Prenatal diagnosis of a Swyer Syndrome as a result of gender discrepancy in NIPT and ultrasound

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Case

A 36 years old primigravida made a NIPT because of advanced maternal age. The result was negative for trisomy 13, 18, 21 and described a male gender. With 22 weeks of gestation a detailed ultrasound was done. Here we diagnosed a female phenotype.

The prenatal diagnosis of disorders of sexual development (DSD) is a stepwise approach. Here we had a male chromosomal gender and a female phenotype. First, the gonadal gender was unknown. This constellation corresponds to a 46, XY DSD.

46,XY DSD:

- **gonadal dysgenesis**: Anti Mueller Hormone (AMH) is not measurable. These people have a uterus.
- **androgen receptor insensitivity**: AMH is measurable. These people have no uterus.

Ultrasound

In the second half of pregnancy it is possible to see the uterus. In the axial plane we measured the rectovesical interspace (see figure 1 and 2).

In our case a uterus was seen. So we had the suspicion of a gonadal dysgenesis in 46, XY DSD (Swyer Syndrome). This was confirmed after birth: molecular genetic was normal for androgen receptor gen, AMH was not measurable and the ultrasound showed a uterus as we suspected prenatal.

Conclusion

In cases of a discrepancy of fetal phenotype in ultrasound and fetal karyotype the presentation of the fetal uterus is very helpful and pointing the way.