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CASE REPORT

FEEDING APPLIANCE FOR AN INFANT WITH HOLT-ORAM SYNDROME ASSOCIATED CLEFT LIP AND PALATE: REPORT OF A RARE CASE

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ABSTRACT

A three-month-old male infant was brought to the Department of Pedodontics and Preventive Dentistry with the chief complaint of feeding difficulties. On examination, bilateral cleft lip with complete cleft palate and upper limb deformities were found. In radiographic examination of forearms and chest absent radii and thumb, left rib crowding and neonatal right ventricular hypertrophy were evident. The diagnosis of Holt-Oram syndrome was made based on clinical, radiological and echocardiographic findings. A feeding appliance was fabricated and delivered to the patient.

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INTRODUCTION

The Holt–Oram syndrome (HOS) or atriodigital dysplasia, a developmental disorder of the heart and upper limbs was first described by Mary Clayton Holt and Samuel Oram in 1960. It has autosomal dominant inheritance, near complete penetrance and variable expression; affecting one in 100,000 live births of which 40% of cases are sporadic (Holt and Oram, 1960).The phenotypic features are represented by upper limb malformations with associated cardiac lesions. Orthopedic signs include dysplasia of the upper limbs, thumb hypoplasia, clinodactyly, brachydactyly, triphalangeal thumb, dysmorphic carpal bones, ulnar shortness, humerus shortness or hypoplasia, absence of radius and phocomelia (James *et al*.,1996).The cardiac disorder involves atrial or ventricular septal defects, patent ductus arteriosus, endocardial cushion defects, hypoplasia of left ventricle or conduction disturbances such as 1st degree heart blocks (Bruneau *et al*., 1999; Webb and Gatzoulis, 2006) . The most common cardiac disorder in HOS is ostium secundum atrial septal defect(ASD) followed by ventricular septal defect (VSD) and ostium primum ASD (Bossert *et al*., 2003). Other features include pulmonary and subaortic stenosis, tetralogy of Fallot, Cardiomyopathy etc.

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Cleft lip and palate is one of the most common developmental defects of the head and neck region. Neonates with cleft palate face extreme difficulty in eating, which often leads to failure to thrive (Goldberg WB, Fergusson FS, Miles RJ, 1988). The association between HOS and cleft lip and palate has been well documented and explained based on their genetic backgrounds. This article presents a case report of a three-month-old infant having HOS associated cleft lip and palate in which feeding plate was delivered.

CASE REPORT

A three-month-old male infant was brought to the Department of Pedodontics and Preventive Dentistry with the chief complaint of feeding difficulties. On examination, it was found that the child was born with bilateral cleft lip and complete cleft palate (Fig. 1) along with upper limb deformities (Fig. 2, 3). Antero-posterior view of both forearms and chest showed absent radii and thumb, left rib crowding and neonatal right ventricular hypertrophy (Fig. 4, 5). Echocardiography showed evidence of large ostium primum ASD, large subaortic VSD with more than 50% aortic overriding, pulmonary stenosis and malposed great vessels. There was no significant family history. After examination it was decided to fabricate a feeding appliance to reduce the feeding problem. Primary impression (Fig. 6) was taken with impression compound holding the child upright by his mother.

