

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 SNP array).

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)

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

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):

- Prenatal – chromosome analysis (karyotyping or SNP array)
- Neonatal – chromosome analysis (karyotyping or SNP array)
- Post mortem solid tissue – chromosome analysis (SNP array)

Prices* & Turnaround Times (TAT)

*Available on request

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

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