

Confined Placental Trisomy 18 Mosaicism Detected by Maternal Serum and Prenatal cfDNA Screening

Background

- Maternal serum screening (MSS) and prenatal cell-free DNA (cfDNA) screening can each be used to identify fetuses at increased risk of trisomy.
- A fetus usually has the same genetic makeup as the placenta, and these techniques measure components that enter the mother's bloodstream from the placenta. Therefore, results from these tests may also happen to identify confined placental mosaicism (CPM), a condition in which the genetic makeup of the fetus differs from that of the placenta.^{1,2}
- Because CPM carries risks such as fetal growth deficiency during pregnancy, early identification can help guide care.³
- **Objective:** The investigators describe a case in which trisomy 18 was initially suspected based on MSS and prenatal cfDNA screening, but CPM was subsequently identified using follow-up cytogenetic testing.

Case Description/Details

- A sample from a pregnant woman was submitted to Quest Diagnostics for prenatal cfDNA screening via massively parallel shotgun sequencing.
 - The woman had previously undergone MSS, which revealed a 1:22 risk for trisomy 18.
 - Results from cfDNA screening were consistent with trisomy 18.
- Following prenatal cfDNA screening, the woman underwent ultrasound examination and amniocentesis with routine cytogenetic and microarray analyses.
 - No abnormalities were revealed by these tests, which rely on fetal, rather than placental, components.
- After delivery, microarray analysis of placental tissue revealed mosaic trisomy 18, consistent with CPM of trisomy 18.
- All the test results taken together indicated CPM for trisomy 18.

Implications

- This case underscores the need to consider CPM when using MSS and/or prenatal cfDNA screening, as both methods analyze components that originate from the placenta.

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Authors

Carrie Guy, Megan D Maxwell, James L Waurin, Arlene M Buller-Burckle, Douglas S Rabin, and Charles M Strom

Affiliation

Quest Diagnostics Nichols Institute, San Juan Capistrano, CA

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References

1. Towner DR, Shaffer LG, Yang SP, Walgenbach DD. *Prenat Diagn.* 2001;21:395-398.
2. Masuzaki H, Miura K, Yoshiura KI, Yoshimura S, Niikawa N, Ishimaru T. *J Med Genet.* 2004;41:289-292.
3. Lestou VS, Kalousek DK. *Arch Dis Child Fetal Neonatal Ed.* 1998;79:F223-F226.