

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

<p style="text-align: center;">Referrals / Indications</p> <ul style="list-style-type: none"> • Abnormal ultrasound scan • Higher chance serum screening result for trisomy 13/18 and/or 21 • Family history of a chromosomal anomaly <p>All referrals are processed as urgent</p>	<p style="text-align: center;">Sample Requirements</p> <p>Amniotic fluid – 15-20ml Chorionic villus sample (CVS) Foetal blood – >0.5ml in lithium heparin</p> <p>NB. 5ml maternal blood sample is required to accompany amniotic fluid samples referred for abnormal ultrasound scan and all CVS samples</p> <p>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</p>
<p style="text-align: center;">Analysis / Services</p> <ul style="list-style-type: none"> • Chromosome analysis by karyotyping • Rapid aneuploidy testing by QF-PCR • Fluorescence in situ hybridisation (FISH) • Array Comparative Genomic Hybridisation (array CGH) 	<p style="text-align: center;">Contact Details</p> <p>All Wales Medical Genomics Service, Institute of Medical Genetics, University Hospital of Wales, Heath Park, Cardiff CF14 4XW Tel: 029 218 44674 Fax: 029 218 44043 Lab.genetics.CAV@wales.nhs.uk www.medicalgenomicswales.co.uk Accredited to ISO 15189:2012 (8988)</p>

Links

Orphanet
<http://www.orpha.net/>
EDDNAL
<http://www.eddnal.com/>
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
Rapid aneuploidy testing (QF-PCR)	3cd
Urgent Chromosome analysis	14cd
Urgent FISH analysis	14cd
Urgent array CGH analysis	14cd