

Edwards Syndrome In A Neonate From A Developing Country; Reasons For Concern: A Case Report

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Abstract

We report the case of a baby boy who was brought by the parents to a Nigerian teaching hospital on the 4th day of life. He had multiple congenital anomalies, the features of which were consistent with Edwards syndrome. The challenges of managing such a child are hereby discussed.

INTRODUCTION

Edwards syndrome is the second most common trisomy. It has been reported as very rare among Nigerians. A previous study reported a prevalence figure of 0.08 per 1000 live births. ¹ The outcome is usually fatal, even in the best of centres. We report the case of a four-day-old neonate with clinical features consistent with Edwards syndrome who was managed at the special care baby unit of the Ladoke Akintola University of Technology Teaching Hospital, Osogbo, South western Nigeria. The parents unexpectedly, requested for discharge against medical advice after 20 hours of hospitalisation. The aim of this article is to highlight the challenges associated with managing the child, the parents and the problems posed by the Nigerian health care system.

CASE REPORT

AB, a four-day-old male baby, was delivered on 1st of April 2006, at a rural local government primary health centre, approximately 30 kilometres from the State Hospital, Osogbo. The staff nurses on duty at the health centre took the delivery of the baby and immediately advised the parents to take the baby to the Ladoke Akintola University of Technology Teaching Hospital, Osogbo. They did not write a letter because they felt that the grotesque anatomical features of the baby would cause immediate social and management problems and that such a baby needed immediate transfer from that location, to a teaching hospital centre. The parents however felt there was no immediate life threatening danger and so presented on the fourth day of life.

The mother a 27-year-old petty trader, booked at the antenatal clinic at an estimated gestational age of 3 months.

There was no previous history of abortion. Attendances at the antenatal clinics were good and all consultations were with nurses. Pregnancy was not adversely eventful and there was no history suggestive of infections, exposure to radiations or use of any other drugs apart from the prescribed routine haematinics. There was no history suggestive of polyhydramnios and uterine ultrasound of the foetus was not done. Labour and delivery were normal and the baby breathed and cried immediately.

The parents of the baby are muslims in a monogamous family setting. The father is a 35-year-old automobile spare part-seller; studying physics in a teachers' college, while the mother is a 27-year-old senior secondary schools drop out. The patient has three elder siblings, aged 8, 6, and 3 years who are all well, and have no similar disease history.

On examination, the baby had features of a term, low birth weight baby, weighing, 2.4kg. He was acyanosed in room air and had a rectal temperature of 36.5°C. The skull was narrow with an elongated bi-frontal diameter and a prominent occiput. In addition the ears were low set and micrognathia was present. The central nervous system examination revealed a small head with a circumference of 28.5cm, a small patent anterior fontanelle, normal muscle tones, deep tendon and sucking reflexes. The limbs revealed hyper-extension deformity at the knee, characteristic clenched hand posture with overriding fifth and middle finger (camptodactyly), syndactyly of the second and third digits, single transverse palmar crease, rocker-bottom feet with prominent calcanei and talipes equinovarus deformity of the ankles. No abnormality was found in the abdomen except

that the rectum terminated in a patulous anus. However, physical examination of the spine and back showed no defect. Also, no abnormal physical findings were detected in the cardiovascular and respiratory systems. Two different picture views of the baby, displaying some of these features are shown in figure 1 and 2.

Figure 1



Figure 2



An assessment of Edwards syndrome was made and the patient was immediately admitted to the special care baby unit. Nasogastric tube feeding was ordered but this was changed to feeding by cup and spoon, because the passage of the feeding tube was provoking apnoeic attacks. The investigations ordered include, full blood count, chest and skeletal radiographs, echocardiography of the heart, ultrasound of the abdomen and karyotyping.

Barely after 20 hours of hospitalisation the father of the child requested for the baby's discharge (against medical advice) after being overwhelmed by the hospital bills. The only investigation that had been paid for and done so far was the packed cell volume, which was reported to be 55%. The prospects of waiving the total bills accruing from the admission and further management were being considered and this was fully discussed with the parents. As part of the plan, the radiologist offered to pay for the radiological investigations. However, the father refused all the overtures and help planned and the baby was eventually discharged against medical advice to the parents. As at the time of discharge, the parents had no plans for managing the baby, but the father promised glibly that the baby would be well cared for and brought for review in one-week time. A week later the parents failed to show up at the clinic and all attempts to contact them failed.

DISCUSSION

Edwards syndrome or trisomy 18 was first described in 1960 by Edwards et al. ² An aberration of the chromosomes, specifically autosomal trisomy 18 is the cause. In most cases the disease arises as a result of non-disjunction and the affected individual usually has a regular trisomy 18. Rarely the aetiology is due to translocations and mosaicism. The phenotypic expression of the disease is most severe in the regular non-disjunction and fairly mild in the translocations and mosaics types. Our case is not true to pattern in terms of the sex, absence of symptoms and signs of cardiac defects, survival for the first 4 days without special care and relatively young maternal age. Perhaps the increased paternal age and a possible trisomy variant may explain some of these phenotypic expressions. The Pena shoiker syndrome or phenotype, in which there is an absence of flexion creases on the hands and fingers is also a possible differential. ³ Karyotype analysis is therefore necessary in diagnosis and classification of chromosomal disease and this ultimately determines the prognosis. ^{1,4} Other associated findings in patients with Edwards syndrome include severe psychomotor retardation, microencehphaly, micrognathia,

arthrogryposis, rocker bottom foot with prominent calcanei and characteristic hand posture. ⁵

The antenatal delivery and postnatal management of a baby with this kind of defect presents enormous challenges to a health system such as ours. A study among the inhabitants of Tunisia suggested the promotion of cyto-genetic prenatal diagnosis through, education, genetic counselling and fetal ultrasound as strategies of preventing handicapping conditions like trisomies. ⁶ However, foetal ultrasound was not done on this baby and opportunities to diagnose some of the abnormalities may have been missed.

Discharge against medical advice by these parents may partly be due to the financial constraints and partly psychological stress from nursing a physically abnormal baby. It is virtually impossible for this parent who could not pay the bills for investigation to afford the cost of correcting any of the abnormalities. Previous studies in this region have shown that discharge against medical advice is a common decision taken by parents of children having congenital and other serious disease, which may culminate in a fatal end. ^{7,8} The discharge against medical advice provides an escape to the social embarrassment posed to this parent on the open ward. Management of the patient at home therefore allows the parents to enforce their rights to privacy, which paradoxically puts the baby at a potential risk of infanticide.

Significant development in the social, economic and health systems in Nigeria and similar countries will be needed, before this baby can be correctly managed. Such developments should include substantial improvements in the organisation and funding of the health and social welfare

systems. This will ensure that needy patients and their relatives can be relieved of the expenses associated with health care services and assist health care personnel to provide better health service including antenatal screening and other tests and appropriate management.

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