NIPS for microdeletions; an update

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Single-nucleotide polymorphism (SNP)-based non-invasive prenatal testing (NIPT) can help predict a subset of submicroscopic abnormalities associated with severe clinical manifestations. Retrospective review of actual clinical practice for a SNP based NIPT for over 80,000 referrals for 22q11.2 deletion syndrome and over 42,000 referrals for 1p36, cri-du-chat, Prader-Willi, and Angelman microdeletion syndromes will be described. A revised protocol that reflexively sequenced high-risk calls at a higher depth of read will be presented. The revised protocol substantially reduces false-positive results and can provide positive predictive values comparable to that seen for fetal aneuploidies.