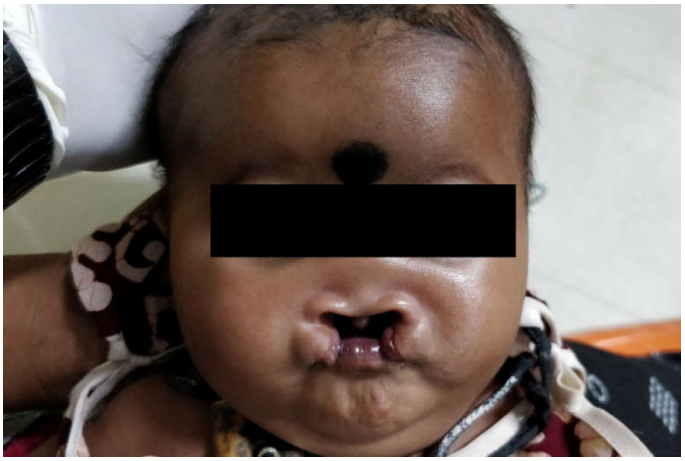

Fig. 1. Bilateral cleft lip and complete cleft palate

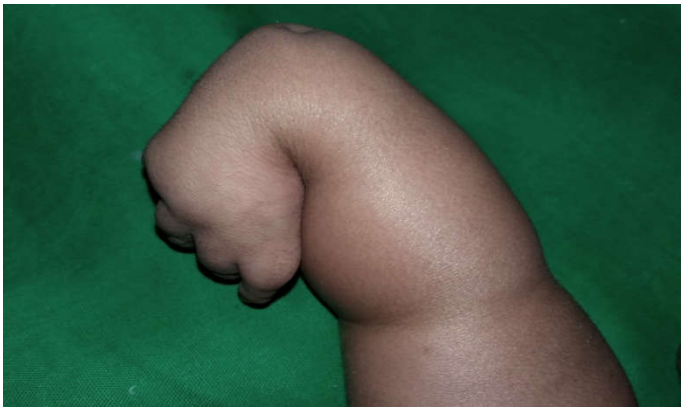

Fig. 2. Deformity of right arm

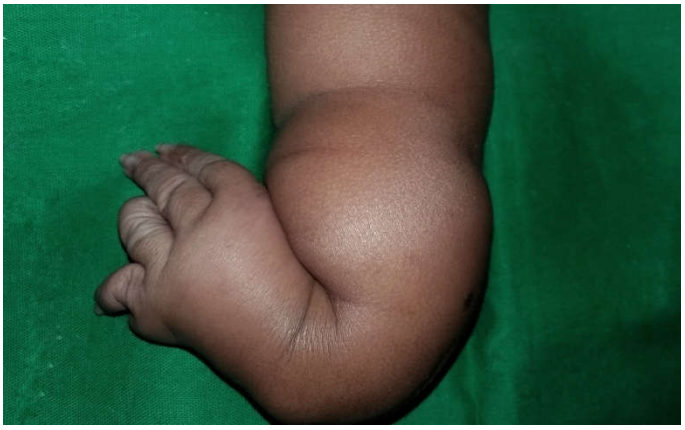

Fig. 3 Deformity of upper left arm

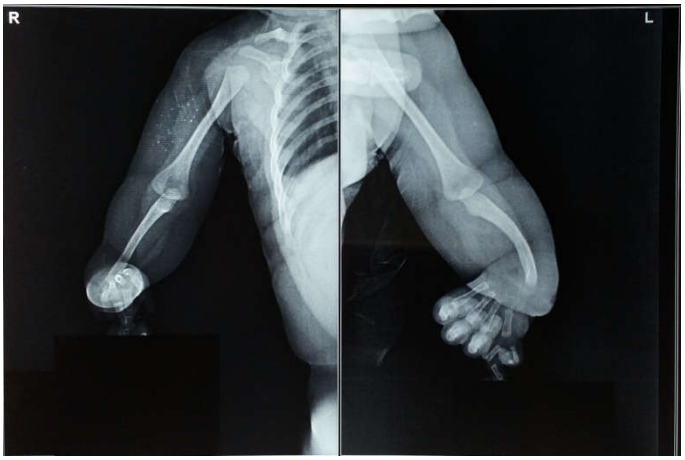

Fig. 4. Absent radii and thumb

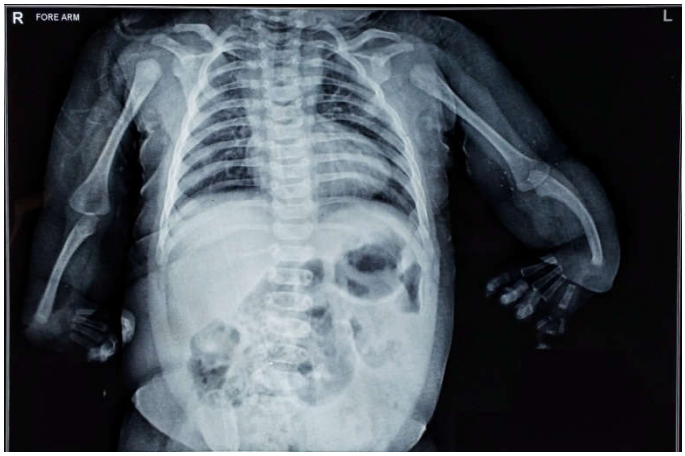

Fig. 5. Left rib crowding and neonatal right ventricular hypertrophy

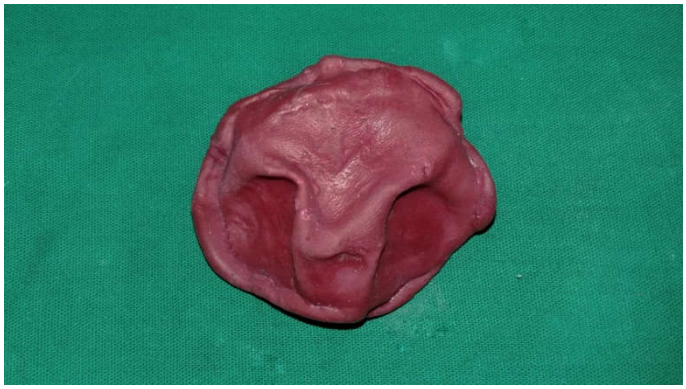

Fig. 6. Primary impression

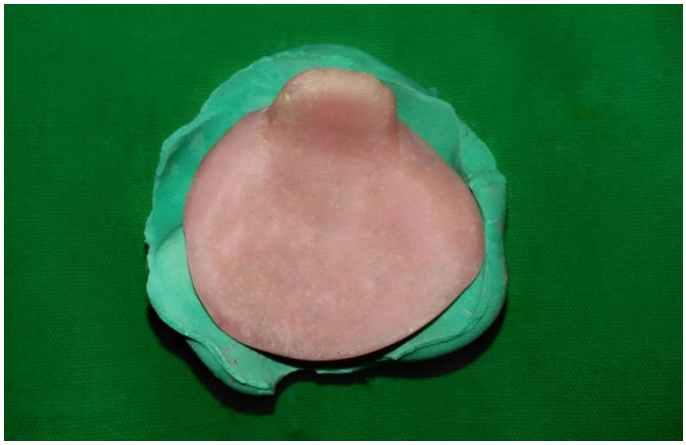

Fig. 7 Special tray

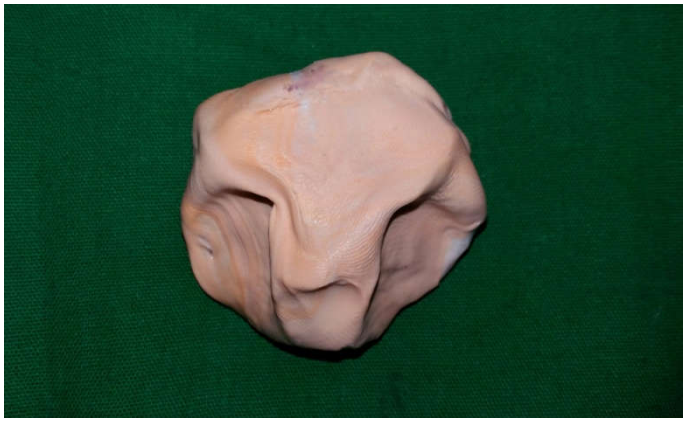

Fig. 8 The final impression

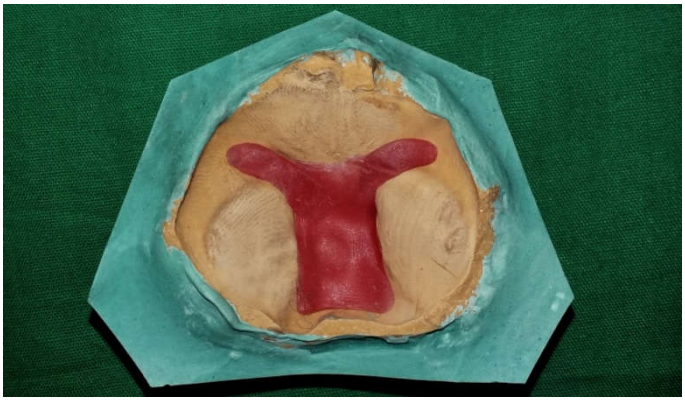

