



*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 Chorionic Villi	Sub-type: Chorionic Villus
Indication: Screening risk positive using NIPT	Date Collected:
Cystic hygroma on USS	Date Received:

REPORT

Karyotype: **47,XX,+21**

Summary: An abnormal female karyotype consistent with Down syndrome. Genetic counselling is available.

A chromosome count of 47 with an additional chromosome 21 was observed in 14 cells examined from 2 cultures established from the chorionic villus sample of this patient, the karyotype being 47,XX,+21.

This karyotype is consistent with Down syndrome in a female fetus and with the presence of cystic hygroma on ultrasound scan.

This result is consistent with that for the chorionic villus 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 and the report no.

Copies to:



Cytogenetics Section
Genetic Diagnostic Laboratory

XXXXXXX was issued on XX/XX/XXXX.

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

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.

Genetic counselling is available from staff at the All Wales Medical Genomics Service should you wish to refer this family.

Cytogeneticist:

Head of Section

Report Date

Copies to: