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Original Research Paper

Neonatology

HOLT-ORAM SYNDROME: RARE CASE SERIES

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ABSTRACT The upper limb malformations in association with congenital heart defects occurring as autosomal dominant disorder are seen in Holt-Oram syndrome. It is a very rare disorder which can be detected with early prenatal ultrasound checkups. Here we are reporting two cases of holt-oram syndrome.

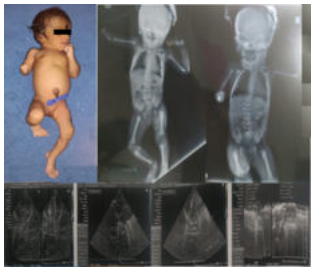
KEYWORDS : autosomal dominant, congenital, heart defects, upper limb, ultrasound.

INTRODUCTION

Holt-Oram syndrome is a rare autosomal dominant disorder with incidence of 1 in 100,000.[1] It is characterized by upper limb malformations and congenital cardiac defects which is first described by Holt and Oram in 1960.[2] Upper-limb malformations can be unilateral or bilateral range from triphalangeal or absent thumb(s) or radial bone to phocomelia [1]. The most common cardiac disorder is an ostium secundum atrial septal defect (ASD), followed by ventricular septal defect (VSD) and ostium primum ASD [3]. It is caused by mutations in two genes of the T-box (TBX5, 601, 620 and TBX 3) located on the 12q24.1p[4]. Diagnosis can be made with family history, clinical findings, X-ray imaging and 2D-Echo. If findings are not enough to diagnose, genetic testing is advisable [1].

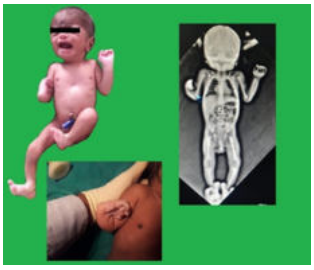
CASE 1

A term female neonate born to a non-consanguineously married couple, 3rd in birth order with antenatal history of mother had threatened abortion at around 10 weeks of gestation (which was managed via progesterone supplements.) and not had regular ANC visits. Baby was delivered by LSCS, birth weight 2.56 kg, cried immediately at birth, APGAR scores were normal. On examination at birth, neonate found to have absence of right distal half of forearm and hand but vitals were within normal limits. An Infantogram, revealed absence of radius in right proximal half of forearm.2-D Echo was suggestive of atrial septal defect of6 mm, patent ductus arteriosus, pulmonary arterial systolic pressure of 38 mm hg, but no clinical signs of respiratory distress were observed and treatment was initiated for findings. Ultrasound abdomen-pelvis and blood investigations were within normal limits. Child was referred to higher centre for further management.



CASE 2

A post term male neonate born to a 30 consanguineously married couple, 1st in birth order with antenatal history of mother consumed unknown variety of medications to relieve morning sickness and not had initial ANC visits. Baby was delivered via emergency LSCS due to severe oligo hydramnios, birth weight 2.32kg, cried immediately at birth, APGAR scores were normal. On examination at birth, neonate found to have malformed right distal half of forearm and hand, but vitals were within normal limits. Infantogram revealed complete absence of radius in right forearm. 2-D Echo was suggestive of atrial septal defect of 5 mm, patent ductus arteriosus and treatment was initiated for findings. Ultrasound abdomen-pelvis and blood investigations were within normal limits. Child was referred to higher centre for further management.



DISCUSSION

Holt-Oram syndrome is inherited in an autosomal dominant manner. Offspring of an affected individual are at a 50% risk of being affected. In pregnancies at 50% risk, detailed high-resolution prenatal ultrasound [7] examination will detect upper-limb malformations and/or congenital heart malformations. Prenatal molecular genetic testing can be used to confirm an affected individual [1]. Virdis et al reported a one case with similar findings.[4] Tidake et al described a case of holt oram syndrome with tetralogy of pallot.[5]. Osonuga et al reported one case of possible holt oram syndrome.[6]

CONCLUSION

Even though Holt-Oram syndrome is a rare disorder, limb anomalies and cardiac lesions can be detected during antenatal screening so it is advised to go through regular ANC visits. If it is detected early termination is advisable. [1].

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