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Complete Endocardial Cushion Defect in the Presence of down Syndrome in a Nigerian Child: A Case Report

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Abstract: Down syndrome is the most commonly recognizable chromosomal anomaly with many associated complications. Though many of these have been documented in the Nigerian child but it thus appears to the best of the knowledge of this author that the cardiology complications have been scarcely reported. Thus, the need to report a case of completed endocardial cushion defect in the presence of trisomy 21 in a Nigerian child and make a case for routine echocardiograph in all clinically diagnosed trisomy 21 for the purpose of early intervention.

Key words: Endocardial cushion defect, down syndrome, congenital heart disease

INTRODUCTION

Down syndrome is the most common and easily recognizable chromosomal anomaly found in children. It is seen in 1-1.94 per 1000 live birth in Africans (Verma *et al*., 1992; Adeyokunnu, 1982; Deiport *et al*., 1995) and commonly associated with endocardial cushion defect (ECD). In a study ECD accounted for 43% of congenital heart disease (CHD) amongst children with down syndrome, followed by ventricular septal defect which accounted for 32% (Park *et al*., 1977). Most reported cases of ECD have been from developed countries while to the best of the knowledge of this author none has been reported from Nigeria in a patient with Down syndrome. In view of this, we report a case of complete ECD in a down syndrome patient with the aim of sensitizing and raising the index of suspicion of the medical community to the presence of this CHD and the need for routine echocardiography for all newborn with down syndrome for the purpose of early diagnosis and intervention.

CASE REPORT

Patient O.D is a 3 month old male infant, who was brought into the emergency unit of the hospital on account of difficulty in breathing, fever and grunting of one day duration. Mother denied history of aspiration and force feeding of the child. Child was on exclusive breastfeeding but will suck intermittently. Patient had a history of prolonged jaundice in neonatal period with no clinical evidence of a heart disease. There was a history of recurrent upper respiratory tract infections requiring frequent hospital visits prior to presentation. Patient is the second child of a 28 year old mother and a 34 year old father. Mother's antenatal history was not adversely affected.

Examination showed an infant with a mongoloid facie in severe respiratory distress, febrile (39°C), acyanotic, pale, anicteric, flaring alae nasal with no significant lymph node enlargement. Respiratory rate was 72 cycles min^{-1} with intercostal and subcostal recession. Heart rate was difficult to count because of gallop rhythm and there was no mummur. Liver was 3 cm enlarged, soft and tender, spleen was not enlarged. Patient had no neck control and was hypotonic, Anterior frontanelle was patent and normotensive, Head circumference was 41 cm, normal deep tendon reflex. Musculoskeletal system was essentially normal. An assessment of bronchopneumonia in congestive cardiac failure to rule out CHD in a patient with suspected trisomy 21 was made.

Investigations revealed pack cell volume of 43%, total white blood cell of 3700 mm^3, Neutrophil of 43%, lymphocyte of 57%. Malaria parasite was nil on peripheral blood film. Blood culture could not be done because the facility was not available. Chest x-ray showed cardiomegaly with cardiothoracic ratio of 0.68, mild biventricular enlargement, left sided aortic arch and hazy abnormal consolidation in right mid zone. A repeat chest x-ray was done on the 7th day of admission because of persistent fever and mild tachypnoea which did not differ from the first x-ray. In view of these findings an echocardiogram was done which showed a posterior wall thickness of 0.75 cm, left atrial diameter of 1.2 cm, right ventricular anterior wall thickness of 0.6 cm, aortic root of 0.070 cm with complete ECD and single atrioventricular valve (measuring 1.5 cm).

Child was admitted and put on oxygen at 2 L min^{-1}, IV 1 mg kg^{-1} of as Frusemide, rapid digitilization with IM Digoxin 0.02 mg kg^{-1} stat then 0.01 mg kg^{-1} 8 h × 2 doses and intravenous antibiotic with cefuroxime and aminoglycoside (Genticin). On the 7th day of admission, patient was discharged home to continue with daily Digoxin, Frusemide and Cefuroxime. At the point of discharge patient was active, sucking well and no longer in respiratory distress, his heart rate was 150 beats min^{-1} and he had no murmur. In the course of follow-up, patient had another episode of pneumonia without congestive cardiac failure. He was treated in the out-patient clinic with oral antibiotics with good recovery. Unfortunately, patient was brought in dead when arrangement was almost concluded for him to travel to South Africa for surgical repair of the heart defect.

RESULTS AND DISCUSSION

ECD, also known as atrioventricular septal defect is a CHD commonly seen among Down syndrome patient (Grech, 1999). There are 2 types of the disease: the partial type consisting of atrial septal defect with both atrioventricular valves while the complete type consists of atrial septal defect, ventricular septal defect and a single atrioventricular valve (Cooper, 2004). Studies have shown the complete type to be more common and usually associated with Down syndrome which was the case in this report. Mothers whose ages are below 32 years have been reported to have a higher risk for the development of CHD in down syndrome (Chaohab *et al*., 2007). This assertion is supported by the fact that the mother of the patient in this case was 28 years old.

Patients with ECD, particularly the partial type may be asymptomatic at birth, infancy, adolescence and may not develop congestive cardiac failure until adulthood while the complete type is associated with early manifestation of heart failure as was the case in this patient who presented at age 3 months. Patient also presented with recurrent upper respiratory infection and pneumonia which are known complications of ECD. The presence of these complications in Down syndrome patient was corroborated by Zhonghua in his postmortem findings as the leading cause of death (Hou *et al*., 1989). The complications of CHD, Congestive cardiac failure and pneumonia may have been responsible for the death of this patient though parents did not consent to post mortem after the child was brought in dead. This underscore the low acceptance level of post mortem in our environment partly because of socio-cultural reasons.

The delay in diagnosis of this condition in the patient is not unconnected with the fact that clinical examination at birth failed to support the need for echocardiography assessment as this procedure was not routinely done in our hospital and many other hospitals in Nigeria. Where it is indicated, there is the challenge of cost as most patients cannot afford to pay for the procedure. This finding was corroborated by Bhatia *et al*. (1992) who studied CHD amongst 50 children with chromosomally proven down syndrome using clinical examination, chest x-ray, electrocardiography and Echocardiography and discovered that 3 children had normal clinical examination, despite an abnormal echocardiography and concluded that clinical examination was not sufficient to rule out heart disease in patients with down syndrome.

An unexplained interesting finding in this patient was the absence of a murmur at birth, follow-up clinic, period of illness and post illness period as the author was opportuned to have managed the patient from birth till demise. It was unfortunate that postmortem was not done as this could have assisted in explaining this unusual clinical finding in the child.

CONCLUSION

In conclusion, congenital heart disease is an important cause of mortality in patients with down syndrome and as such routine echocardiography assessment of all clinically diagnosed newborn with down syndrome is advocated for the purpose of early surgical intervention where needed.

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