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A Rare Case of Complete Penoscrotal Transposition with Multiple Congenital Malformations

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Abstract

Background

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.

Significance

This case study has the aim of identifying missed opportunities of early identification of congenital malformations during antenatal care period, by early imaging and karyotype test. Having knowledge on identified congenital malformations during prenatal period is useful to obtain baseline rates and identify clue to the etiology of condition, which will be helpful to plan ongoing antenatal care.

Material and Methods

This case study was conducted after obtaining ethical clearance from Training, Research and Consultancy Unit (TRCU) at Muhimbili National Hospital and information was collected through in-depth interview (IDI) with the patient's mother and health care providers who took care of a deceased baby. Also, other data were obtained in patient case file and pictures that were taken. Informed consent was sought from the mother to agree for interview and to allow pictures of her baby to be used in this case study.

Outcome

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

Recommendations

1. Strengthening of antenatal care services in a primary health facility is a key for positive outcome of pregnancy. 2. Referring hospitals in low-income settings should be strengthened with well knowledgeable radiographers. 3. There is a need of strengthening neonatal ICU by ensuring bedside radiological equipment's is available also other ICU equipment's are enough. 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 was any other internal congenital anomaly and other cause of death to this newborn.

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Introduction

Penoscrotal transposition (PST) is a rare anomaly of the external genitalia, characterized by malposition of the penis in relation to the scrotum(1,2,3). 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 (1). PST was first reported by Appleby in 1923. Patients with PST often have accompanying urological abnormalities, such as chordee, hypospadias, and vesicoureteric reflux (4).

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 9^{th}–11^{th} week, the scrotal swellings migrate infero-medially and fuse in the mid-line caudal to the genital tubercle that forms the penis by the 12^{th} 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[3]. Somoza et al suggested that an abnormal positioning of the genital tubercle at the 6^{th} gestation week (GA) concerning the scrotal swellings or a defective gubernaculum leads to PST (2).

Complete penoscrotal transposition (CPST) is frequently characterized by major and often life-threatening anomalies involving the urogenital, cardiovascular, gastrointestinal, and skeletal systems (2). Common genital anomalies include hypospadias and chordee, and 100% of cases have a renal defect.

Complete PST can be diagnosed by ultrasound (USS) in the sagittal plane when the scrotum is located above the penis; incomplete PST is identifiable in the coronal plane when the phallus is located in the middle of a divided scrotum. Amnio-infusion in the context of reduced liquor, 3D/4D scanning and fetal magnetic resonance imaging (MRI) are further modalities of imaging that may increase diagnostic accuracy to help in counseling parents (5). An early scan performed at 12 – 13 weeks 'gestation by a competent sonographer can detect about half of the prenatally detectable structural anomalies and100% of those expected to be detected at this stage (6).

In a study performed by Mohamed A. Baky Fahmy (2014), sixty-three cases of penoscrotal positional anomalies were identified from a total of 2400 babies examined, providing an

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approximate overall incidence of 2.5%. Fifty six cases presented different grades of PST, among them only three cases (18%) had the penis completely caudal to a cephalically migrated scrotum, those cases are considered as complete PST (7).

Survival among reported cases of PST varies depending on the associated abnormalities. In uncomplicated cases, survival into adulthood has been reported, and in some cases with fertility intact, but paternity in cases of PST has not yet been studied. In complicated cases, the mortality rate is significantly high due to associated visceral organ anomalies (8).

Case Presentation

A gravida 5, para 1, living 1 with 3 abortions woman aged 27 years was referred from Mwananyamala Referral Regional Hospital and admitted to Obstetrics and Gynecology department at Muhimbili National Hospital - Mloganzila with a referral diagnosis of antepartum hemorrhage and severe oligohydramnious at GA of 30 weeks 2 days. She had three previous pregnancy loses: 1st and 2nd loses were 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 history 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 was normal. 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 week 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) tablets 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 MNH at Mloganzila she had active vaginal bleeding. A bed side ultrasound revealed grade 3 placenta previa 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

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minute, respectively. On examination, the 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. 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 is a spiral and hypoplastic penis crooked toward the anal position.

Based on these findings, a diagnosis of complete PST with multiple malformations was made.

Parents' Perspective

"*I am so thankful for the services I received from all hospitals, and I declare to have no experience of having an abnormal baby in my family and even my husband's family. This is my first-time giving birth to a newborn with congenital malformation and I wondered 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. I am so happy about this publication because it will help other doctors to identify the condition and treat it accordingly. Also, those who are in learning schools will learn more about this condition*". Said patient's (deceased baby) mother.

