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Case Report

Holt-Oram syndrome: a rare case report

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ABSTRACT

Holt-Oram syndrome is an inherited disorder that causes abnormalities of the hands, arms and heart. The diagnosis can be established clinically. The diagnostic criteria have been validated with molecular testing. An upper-limb malformation involving the carpal bone(s) and, variably, the radial and/or thenar bones-An abnormal carpal bone, present in all affected individuals and identified by performing a posterior-anterior hand x-ray, may be the only evidence of disease. 24 years unbooked Hindu female G2P1+0 presented in OPD at term. Her USG examination showed-small deformed upper limbs with poorly appreciable upper limb skeleton. Induction of labour was done and patient delivered vaginally a female baby with deformed upper limbs. This case emphasizes the importance of proper history taking (family history), early diagnosis of such anomalies and proper counseling the parents.

Keywords: Holt-Oram syndrome, Upper limb malformations, Heart and hand syndrome

INTRODUCTION

Major congenital malformations are reported in at least 2% of all fetuses and infants and have a major impact on perinatal and infant mortality and morbidity in infancy and childhood. Limb reduction defects can be detected in 36% cases by prenatal USG.[1] Holt-Oram syndrome is an inherited disorder that causes abnormalities of the hands, arms and heart. In the United States, it occurs in approximately one in every 100000 children born, and affects both males and females equally.

CASE REPORT

24 years unbooked Hindu female G2P1+0 presented in OPD at term. Her antenatal period was uneventful. There was no past history of any teratogenic drug intake, any radiation exposure, and no history of any apparent skeletal or heart abnormalities in the family of either the female or her husband. On P/A examination uterus was term size with longitudinal lie, cephalic presentation, head was entered, and FHS was 142 beats/min and regular. On P/V examination cervix was closed, uneffaced and pelvis appeared to be adequate. Her USG examination showed-small deformed upper limbs with poorly appreciable upper limb skeleton (Figure 1); lower limbs appeared unremarkable. Thoracic abdominal ratio was altered, stomach bubble was seen normally, and UB was normal; craniospinal region was unremarkable, amniotic fluid more than adequate, largest pocket 10 cm. Patient was referred to a pediatric surgeon who advised a fetal echocardiography and a confirmatory molecular genetic testing. ECHO showed no cardiac abnormality. Genetic testing could not be done due to non-availability of the facility. Parents were counseled regarding the condition of the baby. Induction of labour was done and patient delivered vaginally a female baby (Figure 2). The baby had bilaterally deformed upper limbs with normal lower limbs no other apparent skeletal abnormality. On right side humerus, radius and ulna were absent with one finger and a thumb attached to the body like stumps. On left side, arm length was normal; forearm was markedly shortened with only three fingers attached. The baby was handed over to the parents as they were not ready to get any further investigations done. This case highlights the importance of proper history taking (family history),

early diagnosis of such anomalies and proper counseling the parents.

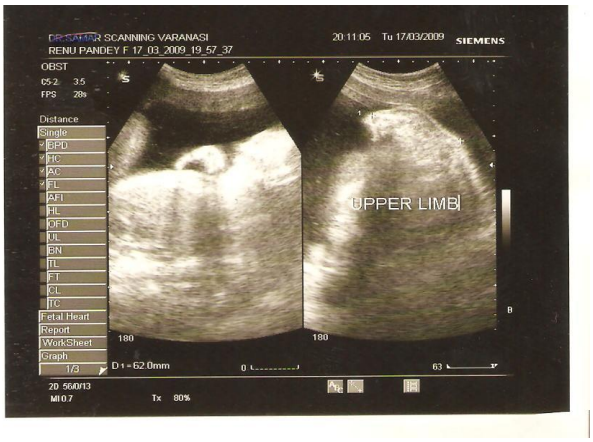

Figure 1: Antenatal USG.

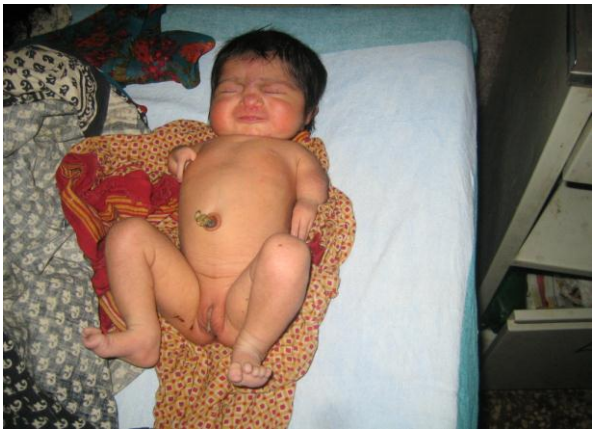

Figure 2: Neonate with upper limb deformity.

DISCUSSION

The baby was a case of Holt-Oram syndrome (Heart and hand syndrome). The diagnosis can be established clinically. The diagnostic criteria have been validated with molecular testing.[2] Clinical findings in HOS are

- An upper-limb malformation involving the carpal bone(s) and, variably, the radial and/or thenar bones- An abnormal carpal bone, present in all affected individuals and identified by performing a posterior-anterior hand X-ray,[3] may be the only evidence of disease.
- A personal and/or family history of congenital heart malformation (75% of individuals with HOS)
- The congenital heart malformations most commonly observed are ostium secundum Atrial Septal Defect (ASD) and Ventricular Septal Defect (VSD)
- Cardiac conduction disease-Although individuals may present at birth with sinus bradycardia and first-degree atrioventricular (AV) block, AV block can progress unpredictably to a higher grade including complete heart block with and without atrial fibrillation.

Management includes evaluations at initial diagnosis to establish the extent of disease. Limb involvement is determined by physical examination. If limb involvement is not grossly obvious, upper-limb and hand radiographs can be performed to detect subtle anomalies of the carpal bones. Echocardiography is the procedure of choice to define the presence of septal defects or other structural cardiac anomalies. ECG is also recommended for the detection of cardiac conduction disease. The management of individuals with HOS optimally involves a multidisciplinary team approach, with specialists in medical genetics, cardiology, and orthopedics, including a specialist in hand surgery. The orthopedic team may be able to guide individuals in decisions regarding surgery for improved upper-limb and hand function as well as physical and occupational therapy options. Those individuals born with severe upper-limb malformations may be candidates for 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. Children with severe limb shortening may benefit from prostheses as well as from physical and occupational therapy.

Prenatal testing is important in pregnancies at risk-detailed high resolution prenatal USG detects limb & heart malformations.[4]

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