Single Nucleotide Polymorphism (SNP) Array Service for Developmental Disorders and Epilepsy/Seizure Disorders

(High-resolution chromosome analysis)

Array analysis by SNP array will identify genomic copy number imbalance at a level of resolution significantly higher than that achieved by conventional karyotyping and will detect microdeletions and microduplications associated with known cytogenetic syndrome regions. In addition, SNP arrays can also detect regions of homozygosity (ROH), which is observed in a proportion of uniparental disomy (UPD) cases. SNP array is currently employed to investigate chromosomal imbalances in patients presenting with learning disability/developmental delay and/or multiple congenital abnormalities and individuals with early onset or syndromic epilepsy conditions.

Test Details

On May 16th 2022, array analysis has moved from array comparative genomic hybridization (CGH) to a SNP array platform. The current platform is the Illumina Infinium CytoSNP-850K BeadChip array. The analysis software is BlueFuse Multi v4.5 giving a backbone resolution of ~50 kb (kilobases) and a gene targeted resolution of ~20Kb. All reports are currently based on Genome Build GRCh38.

Further information about the advantages, limitations and counselling considerations for SNP array testing can be found here:
https://medicalgenomicswales.co.uk/images/awmgsdownload/PD-GEN-SNParrayUpdate.pdf

Clinical Referral Criteria

The appropriate criteria for SNP array testing include:

For syndromic developmental disorders:

- Moderate to severe developmental delay (please specify e.g. growth, motor, speech etc. as separate features)

PLUS one or more of the following:

- Behavioural problems and autism
- Dysmorphism
- Congenital anomalies

For syndromic intellectual disability disorders:

- Moderate to severe intellectual disability

PLUS one or more of the following:

- Behavioural problems and autism
- Dysmorphism
- Congenital anomalies

For syndromic congenital malformations and/or dysmorphism without intellectual disability:

- Presence of congenital abnormalities affecting **two** or **more** organ systems **OR** single organ system congenital abnormality **PLUS** dysmorphism

For early onset or syndromic epilepsy disorders:

- Onset under 2 years, **OR**
- Clinical features suggestive of specific genetic epilepsy, for example Dravet syndrome, **OR**
- Additional clinical features: intellectual disability, autism spectrum disorder, structural abnormality (e.g. dysmorphism, congenital malformation), unexplained cognitive/memory decline
- Testing may occasionally be appropriate where age of onset is between 2 and 3 years and following clinical agreement by a specialist MDT.

For further information, please refer to: https://www.england.nhs.uk/wp-content/uploads/2018/08/Rare-and-inherited-disease-eligibility-criteria-version-6-January-2024.pdf

For non-clinical genetics referrals, please provide a 'SNP Array and Fragile X Test Referral form' completed with as much detail as possible relating to the presenting phenotype in your patient. The clinical information provided is used to assess suitability for testing and for the interpretation of test findings.

Link to SNP array referral form:
https://medicalgenomicswales.co.uk/images/awmgsdownload/PD-GEN-DevDelReq.pdf

Autism as the sole clinical indication does not meet the clinical criteria for SNP array testing as part of this pathway.

For research testing please contact the laboratory to discuss requirements.

Sample requirements

- Child index case ->1ml blood in EDTA & LithHep
- Adult index case -> 4ml blood in EDTA & LithHep

Please label samples with three identifiers and date of collection

All samples must be accompanied by two fully completed request forms – general lab request (purple) form and specific SNP array and fragile X Test Request form

Consent for genetic testing and DNA storage is assumed when the request for testing 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 (calendar days)
SNP array testing (routine)	42
SNP array testing (urgent)	14

Contact Details

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Medicalgenomicswales.co.uk
This test is not yet accredited UKAS to ISO15189 (2012)

For urgent **clinical queries** please contact the on-call consultant on 029 218 42577

Links

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