Figure/Image Captions

(a) **(b)**

Figure 1. (a) shows a horizontal view of complete penoscrotal transposition and (b) shows an oblique view of complete penoscrotal transposition

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The newborn was transferred to the neonatal intensive care unit where he passed on after 4 hours. No radiological or laboratory investigations were completed within this time.

Significance

This case study has the aim of identifying missed opportunities of early identification of congenital malformations during ANC period, by early imaging and karyotype test. Having knowledge on identified congenital malformations during prenatal period is useful to obtain baseline rates and identify clue to the etiology of condition, which will be helpful to plan ongoing ANC. i.e., for life threatening congenital malformations, to consider termination of pregnancy at the early stage rather than late. There is evidence that early termination of pregnancy is less traumatic for the mother than when carried out at a stage when fetal movements are already felt, which may push parents into making a rushed decision[6].

Ethical Consideration

Ethics approval was obtained from MNH ethical committee. No name or identifiers were recorded in the interview with patient's mother to ensure anonymity and confidentiality. Individual signed and written consent was obtained from parents before they participated in the case study.

Discussion

PST is a congenital urogenital anomaly described first in 1923 by Appleby (4). 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 swellings (2). 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 is a spiral and hypoplastic penis crooked toward the anal position. Ayamba et al reported the same findings whereby they noticed complete transposition of the external genitalia with cryptorchidism, hypoplastic penis from the perineum just above the blind anal position, and caudal to the scrotum (3). Somoza et al also noted at birth a newborn with a complete transposition of the external genitalia, a 3.5-cm-long, hypospadic, and hypoplasic penis arose from the perineum, just above the anus and beneath a normal scrotum (1–3,9). CPST is often characterized by major associated malformations. The baby had also other multiple physical abnormalities such as short neck, low set ears and talipes equinovarus.

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Unfortunately, we could not complete imaging of internal organs. Previous reports of CPST have also noted presence of other malformations 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 defects (9). In our case, we did not perform imaging to detect renal anomalies, but literature suggests it was most likely to be there. The detection of CPST should warrant careful clinical evaluation to rule out other anomalies.

Although some have reported a family history and genetic basis for the incidence of PST (10), we did not find any evidence of positive family history of phenotypic abnormalities. When associated with severe hypospadias, penoscrotal transposition a staged surgical repair is indicated. The 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 the baby was very pale at birth. However, we could not rule out the possibility of other anomalies such as fatal cardiac anomalies

Learning points/take home messages

1. Strengthening of ANC 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. An early scan performed at 12 – 13 weeks gestation by a competent sonographer can detect about half of the prenatally detectable structural anomalies and100% of those expected to be detected at this stage (6).
2. Referral hospitals in low-income settings should be strengthened with well knowledgeable personnel (radiographers). As we have noticed in this case even a placenta praevia was missed only to be detected following referral to 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 are available for quick imaging and come up with a proper diagnosis which will guide on proper management of the patient (11). As we have seen in this case no radiological investigation was done to the baby due to the fact that the baby was in critical condition.
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

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to detect if there was any other internal congenital anomaly and other cause of death to this newborn.

Recommendations

1. Basic obstetric ultrasound in a first and second trimester should be performed to all pregnant woman to know the progress of the pregnancy especially fetal growth and development.
2. Referral hospitals in low-income settings should be strengthened with well knowledgeable radiographers.
3. Bedside radiological equipment's should be available in Neonatal ICU for quick imaging and guidance in early intervention based on neonates' condition.
4. Learning culture must be strengthened in hospitals by performing postmortem to all idiopathic deaths that occur to a newborn with congenital anomalies so as to know exactly the cause of death and use that opportunity to learn on how to save other newborns with the same condition.

Conclusion

CPST is rare congenital malformation and can be detected by performing early USS scan at 12 – 13 weeks' gestation. CPST Amnio-infusion in the context of reduced liquor, 3D/4D scanning and fetal magnetic resonance imaging (MRI) are further modalities of imaging that may increase diagnostic accuracy to help in counseling parents. Complete penoscrotal transposition (CPST) is frequently characterized by major and often life-threatening anomalies involving the urogenital, cardiovascular, gastrointestinal, and skeletal system. Common genital anomalies include hypospadias and chordee, and 100% of cases have a renal defect. Management of all forms of PST comprises initial treatment of associated urinary and other abnormalities, followed by surgical correction of the PST. In more complex cases of PS, surgery is technically challenging. It is usually performed during early childhood.

Competing interests:

Authors declare to have no competing interests.

