



NIPT for Down (trisomy 21), Edwards (trisomy 18) and Patau syndrome (trisomy 13).

The West Midlands Regional Genetics laboratory is offering non-invasive prenatal testing (NIPT) as a screening test for common chromosome conditions; Down (trisomy 21), Edwards (trisomy 18) and Patau syndrome (trisomy 13). Screening for Turner syndrome (monosomy X) is also available where indicated by scan findings.

The Lucina NIPT test uses next generation sequencing and VeriSeq v2® test technology developed by Illumina. Cell free DNA (cfDNA) is extracted from maternal blood and sequenced by massively parallel sequencing. A log likelihood ratio (LLR) is generated for chromosomes 13, 18, 21, X and Y to determine fetal aneuploidy.

Patients should be made aware that this is a screening test and a high risk result does not mean that the fetus definitely has one of these conditions. **Therefore, high risk results should be followed up by an invasive prenatal diagnostic procedure. If the test result shows a low risk for any of the conditions, a small risk remains that the fetus may be affected.**

Test performance data;

Singleton pregnancies:

Trisomy 21: Sensitivity >99.9% (95% CI 97.1-100%), specificity 99.9% (95% CI 99.63-99.97%)

Trisomy 18: Sensitivity >99% (95% CI 91.4-100%), specificity 99.9% (95% CI 99.64-99.97%)

Trisomy 13: Sensitivity >99.9% (95% CI 87.1-100%), specificity >99.9% (95% CI 99.64-99.97%)

Twin pregnancies:

Trisomy 21: Sensitivity 96.4% (95% CI 86.4-98.9%), specificity 99.9% (95% CI 99.8->99.9%)

Trisomy 18: 95.7% (95% CI 68.3-99.4%), specificity >99.9% (95% CI 99.9->99.9%)

Trisomy 13: 93.6% (95% CI 64.1-98.9%), specificity >99.9% (95% CI 99.9->99.9%)

Referrals: Referrals can be made via a Clinical Genetics Department, Fetal Medicine Unit or by Specialist Midwives.

Sample requirements;

- The minimum gestation (by scan) for accepting a sample is 10+ weeks.
- 10ml venous blood in Streck tube is required for testing.
- Samples must reach the lab within 5 days of being drawn.
- Testing is available for both singleton and twin pregnancies.
- Fetal sexing information is not provided as part of this test.
- If there is a clinical indication from ultrasound scan findings, screening for Turner syndrome (monosomy X) can be performed. Please contact the lab for more details

Laboratory contact

- Dr Stephanie Allen, Consultant Clinical Scientist

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- Dr Elizabeth Young, Principal Clinical Scientist

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- General enquiries

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Cost and target reporting times

- Prenatal testing £275 (private referrals)
- Results are normally available within 7 calendar days of sample receipt

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