Validation of copy number variants using real-time quantitative PCR (qPCR)

Background

Genomic techniques such as exome sequencing, whole genome sequencing (WGS) and array comparative genomic hybridisation (aCGH) have become the frontline tests for a vast range of referrals within genetics laboratories. A significant challenge of these techniques is the ability to confidently identify small scale copy number variants (CNVs). This can result in the need for further testing to fully interpret or validate complex findings. We have an in-house bespoke real-time quantitative PCR (qPCR) assay for validating CNVs, including single exon imbalances. Once confirmed, the qPCR test provides a targeted and economical option for detecting the imbalance in family members.

Referral requirements

Please include the following information on any referral requests to allow efficient primer design:

- Genomic coordinates of the imbalance with genomic build
- Whether a deletion or duplication is expected
- Expected HGVS nomenclature and reference sequence

Molecular Analysis

Primers are specifically designed to target regions within the suspected CNV and are validated before testing on patient samples. If the designed primers do not pass internal QC, we will notify the requester that primer design has not been possible. A minimum charge will be payable to cover assay design. This technique is not available for mosaic CNVs.

Please note there are two levels of service – the first is confirmation of the CNV **only** with a technical report and second is confirmation, interpretation and reporting of the CNV.

Test (price available on request)	TAT (Calendar days)*
Proband confirmation and technical report only	42
Trio confirmation and technical report only	42
Proband confirmation and interpretive report	42
Trio confirmation and interpretive report	42

For urgent requests please contact the laboratory in advance to prepare the assay and indicate this on the request form.

*The turnaround time of 42 cd starts on the receipt of primers and does not include time for primer design and validation. We aim to complete each case within a total of 56 cd of sample receipt.

Contact Details

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Sample Requirements

Blood – 5ml in EDTA (1ml neonates/infants) or >200ng extracted DNA
Please contact lab prior to sending a prenatal sample.
Please label samples with three identifiers and date of collection
All samples must be accompanied by request form. Consent for testing & DNA storage is assumed when request for test received.

Links

Orphanet http://www.orpha.net/
OMIM http://www.omim.org/
GeneReviews https://www.ncbi.nlm.nih.gov/books/NBK1116/
Genetic Test Registry http://www.ncbi.nlm.nih.gov/gtr/
Decipher http://decipher.sanger.ac.uk/
Unique www.rarechromo.org