Fig. 9. The model cast with undercuts blocked

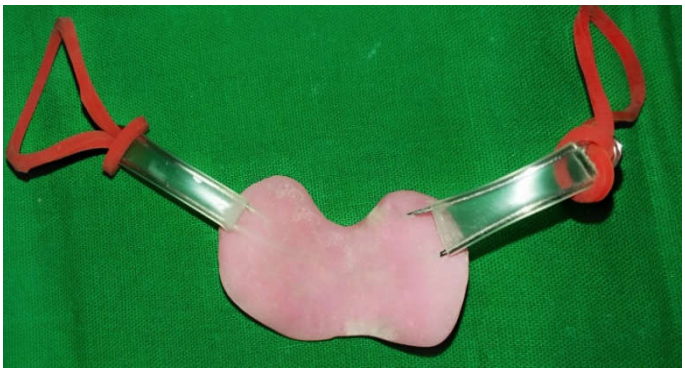

Fig. 10. The feeding plate made with autopolymerising resin

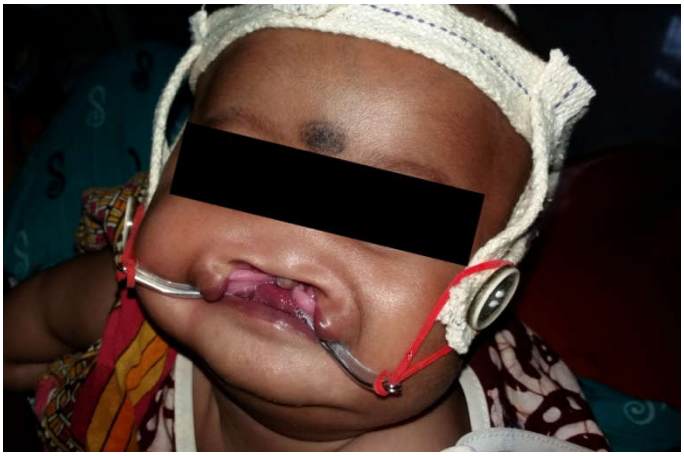

Fig. 11. Appliance inserted into the patient's mouth with customized headgear

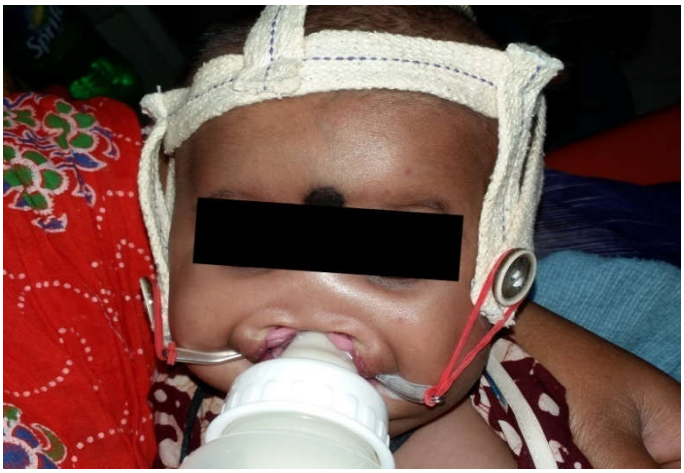

Fig. 12. The child successfully able to be feed with feeding appliance in place

A special tray was made using self-cured acrylic resin (Fig. 7) and final impression was secured with elastomeric impression material. (Fig. 8). Final model cast was made and all undercuts were blocked (Fig. 9). Feeding appliance was fabricated with auto polymerizing resin (Fig. 10), properly polished to avoid injury to the oral mucosa and inserted into the patient's mouth with the help of customized head gear (Fig. 11). This head gear helps to prevent accidental swallowing of the appliance and in easy retrieval of the appliance. The patient's mother was asked to bottle fed the baby and it was seen that there was no nasal regurgitation and the child was successfully able to be feed with feeding appliance in place without any discomfort (Fig. 12). Instructions were given to parents about insertion, removal and cleansing of the prosthesis. A regular follow up was done after 24 hours, 1 week and monthly interval. Weight gain of the infant was evident during regular follow ups.

