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Discovering rare chromosomal abnormalities using WGS-based NIPT

In a recent study published in *Science Translational Medicine,* researchers utilized the power of WGS-based NIPT to identify rare chromosomal abnormalities and assess their impact on pregnancy outcome.[1]

- This large study included nearly 90,000 WGS-based noninvasive prenatal test (NIPT) results from 2 independent clinical laboratories. Researchers used a common quality control (QC) metric to identify appropriate samples for all chromosome analysis in an effort to quantify the frequency of various rare chromosomal abnormalities. Available clinical outcomes were compared with results from all chromosome analysis to better understand fetoplacental biology.
- The rate of rare autosomal trisomies (RATs) in the overall study population was 0.34%, which is consistent with studies of short-term chorionic villus sampling (CVS) culture[2] and smaller NIPT series.[3,4,5]
- Overall, 75% (39/52) of cases of single RATs with outcome data available were associated with relevant abnormal feto-maternal findings including miscarriage, intrauterine fetal demise, intrauterine growth restriction (IUGR), true fetal mosaicism (TFM), uniparental disomy (UPD), congenital anomaly, and constitutional maternal chromosomal abnormality.

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Reference

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3. Lau TK, Cheung SW, Lo PS, et al. Non-invasive prenatal testing for fetal chromosomal abnormalities by low-coverage whole-genome sequencing of maternal plasma DNA: review of 1982 consecutive cases in a single center. *Ultrasound Obstet Gynecol*. 2014;43(3):254-264.
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