

Genome-wide non-invasive prenatal testing using cfDNA: the detection of rare autosomal trisomies

Peter Benn

Dept. of Genetics and Genome Sciences, Univ. of Connecticut Health Center, Farmington, USA

Background:

Genome-wide non-invasive prenatal testing (gw-NIPT) refers to the prenatal identification, through cell-free DNA analysis, of large chromosome imbalances (usually >7Mb) that may involve any chromosome. gw-NIPT is an expansion of the massively parallel shotgun sequencing counting-based approach to NIPT. In addition to the chromosome abnormalities detected by other methods, gw-NIPT identifies rare autosomal trisomies (RATs), monosomies, and partial deletions and duplications.

The most frequent finding with gw-NIPT is the detection of a RAT which, in a non-mosaic state, will be associated with early fetal death. If mosaic, RATs will usually be confined to trophoblasts. Although some of these confined placental mosaics are associated with poor pregnancy outcomes, the overall risks are low. gw-NIPT also detects rare partial deletions and duplications, maternal chromosome imbalances, and rarely, maternal cancer or pre-cancerous chromosome imbalances.

Conclusion:

This presentation will review gw-NIPT and the recommended patient management following a positive result.