OVERVIEW

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NIPD for monogenic disorders

Newly developed non-invasive prenatal diagnosis (NIPD) tests based on analysis of cell free fetal DNA are allowing couples at risk of some single gene disorders to have prenatal diagnosis using a maternal blood test from nine weeks in pregnancy.

In the United Kingdom (UK), NIPD for autosomal dominant conditions such as achondroplasia, thanatophoric dysplasia, and Apert syndrome are now in routine clinical service, as is NIPD for paternal exclusion of cystic fibrosis. NIPD for recessively inherited single gene disorders is more complex as high background level of maternal mutant alleles must be taken into account. Currently the majority of these tests are done using next generation sequencing which is now lending itself to NIPD for recessive disorders such as congenital adrenal hyperplasia. The process by which we have developed these tests and gained approval for use in the UK National Health Service will be described, data relating to patients experience of NIPD will be presented and issues relating to implementation of a prenatal diagnostic service based on NIPD will be discussed.