# Heterotaxy Syndrome With Asplenia And Complex Congenital Cyanotic Heart Disease In A 38 Weeks Neonate: A Case Report

Arwa K. Alharthi [a], Ahmad S. Alharthi [b], Inas A. Saad [c].

**Abstract** — A full term baby girl born at 38 week of gestation, couldn't be weaned of oxygen in spite of timely continuous intervention, oxygen saturation did not raise above 85% .Echocardiogram , Chest x-ray and gastrographine was done. Diagnosed with heterotaxy syndrome with asplenia , situs inversus totalis and complex congenital cyanotic heart disease for future uni-ventricular repair.

**Index Terms**— Heterotaxy syndrome , Asplenia , Dextrocarida , Dextrogastria , Situs inversus.

## INTRODUCTION

It is rare condition where certain thoracoabdominal organs are arranged in a typical pattern.The term "heterotaxy" is from a Greek terms "heteros," meaning "other than," and "taxis," meaning "arrangement. Its broad term that includes many different congenital anomalies in different organ like: heart, intestines, spleen, lungs, liver and other organs (1). Here we report an unusual presentation of an infant with right sided cardiosplenic heterotaxy syndrome associated with complex cyanotic congenital heart defects.We are reporting this case as a rare and first case presented in Taif- Kingdom of Saudi Arabia.

## CASE REPORT

Full term baby girl born at 38 weeks of gestation of birth weight 2.770 KG as a product of Spontaneous vaginal delivery for an un-booked mother. ABGAR (Activity, Pulse, Grimace, Appearance, Respiration) score was 8 at 1minute and 9 at 10 minutes and neonatal Barden Scale score was 26 , cord gas was acceptable and she was routinely resuscitated .On physical examination the apex beat felt on the right side with ejection systolic murmur 3/6 over upper sternal border, mild tachypnea , HR > 100 bpm , oxygen saturation was around 90% . After one week patient couldn't be weaned of oxygen though started on Inderal 3 mg TID. On the third day she developed severe desaturation for which started Prostien 0.05mic/kg/m and Phenylephrine infusion , dose of Inderal was increased and later transferred to the neonatal intensive care unit for ventilator care. In spite of timely continuous intervention, oxygen saturation did not raise above 85% on MV 100% O2 and pulse 150bpm. Echocardiogram done on 1st few hours of life revealed complex heart disease comprising: Dextrocardia , Situs inversus ,Tricuspid Atresia , Large Atrial septal defect nearly coming atrium , hypoplastic Right ventricle and Ventricular septal defect , side by side great vessels with Aorta anterior to left , Pulmonary stenosis . Right sided aortic arch. Small closing PDA . Follow up studies showed progressive restriction of the VSD that compromised pulmonary flow and PDA increased on size upon prostein use. Chest X-ray was done and shows dextrocardia , dextrogastria , and higher position of NJT **(Fig.1)** and intestinal loops in the right regoin of the abdomen **(Fig.2).** On Abdominal Ultrasound spleen was not visualized on anatomical or extra anatomical sites and large liver about 8cm extending to left hypochondrium

a Medical Intern , College of Medicine , Taif University. Saudi Arabia
b Medical Intern , College of Medicine , Taif University.
c Consultant Pediatric Cardiologist , King Abdulaziz Specialist Hospital

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(Fig.4).Gastrographine meal follow through revealed :Dextro-position of the stomach seen at right hypochondrium . Inverted C-shaped duodenal loop seen to right side of the spines. Free flow of the contrast through stomach, duodenum and jejunum (Fig.3).Ultrasound imaging of the brain was normal. Further management for the patient is a referral to a higher center to underwent a cardiac surgery ( uni-ventricular repair ) .

