



*Cytogenetics Section
Genetic Diagnostic Laboratory*



Accredited Medical Laboratory
Reference No: 8988

CYTOGENETIC ANALYSIS

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

REPORT

Karyotype: **47,XY,+21**

Summary: An abnormal male karyotype consistent with Down syndrome. A referral to the Prenatal Genetics Service is recommended.

An abnormal male chromosome complement was present due to an additional chromosome 21 homologue in all 20 cells examined from one culture established from the liquor of this patient. The karyotype is 47,XY,+21.

This karyotype is consistent with Down syndrome in a male fetus.

This result is consistent with that for the amniotic fluid QF-PCR test carried out on this sample, issued on XX/XX/XXXX.

Previous non-invasive prenatal testing (NIPT) showed a high chance of trisomy 21 for this pregnancy,

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and the report, no. XXXXXXXX, was issued on the XX/XX/XXXX.

Prenatal testing (including non-invasive prenatal testing NIPT) should be considered in any future pregnancy of this couple since there is an increased risk of aneuploidy. This would be available by referral to the Prenatal Genetics Service.

Please ensure that a copy of this report is placed in the infant's notes at birth.

In view of this finding, we recommend that this couple receive a referral to the Prenatal Genetics Service for counselling. This would be available from staff at the All Wales Medical Genomics Service.

Cytogeneticist:

Head of Section

Report Date

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