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A Review of Holt-Oram Syndrome

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Abstract:

A congenital heart deficiency, firstly atrium and ventricle septal deficiency, is related to skeletocongenital heart deformities of the upper limb in Hear-Hand syndrome, an autosomal controlling state. Skeleton deformities are bilateral and asymmetrical, and they only result in the arm in the preaxial radial beam diffusion. They reach from ulna hypoplasia and wrist abnormality to hypoplastic radii, nonexistent or digitalized thumbs, hypoplastic or absent first metacarpals, and clinodactyly. Cardiac action might range from minor structural flaws to asymptomatic conduction deformities. Atrium and ventricle septum deficiency are examples of structural flaws that are present in 75% of occasions seldom, more involved cardiac deformity similar to ventricular septum fault, endocardial protect deformity, a dual outlet right ventricle, and an entirely untypical pulmonary venous return are seen. It is rare to pick out an inter atrium septal aneurysm in infants. An atrial septal aneurysm is thought to be a straight cause of thrombus growth. But cyanotic congenital heart deficiency-related coagulopathy might also be a component. In addition to the typical skeleton deformity, our patient had an abnormal combination of compound cardiac injury (congenital heart defect, heart valve, and atrial septal defect), cardiac conduction deficiency, and a gerstmann's syndrome.

Keywords: Skeletocongenital heart deformity, skeleton deformity, new-born, congenital heart Deficiency, syndrome

INTRODUCTION

Anomalies in genotype and phenotype define heart hand syndrome, also popularly known Holt-Oram syndrome. The state is present from delivery in cardiac, conduction deformity, and morphological malformations of the upper limbs are examples of clinical characteristics. During the period 1960, Drs. Mary Holt and Samuel Oram observed a household with atrial septal deficiency (ASD) and a state present from birth in thumb deformities, which led to the beginning profile of Holt-Oram syndrome (HOS). One case per 100,000 births is the rate of this rare genetic order. Since then, descriptions of a few hundred cases have been made on a global scale. The T-box protein 5 gene is the only one that has been connected to Holt-Oram syndrome so a long way. According to research, 74% of human beings with Holt-Oram syndrome had a T-box protein 5 gene alteration. Although the mutation can be passed down via the family. Persons without a history of the condition tend to experience new mutations in the majority of cases.

CASE STUDY

A 23-year-old primigravida gave birth to a full-term (39+4 weeks) male newborn via delivery through her genitals. The baby weighed 2.345 kg and had an uneventful postnatal transit. His APGAR test result at 1 & 5 minutes after birth was a seven and an eight respectively. All maternal serology results were negative, and the newborn's phase was unremarkable. Parents had no prior history of consanguinity. The baby's length was 48 cm at birth, and the head measurement was 33.7 cm (noted after 24 hours of life). The two fontanelles were healthy. Every external orifice was visible. Within 24 hours, a newborn passed meconium and pee. Neonate had a left forearm upper limb malformation upon overall assessment. The skeletons of the left radius and left ulna were absent, and both hands had abnormally extended thumbs. The thumbs have triphalangeal bones. Supination and pronation were restricted in the left forearm. Both of the lower limbs were healthy. An examination from head to toe was normal. On the second day of life, a newborn started having breathing problems. Tachypnoea and subcostal oedema were discovered during the NICU examination. Enlarged in the liver, Levine grade 4/6 pan systolic murmurs, and bilateral crepitation were all discovered throughout the examination. In the beginning, the baby was given oxygen, antibiotics, diuretics, and digoxin because the baby might have had pneumonia or a cyanotic cardiac condition. RDS gradually subsided over the following three days. His radiograph on the left side showed a hypoplastic ulna and a missing radius. A cardiothoracic ratio of 0.67 on the posteroanterior view of the chest indicated cardiomegaly. Both C- reactive proteins and the total count of blood were within typical limits. The number of platelet cells is as usual. The thyroid profile was healthy. The KUB, USG abdomen, and neuro sonogram were all clear. On two-dimensional echocardiography, there were many small ostium secundum and an intramuscular hole in the heart that's present at birth (VSD) of 5 mm with the left to opposite side shunt. Atrial septal anomalies were discovered. A medical evaluation of Heart-Hand Syndrome was determined based on born with cardiac and forelimb defects; the mother and father had no previous experience of limb malformation or cardiac problems; we were unable to perform a genetic analysis due to financial restrictions and lack of resources in our institute; the newborn received care with furosemide, digoxin, and endovenous antibiotics; an orthopaedic consultation was carried out for limb deformities; and the newborn was kept on follow-up for cardiac imperfections

DISCUSSION

Heart-Hand Syndrome and Atrio-Digital Dysplasia are also termed as Holt-Oram Syndrome. While novel variations are responsible for 40–85% of cases, familial transmission accounts for the majority of cases. Holt-Oram Syndrome symptoms and physical results can vary widely, with the severity increasing with each succeeding generation. About 85–95 per cent of patients have heart abnormalities, and there is always upper limb deformity. The most prevalent congenital cardiac defects in HOS patients are ASD and VSD, while there are other forms as well. Other conditions include PDA, endocardial cushion abnormalities, left ventricular hypoplasia, or conduction issues. The severity of the cardiac and orthopaedic lesions determines the prognosis.

Differential diagnosis includes
1. Fanconi anaemia syndrome
2. The syndrome of thrombocytopenia with a missing radius
3. Okihiro syndrome
4. The syndrome of long-thumb brachydactyly
5. Association of VACTERL
6. Atresia of the radial rays.

Even if there are no clinical deformities, a family screening with hand X-rays and electrocardiograms should always be performed, and parents should receive counselling because it is a genetic illness with autosomal dominant inheritance and a high degree of penetrance. The major goal of management comprises physical therapy to at least partially restore function to the distorted upper limbs. It also centres on the early detection of cardiac problems to treat them and increase patient lifetime.

CONCLUSION

An uncommon condition known as Heart-Hand syndrome causes a variety of upper limb and heart abnormalities. This review's objective was to discuss the typical symptoms of Heart-Hand syndrome to seek any structural cardiac problems for early detection and management. These children can lead normal lives if cardiac and orthopaedic issues are treated promptly. Thus, heart problems should be checked in all neonates with upper-limb malformations. It is intended to manage that early.

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