Chromosomal Microarray

✓ Can detect copy number changes down to 200,000 DNA basepairs (0.2Mb)
✓ Provides comprehensive whole genome coverage

INTRODUCTION

Chromosomal microarray (CMA, or Molecular karyotype) is an advanced technique in genetic testing that detects copy number changes in a person's chromosomes at a much higher resolution than conventional chromosome analysis (karyotype). Chromosomal microarray can detect copy number changes down to 200,000 DNA basepairs (0.2 Mb) compared to a resolution of 5-10 million DNA basepairs (5-10 Mb) for conventional cytogenetic analysis.

Single Nucleotide Polymorphism microarray can detect gains (duplication) and loss (deletion) of DNA segments and large regions of homozygosity. The chromosomal microarray platform used by TML Pathology provides comprehensive whole genome coverage to detect microduplication/microdeletion syndromes, aneuploidy, sub-telomeric deletions and duplications. It can also be used to define the genes involved in previously identified chromosome imbalances. This technology does not detect balanced chromosomal alterations, low level mosaicism, point mutations and imbalances of regions not represented on the microarray platform.

INDICATIONS FOR ORDERING

Chromosomal microarray is now regarded as the best practice first line test for patients with intellectual disability, two or more congenital abnormalities, autism spectrum disorder or developmental delay. This technique represents a significant advance in cytogenetic testing with an increase in the diagnostic rate in these clinical settings (15% CMA vs. 2-3% by conventional cytogenetics).

SINGLE NUCLEOTIDE POLYMORPHISM MICROARRAY PLATFORM

TML Pathology uses a Single Nucleotide Polymorphism based microarray platform that is well applied for diagnostic applications. It has over 750,000 probes covering 36,000 genes, all constitutional genes in the Clinical Genome Resource (ClinGen) database, cancer genes, and over 12,000 autosomal and X chromosome OMIM genes. It will detect chromosome imbalances including duplication, deletion and aneuploidy. Single Nucleotide Polymorphism based array platforms will also identify large regions of homozygosity for the detection of syndromes involving imprinted genes such as Prader Willi and Angleman syndromes.

TESTING CRITERIA

The laboratory is accredited to National Pathology Accreditation Advisory Council (NPAAC) standards. The criteria for reporting abnormalities is established according to the Human Genetic Society of Australasia (HGSA) guidelines, and is based on gene content, size of duplication/deletion and clinical significance.

HOW TO ORDER

Request *'Chromosomal Microarray'* on a TML Pathology request form.

SPECIMEN REQUIREMENTS

Neonates-5yrs	1-2ml whole blood in EDTA paediatric tube
Children >5yrs	5ml whole blood in EDTA tube
Adults	5ml whole blood in EDTA tube

TURNAROUND TIME

21 – 28 business days.
Urgent results can be pre-arranged with the laboratory (refer to contact details below). All reports are sent to the referring clinician or clinical geneticist.

COST

A rebate is available subject to Medicare guidelines and criteria.

FURTHER INFORMATION

Dr Nicole L.N. Chia PhD, FHGSA, FFSC(RCPA)
Clinical Scientist, Genetics Department
Genomic Diagnostics (QLD)
T: (03) 6108 9900 / T: (03) 6711 2000

Dr Kym Mina MBBS, PhD, FRCPA (Genetics)
Director of Genomics
Healius Pathology

Genomic Diagnostics NATA ACCREDITED

Healius Pathology Pty Ltd (ABN 84 007 190 043) t/a TML Pathology PUB/MR/1203_V4_Mar22