

Heterotaxy Syndrome with an Isolated Levocardia in Congenital Heart Disease; A Rare Case Combination in Tanzania

Theophylly Ludovick Mushi^{1*}, Naizihijwa Majani¹, Sulende Kubhoja¹, Frederic Lyimo⁴, Godwin Sharau², Godfrey Mbawala¹, Stella Mongella¹, Zawadi Edward¹, Neema Muro², Vivienne Mlawi³, Alexander Mroso¹, Abubakari Sudi² and Pedro K Pallangyo¹

¹Department of Pediatric Cardiology, Jakaya Kikwete Cardiac Institute, Tanzania

²Department of Pediatric Cardiac Surgery, Jakaya Kikwete Cardiac Institute, Tanzania

³Department of Pediatric Intensive care, Jakaya Kikwete Cardiac Institute, Tanzania

⁴Department of Radiology, Muhimbili National Hospital, Tanzania

*Corresponding Author: Theophylly Ludovick Mushi, Department of Pediatric Cardiology, Jakaya Kikwete Cardiac Institute, Dar es Salaam, Tanzania.

Received: January 19, 2019; Published: February 18, 2019

Abstract

Background: Heterotaxy syndrome is a rare congenital malformation usually present with atypical orientation of internal organs, complex cyanotic heart disease and multi-organs anomalies. It has high mortality among infants and early childhood.

Case Presentation: We are reporting a 2 years and 8 months old male in Tanzania, who at age of 6 months he suffered from severe respiratory distress, persistent cough and difficulty in breathing, excessive sweating and failure to thrive. A routine 2-D echocardiogram and CT scan cardiac described a rare congenital anomaly of Heterotaxy syndrome with an isolated levocardia, PDA and pulmonary hypoplasia.

Conclusion: The outcome of this complex is determined by early recognition and its associated cardiac anomalies. The diagnosis of such condition is incidental mainly identified during routine imaging hence high index of suspicion should be considered.

Keywords: Heterotaxy Syndrome; Isolated Levocardia; Pulmonary Hypoplasia

Abbreviations

PDA: Patent Ductus Arteriosus; MPA: Main Pulmonary Artery; RPA: Right Pulmonary Artery; LPA: Left Pulmonary Artery; CT: Computed Tomography; MRI: Magnetic Resonance Imaging

Introduction

Heterotaxy syndrome is a rare congenital malformation usually present with atypical orientation of internal organs may be associated with anomalies of cardiovascular, respiratory, gastrointestinal and urogenital anomalies [1]. It is estimated that one child in every 10,000 people in a population may have this condition. Nevertheless this condition is scarcely documented and believed to be under-diagnosed following uneven distribution of specialized cardiac centres equipped with diagnostic tools and expertise in Africa.

An incidence of this condition is 3 per cent for all congenital heart disease and is associated with dextrocardia [1,2]. Affected individuals suffer from failure to thrive, recurrent respiratory diseases and have significant morbidity and mortality. This article describes a child whose heart on its normal anatomical location with visceral organs abnormally arranged and malformed (Heterotaxy). This condition frequently presents with complex cyanotic heart disease, nevertheless our patient had "acyanotic congenital heart disease" (PDA) and pulmonary hypoplasia which contributed to severe respiratory distress. A study done in Spain for 20 years reported a mortality rate of patient of 47% for patients with congenital pulmonary hypoplasia within first 60 days of life and 75% for the first day of life [3]. This study describe a rare congenital anomaly of Heterotaxy syndrome with an isolated levocardia, PDA and pulmonary hypoplasia.

Case Report

A 2 years and 8 months old male delivered by SVD at term, birth weight 2800g from a 36 years old mother (P_4L_{3+1}) known type 2 Diabetic Mellitus (on metformin and glibenclamide for more than year) and hypertension on regular medication. A child was diagnosed to

have congenital heart disease at age of one month old. At 6 months of age he was admitted in our unit due to severe respiratory distress, persistent cough and difficulty in breathing, excessive sweating and failure to thrive. Had bluish discoloration on the periphery with no history convulsion; he is 4th child in family other siblings were doing well, no family history of consanguinity or congenital heart disease. He had features of congestive heart failure and was kept on furosemide 5 mg bid and spironolactone 3.125 mg orally.

On arrival to our department he was not dysmorphic, afebrile, not dyspnoeic, not jaundiced, no finger clubbing but had peripheral cyanosis, weighed (w) 5.5 kg, height (h) 61 cm (z score - 2). Blood pressure 101/54 mmHg, PR 111 b/min regular full and equal throughout, well perfused, RR 24 b/min, SPO₂ 78% in a room air, reduced air entry on the left, transmitted sound with rhonchi, Heart sound S₁ and S₂ with continuous murmur heard at left upper sternal border grade 2/6 and liver was palpable on the left side.

Chest x ray was done showing homogeneous opacification on the left hemithorax, hilum shadows and filling in of the aortopulmonary window, both costophrenic angles were normal, Heart size appears enlarged. Trachea is central and carina angle is normal, visible osseous structures in the thoracic region are normal.

