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Case Report
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# Complete penoscrotal transposition in the male twin of a dichorionic diamniotic pregnancy from oocyte donation: a case report

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## Abstract

**Objective:** The case highlights the rarity and complexity of penoscrotal transposition (PST), emphasizing the need for multidisciplinary management and early prenatal diagnosis.

**Case(s):** A 36-year-old primigravida with a 20-week oocyte donation dichorionic diamniotic pregnancy was referred due to an abdominopelvic cyst and absent left kidney in one fetus. Prenatal screening by cell-free DNA showed low aneuploidy risk and the presence of a Y chromosome, and no invasive diagnostic procedures were performed due to the patient's refusal. Ultrasound revealed megacystis, hyperechogenic kidneys, and suspected PST in the male twin. The patient underwent cesarean delivery at 35+2 weeks, with subsequent diagnosis of penoscrotal transposition.

**Conclusion:** Prenatal diagnosis of penoscrotal transposition is challenging. This case underscores the importance of genital assessment during referral scans, particularly in the presence of urinary tract anomalies.

**Keywords:** Complete penoscrotal transposition, prenatal diagnosis, megacystis

## Introduction

Penoscrotal transposition (PST) is an extremely rare congenital anomaly characterized by the abnormal positioning of the penis, located caudally and posteriorly to the scrotum.[1] The precise embryological mechanism leading to PST remains unclear; however, it is hypothesized that an abnormal positioning of the genital tubercle during the fourth to fifth week of gestation may disrupt the normal migration and development of the scrotal swellings.[2] Isolated cases of PST are uncommon, as this condition is frequently associated with severe, life-threatening malformations involving the genitourinary, intestinal, cardiovascular, and skeletal systems.[3] Here, we report a case identified prenatally, presenting with megacystis, renal dysplasia, and complete penoscrotal transposition.

## Case(s)

A 36-year-old primigravida with a 20-week dichorionic diamniotic twin pregnancy conceived through oocyte donation was referred to our center due to the presence of an abdominopelvic cystic anechoic formation and the inability to visualize the left kidney in one of the fetuses. The patient had no history of genetic abnormalities, medication use, or substance consumption, aside from a diagnosis of gestational diabetes. Prenatal aneuploidy screening using a cell-free DNA test indicated a low risk for trisomy 21, 13, and 18, and the presence of the Y chromosome. However, the patient did not undergo a first-trimester screening ultrasound.

The pregnant woman was referred to our clinic with suspicion of megacystis at 20+6 weeks and it was confirmed by the ultrasound performed at our center. Female genitalia were observed in one fetus, while the other one, presumed to be male based on the Y chromosome detected by cell-free DNA test, presented with an abdominopelvic cystic anechoic formation (41x37x37 mm) surrounded by umbilical arteries, consistent with megacystis. Both kidneys were present but exhibited a hyperechogenic echostructure, with no evidence of upper

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urinary tract dilatation (Figure 1 A and B). Initially, at the genital level, the scrotum was visible, but no ultrasound evidence of a penile shaft was identified, raising suspicion of aphallia. Subsequent ultrasounds revealed the presence of scrotal sac and a curved penile remnant (Figure 2 A and B), suggestive of hypospadias. These findings are characteristic of congenital anomalies such as PST or severe forms of penoscrotal hypospadias. The female fetus showed no ultrasound abnormalities. Amniotic fluid volume and fetal echocardiography were both normal. At 21+4 weeks of gestation, a fetal MRI was performed to further evaluate the genitalia and suspected urogenital abnormalities. The MRI revealed the presence of a large neck diverticulum of the anterocranial bladder wall, causing posterior-cranial displacement of the remaining abdominal organs and slight elevation of the hemidiaphragm, raising suspicion of urachus persistence (Figure 3 A and B). The kidneys were in their normal anatomical positions but exhibited restricted signals on diffusion-weighted imaging (DWI) MRI, with no evidence of hydroureteronephrosis. The scrotal sacs contained fluid, but the penile shaft was not clearly visualized.

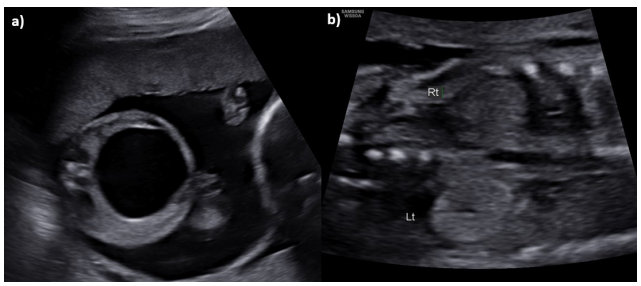


**Fig 1.** a) Axial sonogram of the pelvis showing an abdominopelvic cystic anechoic formation (41x37x37 mm), indicative of megacystis. b) Coronal sonogram of kidneys that appear with a hyperechogenic echostructure.

