COMPLETE PENOSCROTAL TRANSPOSITION WITH MULTIPLE CONGENITAL MALFORMATIONS

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SUMMARY

Penoscrotal transposition (PST) is an extremely rare congenital anomaly of the external genitalia, characterized by malposition of the penis in relation to the scrotum. PST can be either complete or incomplete according to the positional exchanges between the penis and scrotum and both forms of PST are generally linked with hypospadias. Incomplete transposition is the common form of this entity and the penis lies in the middle of the scrotum, but complete transposition, the scrotum almost entirely covers the penis, which emerges from the perineum. Both forms are most often associated with a wide variety of other anomalies. We describe a case of a newborn with complete PST, with other associated malformations.

BACKGROUND

Penoscrotal transposition (PST) is a rare anomaly of the external genitalia, characterized by malposition of the penis in relation to the scrotum. PST can be defined as either complete or incomplete according to the positional exchanges between the penis and scrotum and both forms of PST are generally linked with hypospadias. Incomplete transposition is the common form of this entity and the penis lies in the middle of the scrotum, but in complete transposition, the scrotum almost entirely covers the penis, which emerges from the perineum. PST was first reported by Appleby in 1923. Patients with PST often have accompanying urological abnormalities, such as chordee, hypospadias, and vesicoureteric reflux.

The etiology and embryological sequence abnormalities that occur in PST is still unclear. The genital tubercle and the labioscrotal swellings are the embryological origins of the penis and scrotum, respectively. During normal embryonic development, in the 9th–11th week, the scrotal swellings migrate infero-medially and fuse in the midline caudal to the genital tubercle that forms the penis by the 12th week of gestation. This is usually achieved under the influence of androgens and poor response or absence of androgens results in abnormal migration of the scrotal swellings. Somoza et al suggested that an abnormal positioning of the genital tubercle at the 6th gestation week (GA) concerning the scrotal swellings or a defective gubernaculum leads to PST.

Complete penoscrotal transposition (CPST) is frequently characterized by major and often life-threatening anomalies involving the urogenital, cardiovascular, gastrointestinal, and skeletal systems. Common genital anomalies include hypospadias and chordee, and 100% of cases have a renal defect.
CASE PRESENTATION

A gravida 5, para 1, living 1 with 3 abortions woman aged 27 years was referred from Mwananyamara Referral Regional Hospital and admitted to Obstetrics and Gynecology department at Muhimbili National Hospital- Mloganzila. She has a referral diagnosis of antepartum hemorrhage and severe oligohydramnios at GA of 30 weeks 2 days. She had three previous pregnancy losses: 1st and 2nd loses both at 12 weeks GA with 6 months between the loses; her 3rd loss was 2 years after she had a term healthy baby by normal delivery.

She had no histories of phenotypic genetic abnormalities in their families, illicit drug use, cigarette or alcohol consumption, no chemical, radiation exposure, or any chronic illness. She is married, and is a university graduate, working as a transportation officer. She attended antenatal clinic (ANC) five times and all her laboratory work-up such as blood count indices, blood grouping, urinalysis, stool examination, and microscopy, and fasting blood sugar were normal. She was HIV negative and blood film for malaria parasites came back negative. Record of the ultrasound scan taken at 6th -week gestation indicates early multiple pregnancy (two gestational sacs seen) and the next scan performed at 27th weeks GA show a normal single fetus with no anomaly. She was given iron and folic acid supplements, received Tetanus Toxoid vaccine twice, sulfadoxine-pyrimethamine (SP) tables twice as part of intermittent preventive therapy for malaria and she was dewormed using Mebendazole. With her history of pregnancy loss and complaints of abdominal cramps, she had been kept on bed rest at home from 16th week of gestation as recommended by her obstetrician at Mwananyamala Hospital.

After she was brought to our hospital she had active vagina bleeding. A bed side ultrasound revealed placenta previa grade 3 and she was planned for emergency hysterotomy. She delivered a male baby, 1750 gm, who did not initiate spontaneous breathing after birth. The baby was initially resuscitated with a bag and mask before intubation at age 5 minutes. He was assigned an Apgar score of 3, 1, 4 at 1st, 5th and 10th minute respectively. On examination, baby had severe pallor, occipital-frontal circumference – 29cm, communicating anterior and posterior fontanelle, low set ears, short neck, wide-spaced nipples, undescended testes (empty scrotum), inverted genitals (CPST) with hypospadias (Figure 1), rectal atresia, prominent heel, and bilateral talipes equinovarus.

