Microarray user guide

Introduction

Genome-wide chromosomal microarray (CMA), such as array-CGH and SNP microarray, will detect copy number changes (gains and loss of genetic material) at a higher resolution than conventional karyotyping. These imbalances are referred to as copy number variants (CNVs). In addition, SNP microarray analysis can identify regions with absence of heterozygosity (AOH), which may indicate isodisomy of known imprinting regions and can indicate ploidy level changes (e.g. triploidy).

The aim of microarray analysis is to detect and report pathogenic CNVs. However, it may also identify variants of unknown clinical significance (VUS) or pathogenic CNVs which are not associated with the specific reasons for referral (i.e. incidental findings).

In July 2021 the microarray platform used at the WGLS switched from array-CGH (OGT's CytoSure Constitutional v3 (60K) oligo array) to a SNP microarray design (**Illumina's Infinium Global Screening Array (Cyto) 700K SNP microarray**).

Referrals

Microarray testing can be offered to patients with clinical features highly suggestive of a chromosomal disorder, or with certain congenital abnormalities, and can be used to test fetal losses with specified clinical indications. Please refer to the National Genomic Test Directory for the clinical indications associated with microarray testing to determine eligibility.

Sample requirements

- Postnatal samples: 5ml blood taken into EDTA (>1ml from neonates)
- Tissue samples: see 'solid tissue user guide'

*please contact the laboratory if you require an alternative sample type to be tested

Parental blood samples may be requested to determine the inheritance pattern and thus aid variant interpretation and/or to determine recurrence risks.

Technical

The mean OMIM gene resolution of the 700K GSAcyto SNP microarray is ~10kb; CNVs below this threshold may not be detected. The microarray will not detect balanced chromosome rearrangements, single nucleotide variants or triplet repeat expansions, the genomic position or orientation of copy number gains, and is limited in detecting mosaicism (gonadal mosaicism cannot be detected). AOH may indicate uniparental isodisomy or regions identical by descent; the microarray cannot detect heterodisomy. Only regions of AOH involving known imprinting disorders will be reported. Interpretation is based on the evidence available in the literature and various databases at the time of analysis. CNVs are reported in accordance with best practice guidelines and laboratory policies; therefore, some CNVs will not be reported.

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Microarray technical processing for the WGLS is carried out by the West Midlands Regional Genetics Laboratory whilst analysis, interpretation and reporting is undertaken by the WGLS using NxClinical (Bionano) and the FASST2 CNV calling algorithm on human genome build GRCh37.

Turnaround times

We aim to report routine cases within 42 days and urgent cases within 14days.

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