AWMGS
All Wales Medical Genomics Service

SNP Array testing for the All Wales Psychiatric Genomics Service (AWPGS)

SNP array testing is employed to identify copy number imbalances in patients referred to the AWPGS, including microdeletions and microduplications associated with known cytogenetic syndrome regions.

Test details

The current SNP array platform is the Illumina Infinium CytoSNP-850K v1.2 BeadChip array. The analysis software is BlueFuse Multi v.4.5 giving a backbone resolution of ~50kb (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:

PD-GEN-SNParrayUpdate.pdf (medicalgenomicswales.co.uk)

Clinical Referral Criteria

Please refer to PGS_referral_criteria_2022.pdf (medicalgenomicswales.co.uk) for the appropriate criteria for SNP array testing through the AWPGS.

To refer an individual to the AWPGS, please complete the following form:
PGS_referral_form_2022.pdf (medicalgenomicswales.co.uk)

Sample Requirements

- Blood – >4ml blood in EDTA & LithHep

Please label samples with three identifiers and date of collection
All samples must be accompanied by a request form
Consent for testing & DNA storage is assumed when request for test 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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https://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 Testing Registry
http://www.ncbi.nlm.nih.gov/gtr/
Decipher
http://decipher.sanger.ac.uk
Unique
www.rarechromo.org