. Antenatal check up and vaccination were done as per schedule. On external examination, there was hypertelorism (fig 1), right palpebral fissures were not opened, coloboma of left upper eye lid, persistent upper part of nasomaxillary groove (fig 1), nose deviated to right side (fig 1), Cystic swelling on right parietal region and occipital region (fig 1), Small neck (fig 1), B/L club foot (fig 2) in lateral four fingers of right hand, distal phalanges were atrophied (fig 2). Left hand was normal. Internal examination revealed ankyloglossia, laryngeal hypoplasia, small epiglottis (fig 1), 2 cystic swelling on head were encephalocele (fig 1). Heart was hypertrophied. Radiograph showed defects in skull, mandible, neural arch in lower cervical and upper thoracic spine (fig 3,4).

Discussion

Orofacial clefts were first reported by Couronne in 1989, lower jaw defect may occur alone or part of syndrome. Variable severity with variable presentation. Incidence is 1.7-2%. autosomal recessive inheritance, recurrence in sibs and cousins 36%, parental consanguinity 44 % in Brazilian population. No significance preponderance in sex². Multiple genetic and environmental factors, defect in nucleolar trafficking protein, vitamin A toxicity, steroid, birth control pills, antihistaminic drugs intake by mother during antenatal period were some of etiological factors³

Mandible is derived from first pharyngeal arch cartilage known as Meckel's cartilage which is formed after mesenchymal condensation at 36-38 days of development. In mental region, mental ossicles appear in 7th month. Symphysis menti is converted from a syndesmosis to synostosis in first postnatal year⁴

Most authors say defect is due to lack of mesodermal penetration

into midline structures of mandibular portion of first branchial arch. Disturbing influence occurs at the end of fifth or at the beginning of 6th week of gestation. According to Morton and Jordan, failure of ventral end of mandibular process may keep ventral ends of succeeding arches from uniting as fusion proceeds from above downwards leading to absence of hyoid, thyroid cartilage, strap muscles and manubrium in more severe cases. Factors for impeded migration can be mechanical and vascular anomalies with secondary ischemia and necrosis. Few investigators suggest that pressure on cervical cord by pericardial roof could prevent fusion. Some other investigators suggest role of hyaluronate and collagen. Associated deformities in other part of body may be due to external factors resulting in widespread disturbances in embryological development⁵

Marion classified clefts into type 1-cleft present between midline of face and infraorbital foramen, type 2-cleft between infraorbital foramen to lateral aspect of face AACPR classified into 4 groups.

1-mandibular process cleft- which included mandibular cleft lip, mandibular cleft, lower lip pits

2-nasooocular cleft-extending from nasal region to medial canthal region

3-orooocular cleft- extending from angle of mouth towards palpebral fissure

4-oroaicular cleft- extending from angle of mouth towards ear

According to Boo Chai classification (which was a modification of Marion classification) oro ocular clefts were divided into type 1 and type 2 (oblique facial cleft)⁶

Some more classification were given by Karfik, Lund, De meyer⁷

Most accepted classification was given by Paul Tessier in 1973, according to him, all clefts were numbered from 0-14 with a number 30 for a medial symphyseal cleft in mandible. Classification centered an orbit, with facial cleft numbered from 0-7 and cranial cleft from 8-14 in a counter clock wise rotation being ,30 in midline of mandibular symphysis. Cleft could be complete or manifest as simple notch or complete division or grooving of part⁸

In our present case we came across midline symphyseal cleft.

Congenital heart disease, cleft of tongue, ankyloglossia, oligodontia, absence of hyoid bone, thyroid cartilage, manubrium of sternum, hypoplasia of epiglottis, larynx, strap muscles of neck, neck contractures, malformation of limbs were associated findings⁹

Conclusion

Treatment of these conditions is mainly corrective through surgical interventions. Mandible reconstruction is done after 10 years of age to minimize damage to developing tooth buds. when required, reconstruction of mandible is done with rib graft, iliac bone graft along with titanium reconstruction plate or an acrylic splint.

Figures

Fig 1



Fig 2

Fig 3



Fig 4



References

1. Gorlin RJ. 2001. Branchial arch and oro-acral disorders. In: Gorlin RJC, Hennekam RCM, editors. Syndromes of the Head and Neck. 3rd edition. Oxford: Oxford University Press. p 790–849
2. Monroe CW: Midline cleft of the lower lip, mandible and tongue with flexion contracture of the neck: Case report and review of the literature. *Plast Reconstr Surg* 38:312, 1966
3. Shiho Nakamura,1 Haruyoshi Yamaza,1 Soichiro Nishigaki,1 Keiji Masuda,2 Ken-ichi Yanagita,2 and Kazuaki Nonaka Case Report Clinical Approach to a Suspected Case of First Branchial Arch Syndrome Noboru Yamaguchi, Hindawi Publishing Corporation Case Reports in Medicine Volume 2014, Article ID 506804, 5 pages <http://dx.doi.org/10.1155/2014/506804>
4. Arshad AR. Incomplete midline cleft of lower lip. *Cleft Palate Craniofac J*. 1995;32:167–169
5. Charles Bruce Morton, M.D.; Harvey Ernest Jordan, Ph.D. Median cleft of lower lip and mandible, cleft sternum and absence of basihyoid: report of a case. *Arch Surg*. 1935;30(4):647-656. doi:10.1001/archsurg.1935.01180100093010.
6. Boo chaik, the bifid nose. *plast reconstr surg* 36:626, 1965
7. De Meyer W. Median facial malformations and their implications for brain malformations. *Birth Defects*. 11:155-81, 1975
8. Paul Tessier. Anatomical classification of facial, cranio-facial and latero-facial clefts. 1976. Volume 4, Pages 69–92
9. Roshani E Rana, Vinita A Puri, Rahul K Thakkur, AS Baliarsing CASE REPORT .70 Median cleft of mandible and lower lip with ankyloglossia and ectopic minor salivary gland on tongue. Year: 2004 | Volume: 37 | Issue: 1 | Page: 67-69