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

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Introduction

Bardet-Biedl syndrome is a rare autonomic recessive ciliopathy characterized by a spectrum of clinical features, including retinal dystrophy, postaxial polydactyly, central obesity, renal anomalies, hypogonadism, and varying degrees of intellectual disability. Mutations in more than 26 known genes are implied. The clinical presentation is variable and often evolves over time, making early diagnosis challenging, particularly in the prenatal setting. Renal and urogenital abnormalities are common and may be the earliest detectable signs, with significant implications for morbidity and mortality.

Case Report - Anamnesis

28-year-old primigravida

No significant medical history

Relevant family history: brother with hypospadias, two sisters with patent ductus arteriosus

No significant medical history

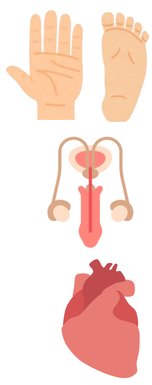
Relevant family history: first degree cousin with intellectual development disorder, polydactyly and unspecified renal and ophthalmological disorder

Non consanguineous couple

Case Report

13WG+1D: 1st scan > **20WG+6D: Morphology scan** > **Amniocentesis**

- Low risk for aneuploidies
- No alterations identified



Bilateral postaxial **polydactyly** of the hands and feet

Suspected **hypospadias**

Complete atrioventricular septal defect

No renal anomalies

PCR for common aneuploidies and **chromosomal microarray** analysis both yielding no abdnormal results

26WG: Referral to our Fetal Center > **36WG: Next-generation sequencing on fetal DNA Results** > **39WG+6D: Hospitalized due to premature rupture of membranes**

Clinic of fetal nephrourological pathology - Multidisciplinary evaluation: the constellation of anomalies was considered highly suggestive of a **ciliopathy.**

Identified two variants in the ***BBS12*** gene:

- one **patho**genic (c.1482_1485del)
- one of uncertain significance **(c.65T>C)**, which was later reclassified as likely **pathogenic**

Cesarean section for arrested labor: male, 3900g, Apgar scores 3/8/9, pH 7.260, base excess 1.6 mmol/L

All the prenatal findings were confirmed except for hypospadias

Conclusion

Early prenatal diagnosis of BBS through 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.

References

Forsyth RL, Gunay-Aygun M. Bardet-Biedl Syndrome Overview. 2003 Jul 14 [Updated 2023 Mar 23]. In: Adam MP, Feldman J, Mirzaa GM, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2025. Available from: https://www.ncbi.nlm.nih.gov/books/NBK1363/