geneteg feddygol
GWASANAETH i GYMRU

medical genetics
SERVICE FOR WALES

Cytogenetics Section
Genetic Diagnostic Laboratory

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:	**Screening risk positive using NIPT**	Date Collected:	
	Cystic hygroma on USS	Date Received:	

REPORT

Report Summary: Evidence of trisomy 21 in chorionic villus sample

Report Interpretation

QF-PCR analysis showed an abnormal result with evidence of trisomy 21, which is consistent with Down syndrome and with the fetal cystic hygroma detected on ultrasound scan. There was no evidence of trisomy 13 or 18 and no evidence of sex chromosome aneuploidy.

This result is for these chromosomes only. 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 confirmed sample identity and excluded the possibility of maternal tissue sampling.

A report on the full cytogenetic analysis of cultured cells will follow separately.

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 and sex chromosome aneuploidy. 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 informative chromosome 21 markers used in this case were: D21S1437 at 21q21.1, D21S1435 at 21q21.3, D21S1446 at 21q22.3 and D21S1411 at 21q22.3. The sex chromosome multiplex includes two XY chromosome markers, seven X chromosome only markers and two Y chromosome only markers. 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

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analysis. The results and their interpretations assume the sample received contains predominantly placental cells/tissue of zygotic origin.

Report by:
Pre-Registration Clinical Scientist

Authorised by:
Clinical Scientist
XX/XX/XXXX

Checked by:
Clinical Scientist

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