Prenatal Microarray

An advanced technique in genetic testing that detects copy number changes in a person's chromosomes at a much higher resolution than conventional analysis (karyotype)

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 analysis (karyotype). Chromosomal microarray can detect copy number changes down to 200,000 DNA basepairs (0.2Mb) 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 segments of DNA and large regions of homozygosity. The chromosomal microarray platform used by our laboratory provides comprehensive whole genome coverage to detect microduplication/microdeletion syndromes, aneuploidy and triploidy.

INDICATIONS FOR ORDERING

Prenatal testing by microarray is the preferred test where there are abnormal ultrasound findings or there is concern of a chromosomal imbalance. This may also include family history of chromosome rearrangement or abnormal pregnancy screening results.

SINGLE NUCLEOTIDE POLYMORPHISM MICROARRAY PLATFORM

Our laboratory uses a Single Nucleotide Polymorphism based microarray platform that is well applied for diagnostic applications. It has over 750,000 probes covering over 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 Angelman 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. This test may be used in conjunction with other cytogenetic screening tests including fluorescence in-situ hybridisation and conventional karyotyping.

This test does not detect balanced chromosomal alterations, low level mosiacism and some forms of tetraploidy. In addition, point mutations and imbalances of regions not represented on the microarray platform will not be identified and may require testing by alternative methods.

HOW TO ORDER

Request *'Prenatal Microarray'* on a QML Pathology form.

SPECIMEN REQUIREMENTS

Amnio	Minimum of 2x10ml amniotic fluid
CVS	25-40mg branched, clean villi
Parental Samples	**Parental samples required** 3-5ml whole blood in Lithium heparin tube AND 3-5ml whole blood in EDTA tube

Note: A conventional karyotype test will be performed if the samples are contaminated with blood.

TURNAROUND TIME

10 – 15 business days. All testing and requests for urgent results should be pre-arranged with the laboratory.

COST

A rebate is available subject to Medicare guidelines and criteria.

FURTHER INFORMATION

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