2D-Echocardiogram: Situs inversus, levocardia, aorta on the anterior and inferior vena cava with azygous continuation, posteriorly, dilated left atrium, no atrial septal defect or ventricular septal defect, patent ductus arteriosus 6 mm left to right shunt, normal coronaries in course and origin, three cuspid aortic valves, left aortic arch and no coarctation of the aorta. Normal pericardium and no pericardial effusion.

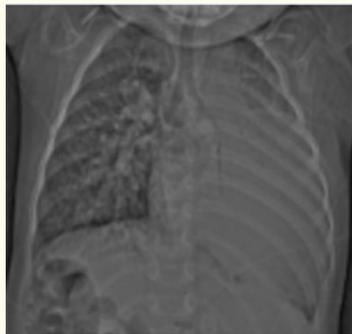


Figure 1: Chest x ray after PDA ligation when the child developed severe pneumonia. There was consolidation at the right hilum and lower lobe (post-PDA ligation).

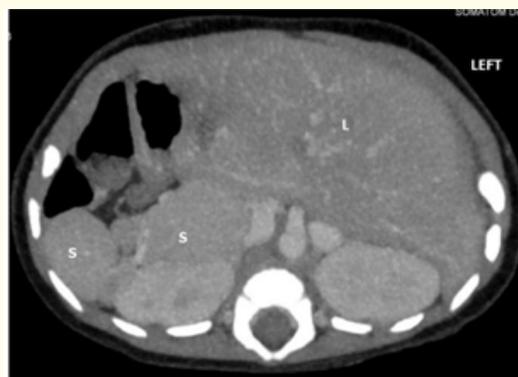


Figure 2: CT scan axial view shows liver on the left side, both stomach and two spleens are on the right.

CT scan of chest and abdomen a coronal reformatted view

Situs inversus, levocardia (apex pointing to the left); Interrupted inferior vena cava with azygous continuation, massive cardiac enlargement with multi-chamber involvement, Hypoplasia of left lung parenchyma, small volume on the left lung, No left diaphragmatic

hernia seen. Liver seen on the left side, both stomach and two spleens were on the right side. Stomach, 2 spleens and small intestine both are located on the right side and liver and large intestine on the left (Lt) side and liver on the right side.



Figure 3: a) Chest + abdominal CT scan coronal reformatted. b) 3-Dimension of the chest and abdomen.

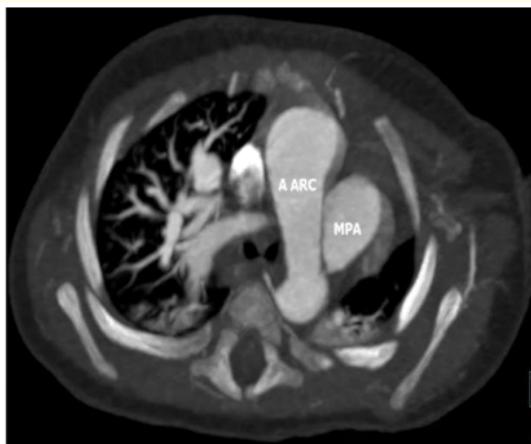


Figure 4: CT scan axial view shows right lung is normal with good size MPA, hypoplasia of the left lung, ascending aorta (Ao) 2.26 cm, Descending aorta 1.29.

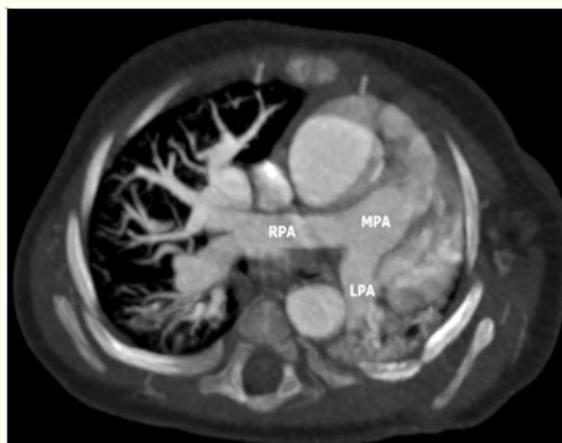


Figure 5: CT scan axial view shows main pulmonary artery (MPA) is 16.1 mm and good size left (LPA) and right pulmonary artery (RPA).

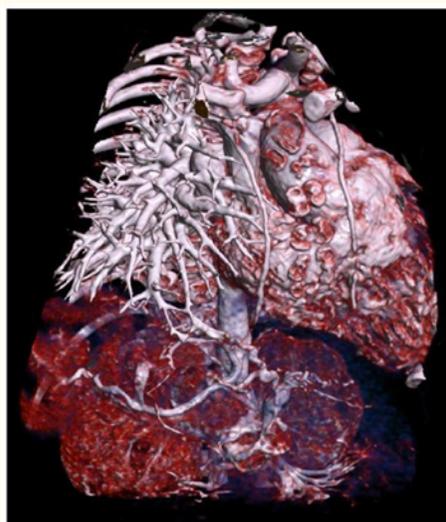


Figure 6: A 3D showing vascular supply of the right lung more than on the left side, an enlarge heart with Liver seen on the left side.

