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Discordant fetal sex between NIPT and ultrasound: a diagnostic challenge at term

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A 34 years old primigravida (G1P0), with no relevant personal or family history was admitted at 39+3 weeks of gestation in active labor, with 7 cm cervical dilation and intact membranes. Pregnancy follow-up had been uneventful.

- First trimester non-invasive prenatal testing (Harmony) indicated low risk for aneuploidy and predicted a male fetus.
- First, second, and third trimester ultrasounds (at 12+5, 21+5, and 35 weeks) consistently demonstrated normal fetal anatomy and clearly visualized female external genitalia establishing a persistent and unequivocal discordance in fetal sex determination.

At birth, neonatal examination confirmed **female external genitalia**.

Postnatal abdominal and pelvic ultrasound did not identify structures clearly attributable to a uterus or ovaries, nor were gonads visualized in the inguinal regions, or pelvic cavity. An ovoid solid structure adjacent to the bladder floor was noted, of uncertain origin (rudimentary prostate vs dysmorphic uterine structure)

A karyotype analysis is currently in progress to further clarify the underlying genetic sex and guide diagnosis.



Figure 1. Female external genitalia on third trimester ultrasound

Conclusion

This case underscores a clinically significant discordance between NIPT and ultrasound findings at term, posing diagnostic and counselling challenges. Potential explanations include confined placental mosaicism, vanishing twin, maternal chromosomal abnormalities, or fetal disorders of sex development.