AWMGS
All Wales Medical Genomics Service

Prenatal Service: Abnormal Scan
Single Nucleotide Polymorphism (SNP) Array Service

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 invasive prenatal samples from patients presenting with an abnormal ultrasound scan.

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 v1.2 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

Inclusion criteria:	Exclusion criteria:
• Scan abnormalities where QF-PCR is normal o Increased nuchal translucency (NT) >3.5mm o Structural anomalies	• High risk combined or Quad screening result • Increased maternal age • Parental anxiety

Contact Details	Sample requirements
All Wales Medical Genomics Service Institute of Medical Genetics University Hospital of Wales Heath Park, Cardiff, CF14 4XW Tel: 029 218 44674 Fax: 029 218 44043 Lab.genetics.cav@wales.nhs.uk Laboratory website: http://www.wales.nhs.uk/AWMGS/ This test is not yet UKAS accredited to ISO15189 (2012)	• CVS (12 to 15 weeks' gestation) • Amniotic fluid – 15-20ml (15+ weeks' gestation) • Maternal sample – 5ml blood in EDTA Please label samples with three identifiers and date of collection **All samples must be accompanied by a fully completed 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

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

Prices* & Turnaround Times (TAT)
* Available on request
Patients in Wales are centrally-funded

*For research testing please contact the laboratory
to discuss your requirement*

Links

Orphanet
http://www.orpha.net/
OMIM
http://www.ncbi.nlm.nih.gov/omim/
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

Analysis	TAT
SNP array prenatal testing	14cd

Prenatal SNP Array Testing Pathway