DISCUSSION

Penoscrotal transposition (PST) is a congenital urogenital anomaly described first in 1923 by Appleby. The embryological sequence responsible for this malformation remains unclear; however, it has been suggested that an abnormal positioning of the genital tubercle in relation to the scrotal swellings during the critical fourth to the fifth week of gestation could affect the migration of the scrotal swellings2. In this case report, there is a complete exchange of position with the scrotum located superior to the penis, which is inferior to scrotum. (Figures 1(a) and 1(b)). Also, there presents a spiral and hypoplastic penis crooked toward the anal position. Ayamba et al reported the same findings whereby noticed complete transposition of the external genitalia with cryptorchidism, hypoplastic penis from the perineum just above the blind anal position, and caudal to the scrotum3. Somoza et al also noted at birth a newborn has a complete transposition of the external genitalia, a 3.5-cm-long, hypospadic, and hypoplastic penis.

Figure 1: (a) shows a horizontal view of complete penoscrotal transposition and (b) shows an oblique view of complete penoscrotal transposition.
arose from the perineum, just above the anus and beneath a normal scrotum1–3,5.

CPST is often characterized by major associated malformations. Our baby had also other multiple physical abnormalities such as short neck, low set ears and talipes equinovarus. Unfortunately, due to our limited resources, we could not complete imaging of internal organs. Previous reports of CPST have also noted presence of other malformation with 100% occurrence of renal anomalies. For example, Parida et al had noted major renal anomalies in the form of agenesis, horseshoe kidney, ectopic and dysplastic kidney, obstructive uropathy, and hydronephrosis. Other systemic abnormalities are mental retardation, anorectal malformations, central nervous system, skeletal and cardiological defects5. In our case, we did not perform imaging to detect renal anomalies, but literature suggests most likely there were there. The detection of CPST should warrant careful clinical evaluation to rule out other anomalies.

Although some reported a family history and genetic basis for the incidence of PST6, we did not find any evidence of positive family history of phenotypic abnormalities. When associated with severe hypospadias, penoscrotal transposition necessitates a staged surgical repair for physiological and psychological reasons.

Our newborn required advanced resuscitation at birth, likely due to hypoxia in utero as a result of significant blood loss (placenta previa grade 3). This is supported by the findings that baby was very pale at birth. However, we cannot rule out the possibility of other anomalies such as fatal cardiac anomalies which are incompatible with life1,5.

OUTCOME

The newborn was transferred to the neonatal intensive care unit for further treatment and passed on after 4 hours. No any radiological or laboratory investigation were completed within this time.

LEARNING POINTS/TAKE HOME MESSAGES

1. Strengthening of antenatal care services in a primary health facility is a key for positive outcome of pregnancy. This is by early detection of abnormal development of fetus in utero by early ultrasound

2. Referring hospitals in low-income settings should be strengthened with well knowledgeable personnel (radiographers) and modern equipment. As we have notice in this case even a placenta praevia was detected after being received in a tertiary hospital despite the woman being scanned in late 2nd trimester.

3. There is a need of strengthening neonatal ICU by ensuring bedside radiological equipment’s are available also other ICU equipment’s are enough. As we have seen no any radiological investigation done to the baby due to the fact that the baby was in critical condition but based on critical care knowledge and experience this could possible by bedside equipment.

4. Learning culture must be strengthened in our institute; if we had good learning culture radiological investigations would have been done to the dead baby for learning purpose to detect if there is any other internal congenital anomaly and other cause of death to this newborn

PATIENT’S PERSPECTIVE

Am so thankful for the services I receive from all hospitals and I declare to have no any experience of having an abnormal baby in my family and even my husband’s family. This is my first time to give birth a newborn with congenital malformation and i wondered the way it was not even discovered early during the antenatal period. Also, I promise to attend the clinic early for the next pregnancy and follow all instructions that i will be given by health care providers for the sake of the good health of herself and next baby. Am so happy about this publication because it will help other doctors to identify the condition and treat it accordingly also for those who are in learning schools will learn more about this condition.

Competing interests: None.

Patient consent: Obtained

References