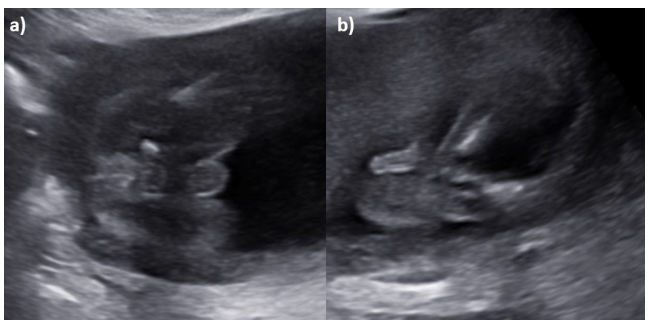


**Fig 2.** Coronal sonogram of the pelvis showing a) a scrotal sac without an identifiable penis and b) a phallic protrusion extending dorsocaudally from the scrotum to the anal orifice. This finding may indicate an abnormality in genital development, such as in cases of penoscrotal transposition or other congenital anomalies

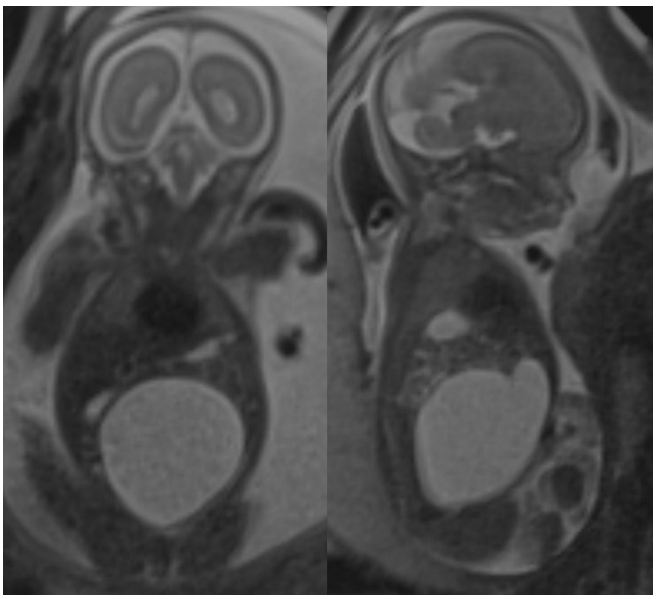

**Fig 3.** Magnetic resonance at 21 weeks of gestation showing megacystis with displacement of the abdominal and thoracic organs and the presence of a large neck diverticulum of the antero-cranial bladder wall raising suspicion of urachus persistence

Multidisciplinary counselling was provided, addressing potential urogenital abnormalities, including megacystis due to a posterior urethral valve or urethral atresia, proximal hypospadias with significant penile recurvatum, continent epispadias, and the likelihood of reduced renal function.

Although the risk was considered low, chromosomal disorders such as Denys-Drash syndrome and complex malformations like CHARGE syndrome could not be entirely excluded. Therefore, amniocentesis for karyotype assessment, analysis by QF-PCR, and array-CGH, were proposed but not performed due to the couple's refusal to accept the risk of complications and potential loss of the unaffected twin.

Weekly ultrasound follow-up was conducted until admission at 35 weeks of gestation, along with consultations with a neonatologist, pediatric nephrologist, pediatric urologist, and maternal-fetal medicine specialist. Interestingly, the affected twin later developed polyhydramnios, diagnosed via ultrasound at 26+3 weeks of gestation.

The patient underwent a cesarean section at 35+2 weeks of gestation due to cardiotocographic abnormalities detected during routine fetal well-being monitoring. The affected twin weighed 2500g, with an Apgar score of 7/9 at birth, while the female twin was healthy with an Apgar score of 9/9.

