

Current Dogma for Antenatal Genetic Counseling: Implementing Cutting-Edge Screening and Diagnostic Tools

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Objectives

This presentation will begin with a brief review of the history of noninvasive prenatal screening (NIPS) followed by various and sundry uses for NIPS.

Methods

Genetic carrier screening panels will be reviewed to facilitate understanding in context of existing evaluations performed for the newborn screen. New NIPS methodologies and prospective future of NIPS will be presented. Select cases will be presented that illustrate knowledge of the provider in counseling and directed screening/testing.

Results

In Case 1, genome-wide cfDNA was selected, demonstrating 6.55Mb loss of material from chromosome 11q. Amniocentesis confirmed findings, noting the specific deletion was associated with risk for thrombocytopenia, platelet dysfunction and immunodeficiency. Patient's care transferred to our tertiary care center to facilitate delivery and postnatal care. In Case 2, patient's fetus had a 5.2mm nuchal translucency in first trimester screen and patient elected for NIPS and referral to our center for counseling. NIPS resulted at time of referral suggested "normal male." Options discussed and patient had single gene NIPS and amniocentesis for karyotype/microarray collected. Microarray/karyotype was normal, but the single gene NIPS demonstrated variant in PTPN11 associated with Noonan syndrome. In Case 3, counseling for a consanguineous couple led to expanded carrier screening. Both parental entities were carriers for R1070 mutation in cystic fibrosis transmembrane conductance regulator (CFTR). Notably, this mutation would have been missed on targeted mutational analysis and/or ethnic-based carrier screening.

Conclusions

Providers and patients have varying viewpoints and awareness regarding utility of classic prenatal screening, NIPS, single gene NIPS, and karyotyping/microarray. Without careful selection of the appropriate test, diagnoses may be missed and in effect thwart ability to provide anticipatory guidance and optimal care.