

All Wales Molecular Genetics Laboratory

Non-invasive prenatal testing analysis

Report on :			
DoB :		Address :	
Sex :		Lab No :	
Sample type : Blood plasma		NHS No :	
Date Rec'd :		Hospital No :	
Date reported :		Your ref :	
		Alt Hosp No :	

Reason for Referral :

Combined screening risk of 1 in 2 to 1 in 150

... weeks gestation by scan

Sample collection date:

Conclusion: High chance of trisomy 21 - INVASIVE TEST RECOMMENDED

Non-invasive prenatal testing (NIPT) showed a high chance of trisomy 21. The current pregnancy is therefore considered to be at high chance of being affected with Down Syndrome. It is recommended that this result is confirmed by invasive testing.

Non-invasive prenatal testing (NIPT) showed a low chance of aneuploidy for chromosomes 13 and 18.

This is a test to detect numerical abnormalities of these chromosomes only. This result will not detect trisomy and monosomy of other chromosomes, structural chromosome abnormalities, including imbalances of small segments, or mosaicism.

This test is a screening test only, and therefore false positive and false negative results can occur. Please refer to the test performance data below. False results may be due to biological factors including confined placental mosaicism (CPM), chromosome abnormalities in the maternal genome or the demise of a co-twin (vanished twin).

Analysed by:

Checked by:

Test details: Massively parallel shotgun sequencing of cell-free DNA extracted from maternal plasma, followed by assessment of the log likelihood ratio values for chromosome 13, 18 and 21 to determine fetal aneuploidy. Test performance data - Trisomy 21 sensitivity 100% (95% CI 47.8-100%), specificity 100% (95% CI 96.7-100%); Trisomy 18 sensitivity 100% (95% CI 63-100%), specificity 100% (95% CI 96.6-100%); Trisomy 13 sensitivity 100% (95% CI 29.2-100%), specificity 100% (95% CI 96.7-100%). The accuracy of the result can be affected by gestation. A minimum gestation of 10 weeks for testing is required. This test has not yet been accredited by UKAS.

Copies to:

Results are dependent on samples being correctly labelled and family relationships as indicated.
Please note, any remaining DNA will be stored in the laboratory.



Accredited Medical Laboratory
Reference No: 5585

Institute of Medical Genetics, University Hospital of Wales,
Heath Park, CARDIFF CF14 4XW.
Tel: 029 20742641, Fax: 029 20744059.

Sefydliad Geneteg Feddygol, Ysbyty Athrofaol Cymru,
Parc-y-Mynydd Bychan, CAERDYDD CF14 4XW.
Ffôn: 029 20742641, Ffacs: 029 20744059

website: <http://www.wales.nhs.uk/awmgs>

