

Check list for submitting samples

- Patient EDTA blood sample ($\geq 2\text{mls}$)
- Patient heparin blood sample ($\geq 2\text{mls}$)
- Patient Clinical Submission Form A
- Patient Consent Form B

Points to note

- The test can only detect changes in gain or loss of genomic DNA material. Balanced rearrangements and small DNA sequence changes including point mutations and small deletions, duplications and insertions will not be identified.
- Low level mosaicism may be undetected.
- Specific genetic syndromes can be caused by other changes apart from deletions and duplications (e.g. Angelman and Prader-Willi syndromes). If a specific disorder is suspected clinically then further testing after microarray (DNA sequence analysis, methylation studies etc) may be appropriate.
- Copy number variants will only be detected for regions present on the microarray.

Chromosome Microarray Analysis

**A new technique revolutionizing
the field of genetic diagnosis**

Information for Doctors



For further information contact:
Dr Karen Woodward, Kirralee Patton or
Dr Narelle Hadlow
Molecular Genetics / Cytogenetics
Western Diagnostic Pathology

Phone: (08) 9317 0999

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74 McCoy Street, Myaree, WA
Telephone: (08) 9317 0999



Chromosome Microarray Analysis

What is Chromosome Microarray Analysis (CMA) or Microarray?

- Microarray is a new test for detecting gains or losses of chromosome material not visible by conventional cytogenetic analysis.
- Microarrays can identify abnormalities in patients with unexplained developmental delay/intellectual disability (DD/ID), multiple congenital abnormalities, autism spectrum disorder and apparently normal karyotype.
- Up to 20% more abnormalities can be detected by microarray after conventional testing.
- The entire genome is examined in a single test at a higher level of resolution than previously possible by karyotyping.
- The gains (duplication) or losses (deletion) of DNA identified are termed copy number variants (CNVs).
- The test uses genomic DNA microarrays containing more than a million short DNA sequences known as oligonucleotides.
- The test is not designed to detect small DNA base pair changes such as the common mutations causing Cystic Fibrosis and Hereditary Haemochromatosis.

What genetic diseases can be detected by Chromosome Microarray Analysis?

- Our microarrays are targeted to detect more than 100 known genetic disorders caused by CNVs. This includes known microdeletion / duplication syndromes (e.g. DiGeorge syndrome, Williams-Beuren syndrome, Smith-Magenis syndrome, Miller-Dieker syndrome) and pericentromeric and subtelomere regions.
- The whole genome is analysed and atypical deletions and duplications can also be identified.

What to consider before testing?

Before offering testing to patients the following are recommended:

- Confirm karyotype
- Confirm Fragile X testing normal
- Fill out patient Clinical Submission Form A
- Witness patient Consent Form B
- Referral for genetic counselling if appropriate

Details of the patient's clinical features are important in interpreting test results and should be submitted with blood samples.

Who should be tested?

- Patients suspected to have a genetic condition with possible genomic imbalance.
- Clinical indication for testing includes one or more of the following and a normal karyotype:
 - ▶ Unexplained developmental delay/intellectual disability
 - ▶ Dysmorphic features
 - ▶ Multiple congenital anomalies
 - ▶ Autism or unexplained autistic features

Microarray results and testing of parents

- The results will be classified into 3 categories as shown below.
- Parental blood samples can often clarify findings when the patient results are inconclusive.

