

## **Non-invasive prenatal diagnosis: from dream to reality**

In 1997, my group reported that fetal DNA was present in the plasma of pregnant women. This finding has opened up the field of non-invasive prenatal diagnosis. We have found that such fetal DNA is present in maternal plasma from the first trimester of pregnancy, at a surprisingly high mean fractional concentration of 15%. Over the last few years, we have shown that through the use of massively parallel DNA sequencing technologies, one can use this approach for the robust detection of fetal Down syndrome. This technology is now available in over 50 countries around the world and has been used in over 700,000 pregnant women. More recently, we have shown that the entire fetal genome, transcriptome and methylome can be sequenced from maternal plasma. Such developments have opened up many new clinical and research applications. In summary, fetal nucleic acids in maternal plasma has created a paradigm shift in prenatal medicine.

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