CASE REPORT

A variant of pentalogy of cantrell in a live birth

HU Okafor, Tagbo Oguonu, SN Uwaezoke, BC Anusiuba[1]

Department of Paediatrics, [1]Internal Medicine, College of Medicine, University of Nigeria Teaching Hospital Enugu, Nigeria.

Abstract

Pentalogy of Cantrell which usually comprises of anomalies of the ectodermal and mesodermal tissues is a very rare congenital condition which in the extreme of cases is incompatible with life. In this report a variant of the condition in a live newborn baby who presented to the University of Nigeria Teaching Hospital with abnormalities of the heart, urinary bladder and sternum is discussed. The aim of the report is to create an awareness and high index of suspicion amongst health professionals

Key words: Pentalogy, cantrell, livebirth

Date of Acceptance: 14-Jun-2010

Case Report

A 5-day-old full-term male neonate of a grand multiparous woman was presented to the pediatric outpatient clinic of the University of Nigeria Teaching Hospital Enugu, Nigeria on the 27th of January 2004.The complaints were an anterior chest wall defect noticed at birth associated with anterior chest wall in-drawing on breathing. The pregnancy was carried to term but the mother had a febrile illness at 8 weeks gestation which was not associated with a rash and was assumed to be malaria which was treated appropriately. There was no history of ingestion of neither herbal or other unorthodox medications nor exposure to ionizing radiation during the pregnancy. She only received hematinics. Delivery took place at a local secondary health facility and was by spontaneous vertex delivery. There was no history of congenital anomalies in other siblings. There was no history of immediate postnatal problems. Patient was brought to the hospital on self-referral for the observed abnormalities. Parents were of the lower socio-economic class, social class four using Oyedeji's classification.[1] Examination revealed a fully conscious active baby that weighed 2.8 kg with a crown-heel length of 52cm, and occipito-frontal head circumference of 33cm. All anthropometric measurements were appropriate for age. Chest examination showed a diamond-shaped mid-line chest wall defect with a membranous base, hypopigmented and discharging sero-fibrinous fluid from the center. It measured 7 cm (length) and 5 cm (width) and was surrounded by a border of hyperpigmented skin. A hyperpigmented line joined the inferior pole of the defect to the umbilicus [Figure 1]. There was deep retraction of the chest wall on inspiration. Physical examination of other systems did not reveal any abnormality. However, on investigation, chest radiography revealed an absence of the sternum, while ultrasonography showed a urinary bladder which extended superiorly through a narrow hypoechoic path (believed to be an urachal diverticulum). Echocardiogram revealed a situs solitus with a secundum atrio-septal defect [Figure 2]. Other abdominal organs were in their normal visceral sites and were normal. The child was in the hospital for 2 weeks before the parents insisted on discharge because at the time he looked apparently healthy. The eventual outcome could not be determined in this patient as he was lost on follow-up.

Discussion

We have reported a case of a child with multiple congenital defects involving the ectodermal and mesodermal tissues. The presence of a congenital heart defect, abnormal position of the urinary bladder with the absence of the sternum (sternal defect) will suggest a variant of Cantrell syndrome. Pentalogy of Cantrell also referred to as

Address for correspondence:
Dr. HU Okafor,
Department of Paediatrics,College of Medicine,
University of Nigeria Teaching Hospital Enugu, Nigeria,
E-mail:huche57@yahoo.com

Access this article online

Quick Response Code:

