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GWASANAETH i GYMRU

medical genetics
SERVICE FOR WALES

Cytogenetics Section
Genetic Diagnostic Laboratory

UKAS MEDICAL
Accredited Medical laboratory
Reference No: 8988

QF-PCR ANALYSIS

Name:		Lab No:	
		Date of Birth:	
Address:		NHS No:	
		Hospital:	
		Ward/Clinic:	
		Consultant	
Sample Type	**Pren Chorionic Villi**	Sub-type:	**Chorionic Villus**
Indication:	**First trimester combined NTand se**	Date Collected:	
		Date Received:	

REPORT

Report Summary: NO evidence of trisomy 13, 18 or 21 in chorionic villus sample

Report Interpretation

QF-PCR analysis showed NO evidence of trisomy 13, 18 or 21.

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

Discordance can occasionally occur between the results of CVS QF-PCR analysis and the results of karyotyping CVS cultures or fetal tissue, for the chromosomes tested. As is the case with direct CVS karyotype preparations, false positive/negative results are usually due to confined placental mosaicism.

Comparison of the chorionic villus sample and the maternal blood sample (no.) QF-PCR genotypes excluded the possibility of maternal tissue sampling.

Extracted DNA has been stored on this sample.

Basis of Test:

Quantitative fluorescence polymerase chain reaction (QF-PCR) analysis of DNA extracted from uncultured cells to identify trisomy for chromosomes 13, 18 and 21. The QF-PCR multiplex routine analysis includes 5 polymorphic microsatellite markers from chromosome 13, 6 markers from chromosome 18 and 4 markers from chromosome 21. The DNA is extracted from part of the dissected, finely cut, and trypsin-digested, homogenised material used for CVS culture to ensure that both cytotrophoblast and mesenchyme cells from all available villi are sampled. The possibility of a triploid genotype has been excluded by QF-PCR analysis. The results and their interpretations assume the sample received contains predominantly placental cells/tissue of zygotic origin.

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NHS CYMRU WALES

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Report by:

Pre-Registration Clinical Scientist

Authorised by:

Clinical Scientist

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Checked by:

Clinical Scientist

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Report on Lab No

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