Bristol Genetics Laboratory

North Bristol NHS Trust

# MICROARRAY COMPARATIVE GENOMIC HYBRIDISATION (CGH)

**Contact details:**
Bristol Genetics Laboratory (BGL)
Pathology Sciences and Bristol Genetics
Southmead Hospital
Bristol, BS10 5NB
Enquiries: 0117 4146168
FAX: 0117 4146464
nbn-tr.geneticsenquiries@nhs.net

www.nbt.nhs.uk/genetics

**Head of department:**
Eileen Roberts FRCPath

**Service Lead:**
Lisa Burvill-Holmes DipRCpath
lisa.burvill-holmes@nbt.nhs.uk

**Sample Required:**
Adult: 5ml blood in EDTA and Lithium Heparin
Paediatric: >1ml in EDTA and Lithium Heparin
Prenatal: please discuss with laboratory

Samples should be accompanied by a FULLY completed request form (available as download at www.nbt.nhs.uk/genetics or from the laboratory).

Please include details of test, family history, address and POSTCODE, NHS number, referring clinician and unit/hospital.

**Consent and DNA Storage:**
All genetic testing requires consent. It is the responsibility of the referring clinician to ensure that appropriate consent has been obtained.

DNA is stored from **all** patients unless consent for this is specifically denied.

Stored samples may be used for quality assurance purposes and may be used anonymously for the development of new tests for the disorder in question.

## Clinical Background and Genetics

- Microarray CGH is the comparative, competitive hybridisation of test (patient) and reference genomic DNA to probes arrayed onto a glass slide (microarray)
- This allows robust detection of copy number imbalance (loss or gain of genetic material) across the entire genome at a very high resolution (i.e. small regions of loss/gain of material)
- Copy number imbalance plays a significant role in the aetiology of learning disability (LD) and multiple congenital anomalies (MCA) and microarray CGH has been reported to detect copy number imbalance in 15-20% of this cohort (Miller *et al*, AM J Hum Genet 2010; 86,749-764)

## Service offered

- Diagnostic:
  - Routine diagnostic analysis is undertaken using an 8x60K, ISCA compliant, oligonucleotide microarray platform. The probe coverage varies across the genome and is highest across the exons and introns of genes important in the aetiology of developmental delay. Average resolution varies between 189kb and 663kb depending on the possible pathogenicity of the region. Some regions may have increased or reduced resolution depending upon syndromic significance or published clinical associations, allowing the robust detection of very small pathogenic imbalance
  - Prenatal diagnosis is undertaken on pregnancies with abnormalities detected by ultrasound scan.and uses the same microarray platform – Analysis and reporting is in accordance with the RCPath/RCOG Recommendations for the use of chromosome microarray in pregnancy (June 2015)
  - Samples from pregnancy losses may also be analysed: duplications <3Mb and deletions <1Mb may not be reported.
- Research:
  - Alternate microarray solutions, including custom designs, gene expression microarrays and genotyping microarrays may be offered. Please contact the laboratory to discuss this further.

## Referrals

- Diagnostic testing: referrals from patients with learning disability, developmental delay and/or multiple congenital abnormalities/dysmorphism
- Research testing: microarray analysis is applicable to a variety of cohorts. Please contact the laboratory to discuss specific projects in more detail

## Target reporting Time

- Routine referrals are reported within 4 weeks
- Urgent referrals are reported within 2 weeks

## Quality

- BGL participates in the *UKNEQAS* scheme (and has UKGTN approval) for this service.

## Clinical Advice

- If clinical discussion is required we would recommend contact with your local Clinical Genetics service.

Information document No.56 Version 7
Active date of this version 16/07/2017
**DETAILS CORRECT AT DATE OF PRINTING ONLY.**
Approved by: Lisa Burvill-Holmes

CPA
Accredited Medical Laboratory
Reference No: 2907