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PRUNE BELLY SYNDROME: CASE REPORT
Mary Kiria Koigi, Senior House Officer, Department of Obstetrics and Gynaecology, University of Nairobi; Koigi Kamau, Associate Professor, Department of Obstetrics and Gynaecology, University of Nairobi.

Corresponding Author: Dr. Mary Kiria Koigi, Department of Obstetrics and Gynaecology, College of Health Sciences, School of Medicine, University of Nairobi. Email: kiriakoigi@gmail.com

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M. K. Koigi and K. Kamau

SUMMARY

Despite typical gross features of Prune Belly syndrome, this case posed immediate diagnostic challenge at birth, depicting paucity of comprehensive knowledge on congenital malformations. Prenatal ultrasound had not been done. Neonatal death was due to the malformations and birth asphyxia. Inadequacies in management of congenital malformations and providing support systems to the parents also emerged. A need for screening and training on prenatal anomalies becomes obvious. In addition, psycho-social and medical support systems should be put in place for the parents in order to enhance preparedness in care.

INTRODUCTION

Prune Belly syndrome (PBS) is a rare congenital disorder affecting about 1 in 30,000 births, predominantly affecting males[1,2]. Although the aetiologic basis is not clear, the possibility of genetic inheritance has been proposed[3]. Observed prostatic defects and urethral obstruction may be due to hindered growth and development as a result of destruction or absence of appropriate primitive mesenchyme during embryogenesis[4]. Whatever the mechanism of causation, the end result is typical features characterized by lack of abdominal muscles causing flabbiness and wrinkling of the skin over the abdominal wall, undescended testes and urinary tract malformations. This may be complicated by multi-systemic involvement such as pulmonary, renal, cardiac, skeletal and gastrointestinal [5]. When severe, perinatal mortality is invariable[5, 6]. When prenatal diagnosis is made, termination of pregnancy may be an option if the degree of damage is deemed incompatible with life[7]. However, if prenatal diagnosis is not made and the mother is therefore not prepared for this outcome, the experience can be devastating. This case report therefore aims at looking into the need for both prenatal preparedness and postnatal counselling of such outcomes.

CASE REPORT

The mother was a 24-year-old Para 2 Gravida 3 waitress whose second delivery was a stillbirth via caesarean section due to abruptio placenta

complicated by massive haemorrhage. Although the gestation of the index pregnancy was unknown, the uterine size was corresponding to a term gestation. There was no previous history of congenital malformations. She had received at least one dose of depot medroxyprogesterone acetate (DMPA) contraceptive before she realised that she was pregnant.

She had made four prenatal visits in a nearby City Council clinic during which she was given the routine prenatal prophylactic haematinics. An obstetric ultrasound had not been performed. Antenatal records were not availed.

Her husband worked in a metal and glass industry where she visited him frequently during pregnancy. She never drank alcohol or smoked cigarettes. There was no history of regular medication for chronic disease. She had used DMPA in the preceding birth interval. There was no family history of congenital malformations.

Obstetric examination revealed a term pregnancy in cephalic presentation. The foetal heart rate was normal. She was experiencing mild uterine contractions. She had vaginal bleeding and the cervix was closed. A diagnosis of antepartum haemorrhage at term in early labour in a patient with a previous caesarean section was made. A decision to deliver by emergency repeat caesarean section was made and effected. The outcome was a live term male infant with undescended testes, abnormal genitalia, flabby abdomen and hypoplasia of the right lower limb. The birth weight of the baby was 2600grams and the Apgar score was 6 in 1 minute and 7 in 5 minutes. The baby was taken to the new-born unit. The respiratory rate was 48 breaths per minute, the pulse rate 162 beats per minute and oxygen saturation on room air was at 72%. He was put on oxygen and supplementary feeds. There was no record of administration of intravenous fluids or urine output. Despite classical features, the substantive diagnosis was not reached until the second day. Literature search confirmed the diagnosis of Prune Belly syndrome.

On the second day, there was marked improvement. The respiratory rate was 38 breaths per minute and oxygen saturation on room air 98%. A decision was made to transfer the baby to the National Referral Hospital for advanced care, including a Kidney, Ureter and Bladder (KUB) ultrasound. While at the referral hospital, the condition deteriorated fast and the baby died at the age of 3 days.

Figure 1: The overall external features of the baby

Figure 2: Abdominal, genital and limb

DISCUSSION

This was a case of Prune Belly syndrome that was diagnosed in retrospect on the second day postpartum in a low resource referral public maternity hospital. The gross abnormalities such as abdominal distension, flabbiness and wrinkling of the anterior abdominal wall due to absence of abdominal wall muscles, undescended testes and underdevelopment of the right lower limb were obvious[5]. Despite these classical features, diagnosis was not prompt which indicates a deficiency in knowledge of the syndrome. Although the patient had made four antenatal visits, no ultrasound was performed, which therefore made it virtually impossible to anticipate such an adverse foetal outcome. Subtle internal changes such as ureteric dilatation, urethral obstruction, bladder distension, and cardio-pulmonary changes needed detailed investigations in order to confirm the extent of multi–organ involvement[8]. These investigations were not available in the institution of delivery. In addition, a post-mortem was not performed. Despite this being a surprise occurrence for the parents, there was no counselling provided. Hence, the whole scenario around the case constitutes a challenge in both the diagnostic processes and optimization of management. This case, therefore, exemplifies the need in addressing the handling of mothers carrying babies with congenital abnormalities in a low resource setting [9].

Observations made in this case underscore the importance of universal education on the risk of congenital malformations for all stakeholders. Prenatal diagnosis would enable anticipation of outcomes and institution of appropriate supportive and targeted corrective management[10]. WHO recommends early ultrasound anomaly screening before 24 weeks for identification of foetal anomalies[11]? Abnormalities such as foetal ascites, bladder distension and pulmonary hypoplasia can be detected by a detailed foetal anomaly ultrasound scan[7]. Elaborate preconception care for affected couples during the subsequent birth intervals would enable preparedness for future possibilities of recurrent or other congenital malformations and heighten awareness on different types of teratogens such as heavy metals[12]. For this mother, there is a need for correcting the possible misconception that DMPA may have been the teratogenic factor[13].

CONCLUSION

This case depicts a state of poor preparedness for diagnosis, and management of newborns and their parents, when there are congenital malformations. It is recommended that specific effort be made to introduce congenital anomaly education of mothers in public hospitals. Efficient ultrasound services should be introduced in public hospitals for routine congenital anomaly screening. Strategy to pool data on congenital malformations would heighten awareness by practitioners on types of congenital malformations, and management of both parents and the newborns affected by congenital malformations. Systems for preconception counselling should also be put in place.

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