



**Non-invasive pre-natal testing (NIPT) can contribute to screening programmes, but cannot be considered diagnostic**

## Accuracy of NIPT using cell-free DNA for detection of Down's, Edwards and Patau syndromes

### Background

- Non-invasive pre-natal testing (NIPT) uses a sample of cell-free foetal DNA from the pregnant mother's blood to assess the risk of three genetic disorders in the foetus – Down's, Edwards and Patau syndrome.
- It is used widely across the world and has been rapidly adopted in the USA, with a decrease in the use of combined tests and invasive testing, such as amniocentesis or chorionic villus sampling.
- A clear summary of test accuracy is needed to ensure that results are not misinterpreted. This is especially important for those who would consider terminating a pregnancy on the basis of a positive NIPT test.
- Although a previous review of accuracy exists, it needs updating and does not provide a summary of findings.
- We conducted a systematic review and meta-analysis to measure the accuracy of NIPT and inform the decision on introducing the test into current foetal abnormality screening in the UK.

## Findings:

- In 100,000 pregnancies from the general obstetric population, NIPT could detect...

Syndrome	True Positives	False Positives
Down's	417 (out of 435)	94 (out of 99,565)
Edwards	89 (out of 102)	154 (out of 99,898)
Patau	40 (out of 52)	42 (out of 99,948)

- NIPT has very high sensitivity and specificity and can contribute to screening programmes, but cannot be considered diagnostic.
- Sensitivity for all three disorders was higher in high-risk pregnancies, in singleton pregnancies, and in the second and third trimester of pregnancy (week 13+).
- As NIPT is broadly accurate without the increased risk of invasive tests, it may be attractive to patients who are not considering a termination, but would like to find out if the pregnancy is affected by one of these syndromes.
- It is vital to follow a positive NIPT result with an invasive diagnostic test to confirm the presence of trisomy if the woman is considering a termination on this basis.
- A parallel economic evaluation suggests that the test is 'cost-neutral' and would result in a reduction of miscarriages associated with invasive tests.

**Reference: Taylor-Phillips S,** Freeman K, Geppert J, et al. Accuracy of non-invasive prenatal testing using cell-free DNA for detection of Down, Edwards and Patau syndromes: a systematic review and meta-analysis. *BMJ Open*. 2016; **6**: e010002. [<http://goo.gl/7Ymuom>]



## Recommendations for Practice

The UK National Screening Committee has endorsed the use of NIPT for Down's syndrome. However, communicating to clinicians and patients that it is not perfect will be key to ensuring safe implementation. Very careful consideration should be given to information provision and counselling.

### What is NIHR CLAHRC West Midlands?

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