



*Cytogenetics Section  
Genetic Diagnostic Laboratory*



Accredited Medical laboratory  
Reference No: 8988

**QF-PCR ANALYSIS**

Name:	Lab No:
Address:	Date of Birth:
	NHS No:
	Hospital:
	Ward/Clinic:
	Consultant:
Sample Type <b>Pren AF</b>	Sub-type: <b>Amniotic Fluid</b>
Indication: <b>Screening risk positive using NIPT</b>	Date Collected:
	Date Received:

**REPORT**

**Report Summary: Evidence of trisomy 21**

**Report Interpretation**

QF-PCR analysis showed an abnormal result with evidence of trisomy 21, which is consistent with Down syndrome. QF-PCR analysis of a second sample aliquot, taken from the original amniotic fluid sample from this patient, confirmed sample identity.

There was no evidence of trisomy 13 or 18.

This result is for these chromosomes only. 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 amniocytes 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 informative chromosome 21 markers used in this case were: D21S1437 at 21q21.1, D21S1435 at 21q21.3, and D21S1411 at 21q22.3. The possibility of a triploid genotype has been excluded by QF-PCR analysis. The results and their interpretations assume the sample received contains predominantly fetal cells/tissue.

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**Report by:**

Pre-Registration Clinical Scientist

**Authorised by:**

Clinical Scientist

**Checked by:**

Clinical Scientist

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