Ethical considerations and consent

Ethics approve were obtained from Muhimbili National Hospital ethical committee. No name or identifiers were recorded on interview with patient mother to ensure the anonymity and

confidentiality. Individual signed and written consent was obtained from parent before they have participated in the study. The participant was well informed that her participation is voluntary, and she has a right to terminate at any time if don't want to continue. It was clear clarified that the information provided will be for the research purposes and would therefore strictly dealt confidential.

Confidentiality was maintained as the information from the respondent will be in a locked place and accessed by the principal investigator only. Participant was informed that, no potential study risks to the participant and if anything, unpleasant will come out from the findings, the participant will be protected.

Authors' contributions

J.L contributed to study conception and design, J.L, M.M and D.K contributed to data collection and analysis of the data. M.M took the photos with the consent from parents. J.L, M.M, D.K writing of the case presentation. R.M review and contribute to writing of the case report. J.L and R.M writing a manuscript with input from all authors.

Abbreviations

3D - Three dimension
4D - Four dimension
ANC - Antenatal care
CPST - Complete penoscrotal transposition
ICU - Intensive care unit
IDI - In-depth interview
GA - Gestation age
MNH - Muhimbili National Hospital
PST - Penoscrotal transposition
SP - Sulfadoxine-pyrimethamine
TRCU - Training, research and consultancy unit
USS - Ultrasound scan

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References

1. Beyazıt, F., Pek, E., & Aylanç, H. (2017, March 1). A rare case of complete penoscrotal transposition with hypospadias in a newborn. Turkish Journal of Obstetrics and Gynecology. https://doi.org/10.4274/tjod.88262
2. Somoza, I., Palacios, M. G., Mendez, R., & Vela, D. (2012). Complete penoscrotal transposition: A three-stage procedure. Indian Journal of Urology, 28(4), 450–452. https://doi.org/10.4103/0970-1591.105775
3. Ali Mahamudu Ayamba, Raymond Saa-Eru Maalman, Yaw OtchereDonkor, John Noah Anyorigiya, "Complete Penoscrotal Transposition with Other Extragenital Anomalies in a Neonate Delivered at Term", Case Reports in Urology, vol. 2021, Article ID 6676301, 4 pages, 2021. https://doi.org/10.1155/2021/6676301
4. Appleby, L. H. (1923). An Unusual Arrangement of the External Genitalia. Canadian Medical Association Journal, 13(7), 514–515.
5. Sexton P, Thomas J, T, Petersen S, Brown N, Arms J, E, Bryan J, Harraway J, Gardener G: Complete Penoscrotal Transposition: Case Report and Review of the Literature. Fetal Diagn Ther 2015;37:70-74. doi: 10.1159/000358592
6. Kenkhuis MJA, Bakker M, Bardi F, Fontanella F, Bakker MK, Fleurke-Rozema JH, Bilardo CM. Effectiveness of 12-13-week scan for early diagnosis of fetal congenital anomalies in the cell-free DNA era. Ultrasound Obstet Gynecol. 2018 Apr;51(4):463-469. doi: 10.1002/uog.17487. Epub 2018 Mar 4. PMID: 28397377.
7. Mohamed A. Baky Fahmy, Alaa Aldin A. El Shennawy, Ayman Mohtdy Edress,Spectrum of penoscrotal positional anomalies in children,International Journal of Surgery,Volume 12, Issue 9,2014,Pages 983-988,ISSN 1743-9191,https://doi.org/10.1016/j.ijsu.2014.08.001.
8. Bhargavi B. Kola, MD; Vivekananda Dasari, MD; and Deepak Bommisetty,Penoscrotal Transposition,Volume 14 – Issue 8 – August 2015,consultant360, https://www.consultant360.com/articles/penoscrotal-transposition
9. Parida, S. K., Hall, B. D., Barton, L., & Fujimoto, A. (1995). Penoscrotal transposition and associated anomalies: Report of five new cases and review of the literature. American Journal of Medical Genetics, 59(1), 68–75. https://doi.org/10.1002/ajmg.1320590115
10. MacKenzie, J., Chitayat, D., McLorie, G., Balfe, J. W., Pandit, P. B., & Blecher, S. R. (1994). Penoscrotal transposition: A case report and review. American Journal of Medical Genetics, 49(1), 103–107. https://doi.org/10.1002/ajmg.1320490120
11. Fujii AM, Sinha B (2012) Bedside Ultrasound in the Neonatal Intensive Care Unit: A Wave of the Future. J Pulmonar Respirat Med 2:e112. doi:10.4172/2161-105X.1000e112