## DISCUSSION

The term "heterotaxy" is from the greek words "heteros," meaning "other than," and "taxis," meaning "arrangement. Its abnormality in arrangement of thoracoabdominal organs. The incidence of heterotaxy syndrome more in males at ratio of 2:1 and is approximately 1:10000 births (1,2). There are two major types: the first one is called right atrial isomerism also called asplenia where there is two right sides with bilateral right atrial appendages and have no spleen ,this form is also associated with more severe heart defects and trilobed two lungs. Second type is left atrial isomerism which called polysplenia those patient have two left sides with isomeric left atrial appendages and multiple small nonfunctioning splenunculi and they are less likely to have mild heart defects or none at all (3). Most of the times 50 to 100% of patient diagnosed with heterotaxy syndrome present with congenital heart diseases during childhood (4). Presentation of heterotaxy syndrome is frequently associated with extracardiac abnormality like intestinal obstruction caused by gastrointestinal malrotation or volvulus , also they may have biliary atresia and abnormality of the renal tract. The radiological recognition of these patients is indicated and indispensable for planning of proper approach and evaluation of the a wide range of variants in each patient(5).Signs and symptoms of heterotaxy syndrome depend on the organs involve can include cyanosis , low immunity which lead to increased risk of infections, shortness of breath,digestion and feeding difficulty(6). However, in the recent brief literature very few cases have described heterotaxy syndrome with asplenia and complex cyanotic congenital heart disease. Abdur-Rahman et al reported a case of situs inversus totalis, asplenia and reverse rotation of intestine(7). We are reporting a case of a full term baby girl diagnosed with heterotaxy syndrome and asplenia , situs inversus totalis and complex cyanotic congenital heart disease which is rare case in Saudi Arabia and particularly in Taif city. Cardiac abnormalities are responsible for the Morbidity and mortality of these patients with heterotaxy syndrome. The prognosis of heterotaxy syndrome with asplenia is poor and usually lethal by congestive heart failure or severe infection (8). Echocardiogram and pulse oximetry screening of newborns is very important if we are suspecting congenital heart disease before discharge from the postnatal ward, regardless of the newborn health status ,so that clinically obscure congenital heart disease are detected early enough for early proper management and intervention(9).The suspicion of heterotaxy syndrome in our case was arisen due to low oxygen saturation of the neonate which was not improving despite prompt management, after doing radiological investigations and confirming the diagnosis, most important step is to increase survival of the patient and to referee her to a higher center to undergo cardiac surgery (uni-ventricular repair).

## CONCLUSION

Heterotaxy with Asplenia is a complex and rare anomaly which may present with severe congenital heart disease. Prompt early identification and timely proper management should be performed to prevent complications and to increase survival rate of these patient.

