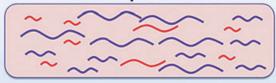
PRENATAL DIAGNOSIS

Maternal euploid cells Fetal trisomic 21 cells





Maternal plasma DNA



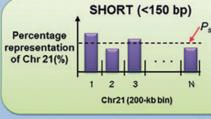


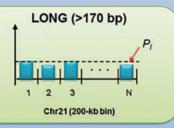
Paired-end sequencing and bioinformatics size classification





Determining the Chr 21 representation in the short and long DNA fragments for 200-kb bins







Significant overrepresentation of Chr 21 in short DNA over long DNA (i.e. P_s v.s. P_l)

Control free noninvasive fetal chromosome analysis
False positive NIPT results due to maternal CNVs
Smith-Lemli-Opitz carrier frequency
Prenatal diagnosis of congenital CMV

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