AWMGS
All Wales Medical Genomics Service

Prenatal Cytogenetic Analysis

Chromosome analysis of prenatal samples using an appropriate investigation method (see **Analysis** below) depending upon the referral reason.

Please contact the laboratory prior to sending a prenatal sample

Referrals / Indications

- Abnormal ultrasound scan
- Higher chance serum screening result for trisomy 13/18 and/or 21
- Family history of a chromosomal anomaly

All referrals are processed as **urgent**

Sample Requirements

Amniotic fluid – 15-20ml
Chorionic villus sample (CVS)
Foetal blood – >0.5ml in lithium heparin

NB. 5ml maternal blood sample is required to accompany amniotic fluid samples referred for abnormal ultrasound scan and all CVS samples

Please label samples with three identifiers and date of collection
All samples must be accompanied by a completed request form
Consent for testing and sample storage is assumed when the request is received – it is the responsibility of the referring clinician to ensure that appropriate consent has been obtained

Analysis / Services

- Chromosome analysis by karyotyping
- Rapid aneuploidy testing by QF-PCR
- Fluorescence in situ hybridisation (FISH)
- Single Nucleotide Polymorphism Array (SNP array)

Contact Details

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Links

Orphanet
http://www.orpha.net/
OMIM
http://www.omim.org/
GeneTests
http://www.genetests.org/
Genetic Test Registry
http://www.ncbi.nlm.nih.gov/gtr/
Decipher
http://decipher.sanger.ac.uk/
Unique
www.rarechromo.org

Prices* & Turnaround Times (TAT)
*Available on request

Analysis	TAT
uploidy testing (QF-PCR)	3cd
romosome analysis	14cd
H analysis	14cd
NP array analysis	14cd