

## QF-PCR Aneuploidy Screening

Quantitative Fluorescence-Polymerase Chain Reaction (QF-PCR) is the analysis method used to test for trisomy 13, 18 or 21, and for sex chromosome aneuploidy screening. This is a targeted test which is usually followed by further analysis (karyotyping or array CGH).

Please indicate on the request form if there is a specific clinical need for a sample to be processed urgently.

*Please contact the laboratory prior to sending a prenatal sample*

### Referrals / Indications

- All prenatal referrals (urgent)
- Neonates with a suspected aneuploidy of chromosomes 13, 18 or 21 (urgent)
- Neonates with indeterminate gender / ambiguous genitalia (urgent)
- Post mortem solid tissue referrals (non-urgent)

### Follow-up Analysis

Aneuploidy screening is a targeted test which is usually followed by further analysis (requiring appropriate samples to be sent to the laboratory):

### Sample Requirements

Prenatal: Amniotic fluid – 15-20ml

Chorionic villus sample (CVS) – by arrangement (5ml maternal blood in EDTA also required)

Foetal blood – >0.5ml EDTA & lithium heparin

Postnatal: Blood in EDTA & lithium heparin

Child index case – >1ml

Adult index case – 5ml

Solid tissue: POCs, foetal skin or other solid tissue samples

– send in sterile transport medium supplied by laboratory or normal saline (sterile)

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

**Prices\* & Turnaround Times (TAT)**

\*Available on request

Analysis	TAT
Rapid aneuploidy testing	3cd
Routine aneuploidy testing	42cd

**Contact Details**

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Institute of Medical Genetics,  
University Hospital of Wales,  
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Accredited to ISO 15189:2012  
(8988)

**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](http://www.rarechromo.org)