DISCUSSION

HOS is also known as atriodigital syndrome, heart-hand syndrome, upper limb-cardiovascular syndrome, cardiac-limb syndrome, or cardiomelic syndrome. The responsible gene is TBX5 which is located on the long arm of chromosome 12q24.1and belongs to the T-box gene family, which encodes a large family of transcription factors (more than 20 members identified in humans), with key role in embryonic development. Mutations in T-box genes are responsible for HOS (TBX5), ACTH deficiency (TBX19), ulnar-mammary syndrome (TBX3), DiGeorge syndrome (TBX1) and cleft palate with ankyloglossia (TBX22) (PACKHAM E. A., BROOK J. D., 2003). It clearly suggests the association between HOS and Cleft lip and palate where both the anomalies occur due to T-box gene mutations. .Target genes are also several cardiac-expressed genes, which include cardiac apha-actin, atrial natriuretic factor, cardiac myosin chains and SALL4. TBX5 and SALL4 interact both positively and negatively to regulate the patterning and morphogenesis of the forelimb and heart (Koshiba-Takeuchi *et al.,* 2006). In this case, the infant had upper limb deformities along with congenital heart defects, which are typical to that of HOS. Based on the typical limb deformities and associated cardiac lesions, a diagnosis of Holt-Oram Syndrome was made. Genetic testing could not be performed due to non-availability of the facility. Differential diagnosis is to be made from Fanconi Anaemia, Vactrel associations and Radial ray choanal atresia .

Conclusion

The management of infants with HOS associated cleft lip and palate requires a multidisciplinary team approach, with specialists in medical genetics, cardiology, orthopedics and pediatric dentistry. Those individuals born with severe upper-limb malformations may require surgery such as pollicization (creation of a thumb-like digit by moving another digit into the thenar position) in the case of thumb aplasia / hypoplasia, for improved function. A cleft palate creates an opening in the roof of the mouth and the infants face difficulties in sucking as necessary negative pressure, which is needed for sucking, cannot be produced in the oral cavity. Thus, the role of pedodontists is extremely crucial in this case because maintenance of adequate nutrition is essential for the growth and development of the infant. The feeding appliance not only helps in feeding the child but also to position the tongue away from the cleft area in the correct position to allow spontaneous growth of palatal shelves towards each other. In our case also it

has been observed that the weight of the baby has improved after proper treatment. Further research is always required so as to explain the exact cause of clefting of lip and palate and its associated genetic factors for prevention of the baby from this distressing congenital anomaly.

REFERENCES

Bossert T, Walther T, Gummert J, et al. 2003. Holt-Oram Syndrome Orphanet Encyclopedia.

Bruneau BG, Logan M, Davis N, Levi T, Tabin CJ, Seidman JG et al., 1999. Chamber-Specific Cardiac Expression of Tbx5 and Heart Defects in Holt–Oram Syndrome, Developmental Biology, 211: 100 –108.

Goldberg WB, Fergusson FS, Miles RJ. 1988. Sucessful use of a feeding obturator for an infant with cleft palate. *Spec Care Dentist,* 8:86-9

Holt M., Oram S. 1960. *Familial heart disease with skeletal malformations*, *Br Heart J,* 22:236–242.

James MA, Mc Carroll HR, Manske PR. 1996. *Characteristics of patients with hypoplastic thumbs. J Hand Surg Am.,* 21(1): *104-13*.

Koshiba-Takeuchi K., Takeuchi J. K., Arruda E. P, Kathiriya I. S., MO R., Hui C. C., Srivastava D., Bruneau B. G. 2006. Cooperative and antagonistic interactions between Sall4 and Tbx5 pattern the mouse limb and heart, *Nature Genet,* 38(2):175–183.

Packham E. A., Brook J. D., *T-box genes in human disorders*, Hum Mol Genet, 2003, 12(Spec 1):R37–44.

Webb G, Gatzoulis MA. 2006. Congenital Heart Disease for the Adult Cardiologist. Atrial Septal Defects in the Adult: Recent Progress and Overview. *Circulation*, 114: 1645-1653.

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