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A secret book written in blood

Non-invasive prenatal testing (NIPT) could soon be available on the NHS according to the BBC News. This would be very good news for pregnant women with high risk pregnancies as invasive procedures such as amniocentesis carry a significant risk of miscarriage.

NIPT tests analyse the maternal blood. During pregnancy the placenta leaks foetal DNA (cffDNA) which circulates in the maternal bloodstream. As a result, a maternal plasma sample contains a mixture of foetal and maternal circulating DNA. Therefore the risk of an affected pregnancy can be estimated.The knowledge that circulating DNA could be found in the bloodstream dates back to 1948, but it was not until 1997 that it was successfully shown that the foetal Y chromosome could be found in the maternal blood of pregnant women carrying male foetuses.

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NIPT tests are typically provided by CLIA-certified centralised labs in the USA such as the verifi Prenatal Test performed by Verinata Health Inc (subsidiary of Illumina Inc) or the Maternity 21 Plus performed by Sequenom Laboratories. However, de-centralised NIP tests are gaining momentum in Europe. For example, Premaitha Ltd offers the first CE marked complete IVD product to clinical laboratories who wish to offer their own Non-Invasive Prenatal Screening Test (NIPT). The test is now being used by Genoma SA ("Genoma", Geneva, Switzerland) and St George's University Hospitals NHS Foundation Trust in London, UK.

NIPT tests also have the potential to uncover the mother's health problems due to genetic abnormalities such as DiGeorge syndrome (associated with learning difficulties and heart defects), reduced fertility due to a sex-chromosome abnormality or more surprisingly the tests can reveal if the mother has transplanted organs taken from male donors in the case of a pregnancy with a female foetus.

The analysis of cell free DNA is also gathering pace in oncology with the so-called 'liquid biopsies': when cancer cells die, they shed circulating tumour DNA (ctDNA) into the blood stream. This ctDNA is much harder to detect than cffDNA and tests have yet to become mainstream despite the fact that ctDNA was shown to be present in the blood back in 1977.

While there is acknowledgement of the great potential of liquid biopsy tests, their integration into everyday clinical practice has yet to occur. Not enough has yet been done to fully understand the accuracy of these tests and large-scale studies still need to be carried out in order to assess the test's efficacy.

It will take several years and substantial financial investment, but blood biopsies will be key to delivering personalised medicine in the next decade.

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