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A Rare Prenatal Encounter: Diagnosing Bardet-Biedl Syndrome In Utero

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Objective

To describe a rare case of Bardet-Biedl Syndrome (BBS) in which prenatal diagnosis enabled timely, multidisciplinary management.

Methods

Retrospective description of a clinical case.

Results

We present the case of a 28-year-old primigravida. The first-trimester combined screening indicated a low risk for aneuploidies and no fetal structural anomalies were detected. The second-trimester scan revealed bilateral postaxial polydactyly of the hands and feet, suspected hypospadias, bilateral choroid plexus cysts, and a complete atrioventricular septal defect. In light of these findings, an amniocentesis was performed, with PCR for common aneuploidies and chromosomal microarray analysis both yielding normal results. At 26 weeks' gestation, the patient was referred to our clinic of fetal nephrourological pathology. After a multidisciplinary evaluation, the constellation of anomalies was considered highly suggestive of a ciliopathy. Next-generation sequencing (NGS) on fetal DNA identified two variants in theBBS12gene: one pathogenic (c.1482_1485del) and one of uncertain significance (c.65T>C), which was later reclassified as likely pathogenic. All the prenatal findings were confirmed except the hypospadias. This confirmation of the diagnosis ofBardet-Biedl Syndrome (BBS) enabled the application of a multidisciplinary postnatal care plan, including psychological support, pediatric cardiology, urology, and orthopedics follow-up.

Conclusion

Early prenatal diagnosis ofBBSthrough imaging and molecular testing allowed for timely genetic counseling and the development of a multidisciplinary care plan. This approach ensured comprehensive management and support for both the mother and the newborn.