NEW METHODS IN PRENATAL DIAGNOSIS

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The development of genotyping and sequencing techniques has been dramatic during the recent years. Now it is possible to obtain a full view over an individual' s genetic landscape in the form of a million common single-nucleotide polymorphisms (SNPs) in single and affordable experiments. Similarly, the second generation sequencing technologies now make sequencing of whole genomes possible, but perhaps still not quite affordable for diagnostics. Clinical applications where there exist severa l genetically distinct populations of cells/DNA present special problems. One important example concerns prenatal diagnosis using maternal blood samples, containing only 3-6% fetal DNA. It is already possible to diagnose fetal abnormalities of known paternal origin this way. However, diagnosis of common fetal chromosome disorders such as trisomy 21 Down syndrome, where there is only a quantitative difference between maternal and fetal DNA still presents a challenge. I will in this talk describe some DNA circularization approaches that have the required capacity to genotype individual fetal DNA molecules in a large pool of maternal DNA. The circularization approach can thus be used for identification of fetal DNA in maternal blood plasma that have differential epigenetic signatures and therefore for the non-invasive prenatal genetic disease, including in particular common chromosome disorders such as trisomy 21 Down syndrome.

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