## FIGURES

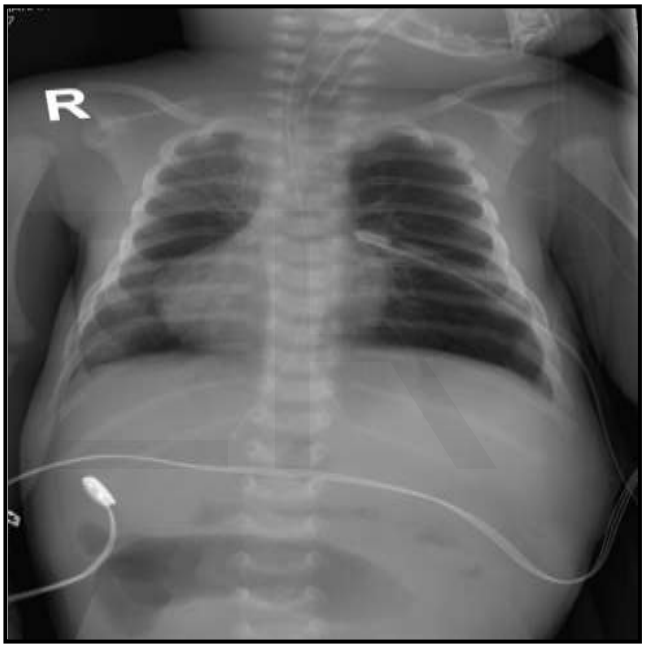


**Figure 1:** Plain X-ray Showing Dextrocardia Dextrogastria , Higher position of NJT

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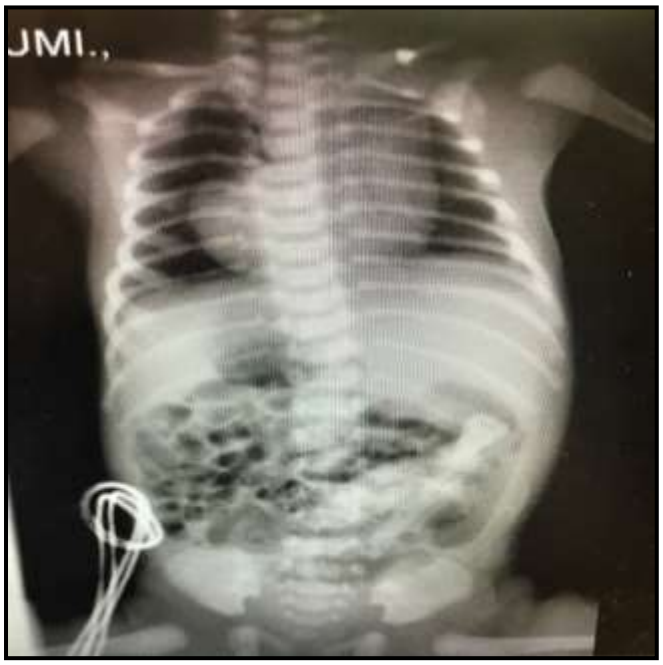


**Figure 2:** Plain X-ray Showing Dextrocardia and bowel loops in the right abdomen.

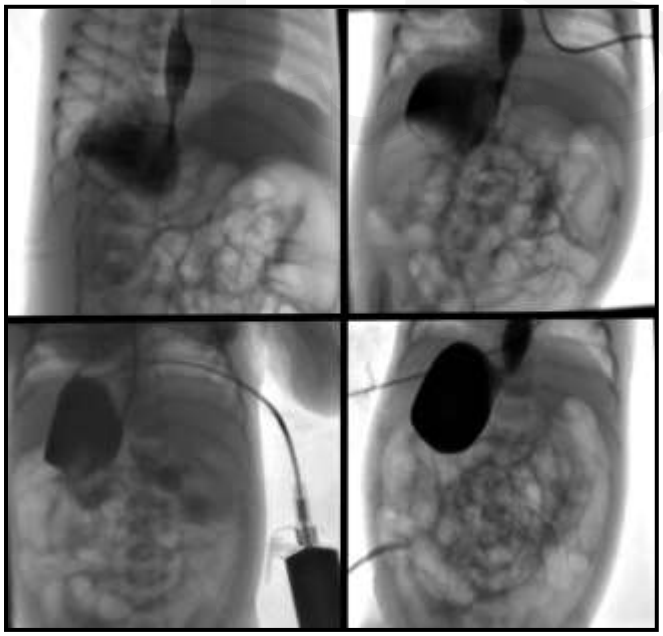

**Figure 3:** Gastrograffin meal flollow through confirmed dextro-position of the stomach, Inverted C shape duodenal loop

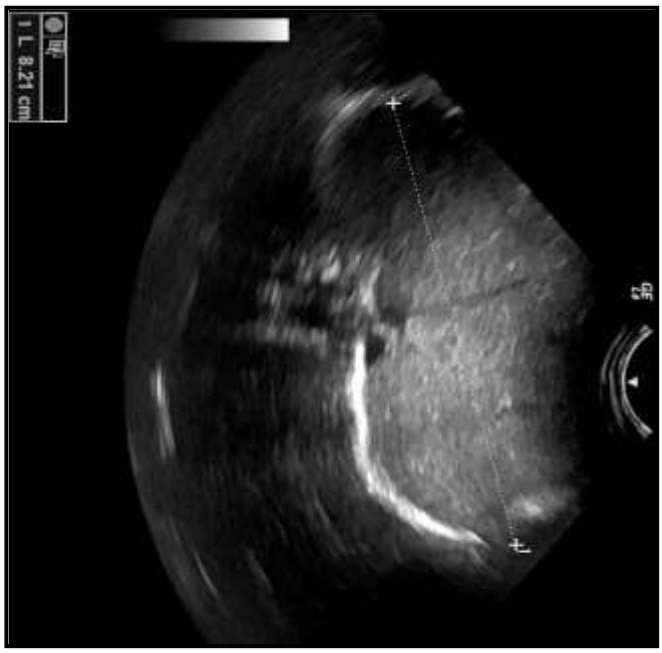


**Figure 4:** Abdominal Ultrasound shows large liver extending to the left hypochondruim and absence of spleen.

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