Discussion

Our child had multiple congenital defects but developed features of congestive heart failure due to large PDA and hence surgical ligation was performed at the age of 6 months. He is now 2 years and 7 months old of age doing well to date and other defects were managed conservatively. The disorderly arrangement of his internal organs of which liver, gall bladder were on the left side, while stomach, small intestine were located on the right side, with underdevelopment of left lung. Such condition is known as “Heterotaxy” [4].

The term “Heterotaxy” is a Greek words “hetero” means other than and taxis “arrangement”. In this condition internal organs are abnormally arranged in the chest and abdomen [5]. Normally, right side organs include liver, gall bladder, tri-lobed lung and inferior vena cava and left side organs include stomach, one spleen, bi-lobed lung and aorta which is a normal orientation situs solitus; and if the apex of the heart is in its normal position is termed as levocardia. When internal organs are disorderly arranged then if the right atrium develops on left side and left atrium on the right side this condition is described as situs inversus. Studies have shown Heterotaxy syndrome is associated with dextrocardia and complex cyanotic heart disease with high mortality during infancy and early childhood, but our patient had isolated levocardia with pulmonary hypoplasia (left) and acyanotic heart disease (PDA) [6-8].

The exact cause of situs inversus with isolated levocardia is unknown [9]. A number of risk factors has been indicated, Genetic predisposition of which Kartagener syndrome a ciliopathic, autosomal recessive genetic disorder affecting cilia lining respiratory tracts, however, most of studies suggest of this condition being associated with dextrocardia unlike our patient who had isolated levocardia [10]. There is familial history of congenital heart disease and his brother died at the age of 1 month old due to CHD. Her mother is known to be diabetes on regular medication.

He had polysplenia (left isomerism) with interrupted IVC and azygous continuation, which is similar to studies reported elsewhere [11]. Right lung was completely developed, well vascularised, with good lung volume and tri-lobed but the left sided lung is hypoplastic, reduced in size, very few blood vessels.

Conclusion

The outcome of this complex is determined by early recognition and associated cardiac anomalies. The diagnosis of such condition is incidental mainly identified during routine imaging.

Recommendation

Clinician should have an index of suspicions following a long standing history of respiratory distress for lung pathology and features of heart failure, use of diagnostic tools such as ECHO, X- ray, CT scan and MRI should be indicated for detailed evaluation of such condition.

Author’s Contributions

TLM wrote initial draft of the manuscript, also together with TLM, GS, SM, VM, NM, AM, AS, PP, ZE, GM took history, and performed physical examination. NM, SK, MJ performed echocardiography; FRL interpreted the all CT scan images. All authors reviewed and contributed to the final version of this case report. All authors read and approved the final manuscript.

Ethical Approval and Consent to Participate

Ethical clearance was granted from the directorate of research of the Jakaya Kikwete Cardiac Institute.

Consent for Publication

Written informed consent was obtained from patient's legal guardians for publication of this case report and any accompanying images.

Acknowledgement

The authors wish to thank Prof. Mohammed Janabi, Executive Director and his entire management team of JKCI, Directorate of Cardiology, Directorate Clinic supports, Dept. Research, Training and Publication, as well as the patient, who promptly agreed to make the images available for the present case report.

Bibliography

1. Heterotaxy syndrome. Genetics Home (2016).
2. Heterotaxy syndrome (2019).
3. Yanny Paola Delgado-Peña, *et al.* "Pulmonary hypoplasia: An analysis of cases over a 20-year period". *Anales de Pediatría* 85.2 (2016): 70-76.
4. Kim SJ. "Heterotaxy syndrome". *Korean Circulation Journal* 41.5 (2011): 227-232.
5. Daniel SC., *et al.* "Heterotaxy syndrome: a case report". *Radiologia Brasileira* 46.3 (2013).
6. Wang Xiaofang, *et al.* "Comparing levocardia and dextrocardia in fetuses with heterotaxy syndrome: prenatal features, clinical significance and outcomes". *BMC Pregnancy and Childbirth* 17.1 (2017): 393.
7. Wolla CD., *et al.* "Cardiovascular manifestations of heterotaxy and related situs abnormalities assessed with CT angiography". *Journal of Cardiovascular Computed Topography* 7.6 (2013): 408-416.
8. Jacobs JP, *et al.* "The nomenclature, definition and classification of cardiac structures in the setting of heterotaxy". *Cardiology Young* 17.2 (2007): 1-28.
9. Jo DS., *et al.* "A case of unusual visceral heterotaxy syndrome with isolated levocardia". *Korean Circulation Journal* 43.10 (2013): 705-709.
10. Pandit S., *et al.* "A rare case of Kartagener's syndrome". *Journal of Natural Science, Biology and Medicine* 5.1 (2014): 175-177.
11. De Paola N., *et al.* "Prenatal diagnosis of left isomerism with normal heart: a case report". *Journal of Prenatal Medicine* 3.3 (2009): 37-38.

Volume 6 Issue 3 March 2019

©All rights reserved by Theophylly Ludovick Mushi, *et al.*