

Technical and Clinical Implications of Maternal Chromosome Xp Deletion Identified by Prenatal Cell-Free DNA Screening

Background

- Turner syndrome is a genetic condition that affects approximately 1 in 2,500 newborn girls.¹
- The most common cause of Turner syndrome is the absence of an X chromosome. But variant forms of Turner syndrome can be caused by partial deletion of an X chromosome or by other X chromosomal abnormalities.
- Emerging evidence shows that maternal copy number variations (CNVs) can be detected by prenatal cell-free (cfDNA) screening.^{2,3}
- **Objective:** The investigators report a case in which maternal variant Turner syndrome was initially identified with prenatal cfDNA screening and confirmed with cytogenetic studies.

Case Description

- A blood sample from a 39-year-old woman at 11 weeks' gestation was submitted for prenatal cfDNA screening using massively parallel shotgun sequencing and advanced bioinformatics via the QNatal[®] Advanced test at Quest Diagnostics.
- The cfDNA sequencing result indicated a deletion at the end of the p arm of the X chromosome (chromosome Xp deletion).
- The deletion was suspected to be maternal in origin because the level of sequencing product at Xp was lower than expected for a fetal deletion (given a fetal fraction of 9.7%).
- Follow-up chromosomal microarray analysis using Affymetrix CytoScan[®] HD SNP-array platform and Chromosome Analysis Suite confirmed the presence of a 39.5-Mb maternal Xp deletion and a diagnosis of variant Turner syndrome.
- Based on these findings, diagnostic testing of the fetus was recommended to determine whether the fetus also carried the deletion.

Implications

- This case report adds to growing evidence that prenatal cfDNA screening can accurately identify maternal CNVs.
- As in this case, identification of maternal variant Turner syndrome can aid in appropriate clinical management of the patient and the fetus.

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References

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