

CASE REPORT**PENILE AGENESIS WITH CROSSED FUSED RENAL ECTOPIA**Milliard Derbew¹, Elbet Ketema¹**ABSTRACT**

Penile agenesis is one of the rarest urogenital anomalies with only less than 100 cases reported worldwide so far. Only 3 cases have been reported from Africa and to our knowledge none has been reported from our country Ethiopia. Viability depends on associated anomalies. Urogenital anomalies are the most common associated ones accounting for 54% of cases. This case report is unusual presentation, which is the first reported case of penile agenesis associated with left to right, crossed fused renal ectopia.

Key words: aphallia, phalloplasty, crossed fused kidney

Case presentation History: The baby was born to a 19 year old para II Ethiopian mother. The father is also Ethiopian whose age is unknown (Estimated to be 30?). The parents belong to the same family tree. Father is a smoker but the mother is not. She had no antenatal follow up but pregnancy was uneventful and delivery was by spontaneous vaginal delivery at Hiwot Fana specialized university hospital which is located in Eastern Ethiopia after 9month of amenorrhea. Labor lasted for 6 hours and baby had good APGAR score at 1st and 5th minute. Baby was referred from delivery room to neonatology unit for absent penis.

Physical examination: Baby doesn't have dysmorphic feature on face. Birth weight was 2.25kg and height at birth was 48cm. by using the new Ballard score gestational age was 38 weeks. Systemic examination revealed no abnormality except finding of the genitalia. He has well formed scrotum with rufae and testis are both well descended bilaterally but absent penile shaft (phallus). The anal canal is at normal site and is patent with normal anal tone. We witnessed the baby passing urine per rectum mixed with meconium.

Investigation: Renal ultrasound and abdominal CT showed full bladder with left to right crossed fused ectopic kidney. Rectum was distended and filled with fluid and solid matter. There was no uterus, ovary or hidden penis.

Karyotype analysis of DNA obtained from cheek cells show 46XY with no apparent polyploidy on both sex chromosomes. The SRY region on the Y chromosome does not show any copy number changes. No apparent changes on the telomeres of all chromosomes were observed.

Conclusion: 46XY



Pic: 1. A 45 minute old neonate with penile agenesis.

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DISCUSSION

Penile agenesis is a rare urogenital anomaly defined by absent penis (phallus) (1). It's said to occur once in 30 million live births (2). It was first reported by Immenger in 1853 and since then there had been <100 cases reported worldwide (3,4). Only 3 cases in Africa and to our knowledge none has been reported in our country Ethiopia so far (5).

Penile agenesis occurs as a result of absence or failure of the genital tubercle to develop during 3rd-6th weeks of gestation and the exact etiology is unknown. (6). Early in the 4th week, proliferating mesenchymal cells at the cranial end of the cloacal membrane produces a genital tubercle in both sexes. Under the influence of dihydrotestosterone hormone from the fetal testes, however, the genital tubercle later enlarges to form a phallus. Corpora cavernosa and corpora spongiosum develop from mesenchyme in the phallus. Labioscrotal Swellings (genital swellings) and urogenital folds (urethral folds) soon develop on each side of the Ventral part of the cloacal membrane. As the phallus enlarges and elongates to become the penis, urogenital folds, lining the lateral walls of the urethral groove on the ventral surface of the penis, fuse in the midline to form the spongy (penile) urethra and Labioscrotal swellings of each side fuse to form the scrotum (7).

Penile agenesis can be classified into three based on the opening of the urethral meatus according to skoog and bellman. These are the post spenecteric, presphenecteric and urethral atresia. Most patients (60%) have presphenecteric and the urethral opening is at the anal verge. They have few associated anomalies and mortality is low. In case of postsphenecteric the urethra open on the anterior rectal wall and they have associated anomaly in one third of cases. Our patient belongs to this group since the urethral opening is on anterior rectum and has associated renal anomaly, Urethral atresia occur in 12% of patients and associated urogenital anomaly is universal. In general higher-level communications are associated with major anomalies and mortality is high (8).

Associated anomalies in Penile Agenesis, PA, include those of the caudal axis, genitourinary and gastrointestinal tract anomalies. Urogenital anomalies are the common associated anomalies which ac-

counts for around 54% (8). They include renal agenesis, hypoplastic kidneys, cystic kidneys, hydronephrosis, abnormal renal rotation, pelvic kidneys, horseshoe kidneys, vesicoureteral reflux, hypoplastic bladder, vesicocolic fistula, blind urethra, agenesis of prostate and seminal vesicles, cryptorchidism, hydrocele and inguinal hernia (9). Our case is the first reported case of penile agenesis which is associated with left to right crossed fused renal ectopia with a single ureter.

Crossed fused renal ectopia is a rare anomaly which is second frequent fusion abnormality of the urinary tract after horse shoe kidney (10). It is an anomaly where the kidneys are fused and located on the same side of the midline. It is said to occur 1 in 1000 births and is thought to result from the abnormal development of the ureteric bud and metanephric blastema during the 4th to 8th weeks of gestation. Patients may have symptoms associated with infection, obstruction and urolithiasis but largely they are asymptomatic (11).

In penile agenesis the testes and the scrotum are almost always normal and all reported cases so far had 46XY chromosome except for 1 case with mosaic XX,XY karyotype (12).

Management of a patient with penile agenesis is a major challenge to the care giving physicians and the parents as well because of the significant psychosocial trauma, which follows. Management options include female sex reassignment with feminizing genitoplasty or multi staged phalloplastic surgery (13). In many of literatures early sex reassignment to female gender is the recommended surgical treatment of this genetically male infant especially if the patient present at an early age (14). However there is growing evidence that sexual orientation and gender identity that is the conviction of belonging to either the male or female gender independent of the anatomical reality of the sex are programmed into various areas of the brain even before birth (15,16) and this will result in gender dysphoria and sex reassignment back to male later in life. The current trend is therefore to rear these children as males (17, 18). In our case the parents refuse any treatment option and disappeared, despite the arrangement made for treatment abroad.

Consent: Written informed consent was obtained from the patient for publication of this case report and any accompanying images. A copy of the written consent is available for review by the editors of this journal.

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