Physical examination revealed a distended abdomen, complete PST, non-palpable testes in the scrotal sac, an inverted penile shaft with a posterior apical portion, and an inverted scrotal raphe (Figure 4 A, B and C). A punctiform apical urethral meatus was noted. No other external dysmorphic features were observed. These findings confirm the initial ultrasound suspicion of PST. Abdominal ultrasonography revealed an overdistended bladder extending above the transverse supraumbilical line, with both kidneys showing hyperechoic parenchyma and minimal cortico-medullary differentiation. No urinary tract dilatation was observed bilaterally. The nephrological findings were suggestive of chronic renal failure, consistent with bilateral renal dysplasia associated with malformative uropathy. Testicular ultrasound confirmed left-sided cryptorchidism. Developmental dysplasia of the hip (DDH) was diagnosed, and spinal ultrasound revealed the absence of the coccyx and abnormalities of the conus medullaris, which require further investigation and confirmation by MRI. Genetic evaluations, including karyotyping, 400 Kb CGH arrays, and an NGS panel targeting genes associated with urogenital malformations, were performed and no abnormality was detected in the evaluation. No genital reconstruction surgery has been performed to date.

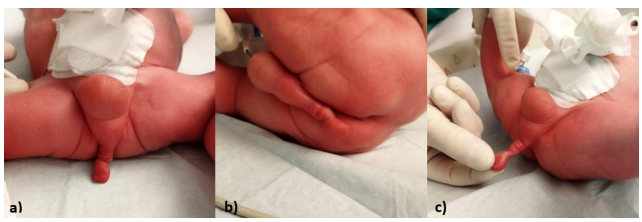


**Fig 4.** a and b) Anterior and lateral view of external genitalia showing complete penoscrotal transposition with penis under the scrotum, inverted penile shaft with a posterior apical portion and inverted scrotal raphe. c) Punctiform apical urethral meatus

## Discussion

Penoscrotal transposition (PST) is an extremely rare congenital anomaly characterized by the abnormal positioning of the penis, located caudally and posteriorly to the scrotum. Various classifications exist in the literature, typically categorizing PST as complete or partial. In the complete type, an intact scrotum is situated above the penis, as observed in our case. In the partial or incomplete type, the scrotum remains unfused, with the penile shaft positioned along its length.[1] PST is frequently associated with severe, life-threatening malformations involving the genito-urinary, intestinal, cardiovascular, and skeletal systems.[3] In our case, the patient was diagnosed with DDH, coccyx aplasia, residual urachus, megacystis, and bilateral renal dysplasia leading to chronic renal insufficiency. Most cases of PST are sporadic, with no evidence of maternal exposure to radiation, infections, or teratogens, suggesting a potential genetic etiology.[4] Chromosomal anomalies, such as mosaic trisomy 18[5] or 13q22 deletion[6–8] have been reported in the literature, as well as associations with Klinefelter syndrome.[4] These findings underscore the importance of performing karyotype analysis in all cases of PST.[1]

Prenatal diagnosis of PST is infrequent and often overlooked. However, it should be considered in the differential diagnosis when ambiguous genitalia or a major urogenital abnormality are suspected. In our case, the patient was referred to our center due to the detection of an abdominopelvic cystic anechoic formation and the inability to visualize the left kidney in one of the fetuses. Complete PST can be identified via ultrasound in the sagittal plane when the scrotum is located above the penis. Incomplete PST, on the other hand, can be recognized in the coronal plane when the penis is located in the middle of a divided scrotum. The diagnosis can be enhanced by the combined use of 3-D and 2-D ultrasound, as demonstrated by Wang et al., who reported significantly higher detection rates with 3-D ultrasound compared to 2-D ultrasound alone (90.9% vs 63.6%, $P < 0.05$) .[9]

Early prenatal diagnosis is crucial, as it facilitates improved parental counselling, referral to specialized delivery centers, and thorough preparation of the medical team for postnatal care. The differential diagnosis for PST should include conditions such as pseudohermaphroditism, micropenis, intrauterine penile amputation, penoscrotal hypospadias, and penile agenesis accompanied by a midline skin tag anterior to the anal region.[10] A comprehensive physical examination is essential to identify associated abnormalities across various organ systems, given the wide range of clinical manifestations that may result in significant morbidity and mortality.[11]

The gold standard for managing PST is surgical intervention, typically recommended between 12-18 months of age.[12] However, in our case, surgery will be postponed until school age due to the presence of other comorbidities.

## Conclusion

PST is a rare congenital anomaly often associated with severe malformations. Although genital assessment is not included in the standard second-trimester ultrasound examination, it should be performed during a referral scan, with particular attention to the labial folds in female fetuses and the position of the penis relative to the scrotum in male fetuses. This evaluation becomes particularly im-

portant in the presence of urinary tract anomalies, such as megacystis, as observed in our case. Prenatal diagnosis of PST remains challenging and is often overlooked. However, advanced imaging techniques, including 3D ultrasound and fetal MRI, can enhance its characterization and diagnostic accuracy. Our case highlights the importance of a multidisciplinary approach in both prenatal and postnatal management, allowing for more accurate parental counseling and optimization of therapeutic strategies.

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