Website: www.njcponline.com

DOI: 10.4103/1119-3077.79257

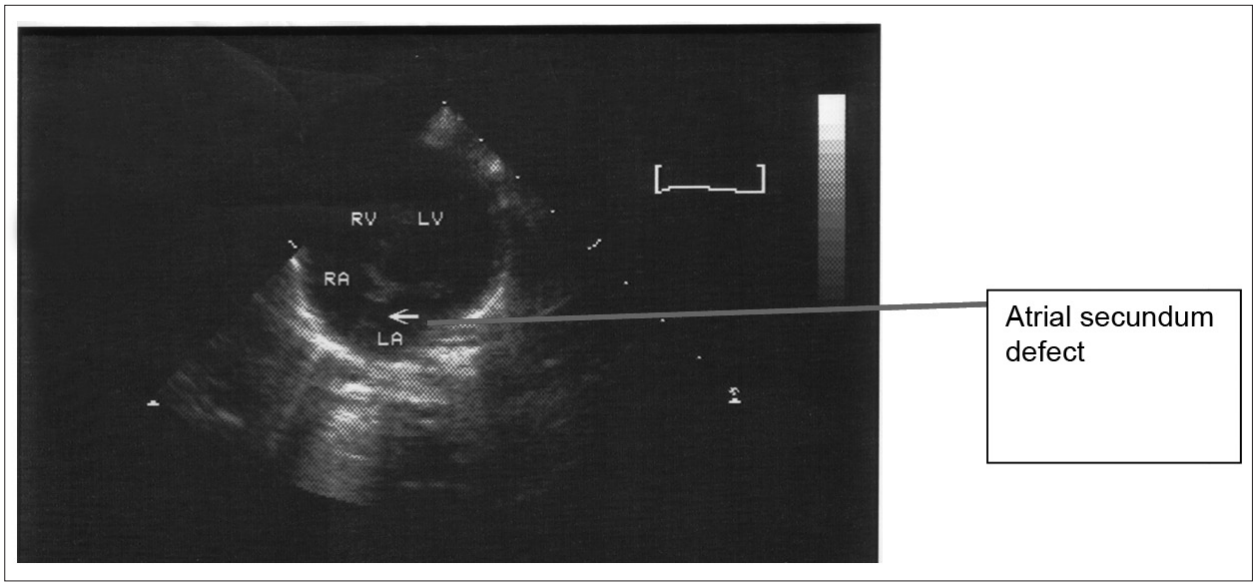

Figure 1: Echocardiogram of patient

Figure 2: Photograph of patient

Cantrell syndrome or Cantrell Haller-Ravich syndrome was first described by Cantrell in 1958.[2] He reported a case with cleft lower sternum, high omphalocele, anterior diaphragmatic hernia, pericardial defect and congenital heart anomaly. Cantrell, Haller and Ravich later described the syndrome of congenital defects of the abdominal wall, sternum, diaphragm, pericardium and heart.[3] Since then, variants of the syndrome with various combinations of the defects have been reported. Diagnosis of the complete syndrome requires the five criteria described by Cantrell, but incomplete variant forms exhibiting three or four of the features have been described.[4] The syndrome is said to be very rare and associated with very poor prognosis, hence most diagnoses are made post-mortem. In a review of the literature in 1972, Toyama reported a survival rate of 20%. This observation included cases with mild defects and incomplete expressions of the syndrome, and all cases were diagnosed after delivery.[4] Its reported incidence is less than 1 in 100,000 live births. It affects both males and females. However, strictly applying the definition of the syndrome will ordinarily exclude our index case. But as mentioned above, there are many variants which have been reported in the literature. For instance Liang *et al*,[5] reported a case diagnosed prenatally at 10weeks of gestation while Lutfu *et al*,[6] reported a case also diagnosed prenatally at 12-weeks gestation. In both cases diagnosis were made initially by ultrasonography and confirmed at autopsy. The pregnancies were terminated following prenatal diagnosis and counselling. In the patient reported by Lutfu *et al*, there was an associated chromosomal abnormality, trisomy 21, this apparently contributed to the decision to terminate the pregnancy. Usually, if a diagnosis is made by ultrasound, chromosomal analysis is always recommended. Associations with trisomy 18, trisomy 13 and Turner syndrome have been reported.[7,8] In our patient, diagnosis was only made after birth and he had no obvious systemic problems except for the defect in the anterior chest wall. Generally there are few reports of this condition in the literature most being diagnosed either prenatally or post-mortem.

Combination of deformities observed in this child is distinct from the classical Cantrell although similarities are shared. A notable feature of this case is the child's age at presentation and duration of stay in hospital without a significant threat to his life by the deformities. In the literature, Cantrell's pentalogy is associated with very poor prognosis hence the need for prenatal diagnosis. When related to the index patient it might be surmised that there may be various degrees of severity and in some cases the defects may be compatible with life despite the overall impression of poor prognosis. Three of the five patients Cantrell reported in 1958 survived, but none of the five had true ectopia cordis. Despite the overall poor prognosis, survival may be related

to the extent of the ventral wall, sternal, and cardiac defects. This may explain our patients survival up to 5 days. There has been no report so far from this part of Nigeria, so we have reported a live birth with the combination of features suggestive of Cantrell's pentalogy. The aim is to create awareness amongst health practitioners in our environment.

Acknowledgment

We wish to acknowledge the contributions of the academic staff of the departments of radiology and cardiology of the College of Medicine University of Nigeria.

References

1. Oyedeji GA. Socio-economic and cultural background of hospitalized children in Ilesha. Niger J Paediatr 1985;12:111-7.
2. Cantrell JR, Haller JA, Ravitch MM. Syndrome of congenital of congenital defect involving abdominal wall,sternum,diaphragm,pericardium and heart. Surg Gynecol Obst 1958;107:602.
3. Magnuson DK, Schwartz MZ. Pentalogy of Cantrell. In: Oldham KT, Columbani PM, Foglia RP, eds. Surgery of Infants and Children: Scientific Principles and Practice. Philadelphia,Lippincott-Raven; 1997:878.
4. Toyama WM. Combined congenital defects of the anterior abdominal wall, sternum, diaphragm, pericardium, and heart: A case report and review of the syndrome. Pediatrics 1972;50:778-92.
5. Liang RI, Huang SE, Chang FM. Prenatal diagnosis of ectopia cordis at 10weeks of gestation with two-dimensional and three-dimentional ultrasonography. Ultrasound Obstet Gynecol 1997;10:137-9.
6. Onderoğlu L, Baykal C, Tulunay G, Talim B, Kale G. Prenatal diagnosis of Cantrells pentalogy. Turk J Paediatr 2003;45:4:1-2.
7. Fox JE, Gloster ES, Mirchandani R. Trisomy 18 with Cantrell Pentalogy in a Stillborn Infant. Am J Med Genet 1988;31:391-4.
8. Bryke CR, Breg WR. Pentalogy of Cantrell. In: Buyse ML, editor. Birth Defects Encyclopedia. Boston: Blackwell Scientific Publications; 1990. p. 1375-6.

Source of Support: Nil, **Conflict of Interest:** Nil.