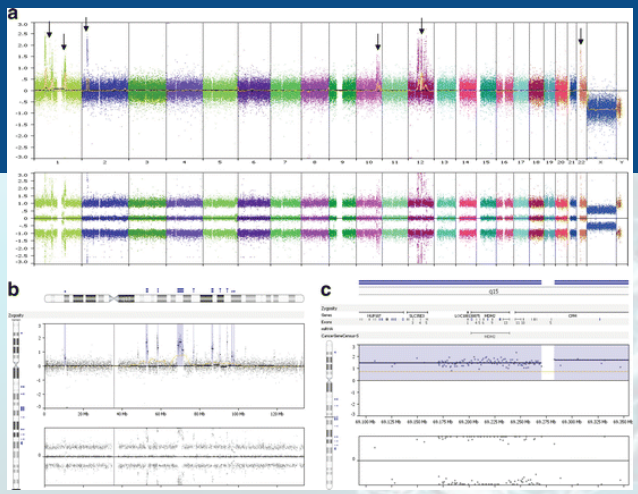
RML
MEHROTRA PATHOLOGY
Committed To Diagnostic Excellence

Turn around time (TAT):
7 Days

CHROMOSOMAL MICRO ARRAY

What is a Chromosomal Microarray (CMA)?

- It is a microchip-based testing plateform that allows automated analysis of many peices of DNA at once.
- CMA analysis offers the capacity to examine the whole human genome in a single chip with high resolution. It offers un paralled screening for deletions, duplications, loss of heterozygosity for all chromosomes.
- CMA chips use probes that hybridize with specific Chromosomal regions to detect copy number variations (CNV).
- CMA offers a combinations of CNV and single Nucleotide Polymorphism's (SNP)



What are the advantages of CMA in clinical practice?

Compared to Karyotyping, CMAs offer the following advantages:

- Higher resolution
- Nearly double the diagnostic yield
- Multiplexing/throughput is possible
- Detection of maternal cell contamination
- Detection of uniparental disomy

← Fig:1 Karyotype View

Registered Office: A-21A, Nirala Nagar, Lucknow-226020 | Corporate Office : B-171 Nirala Nagar, Lucknow-226020
Email : rmllabs@hotmail.com | Mob.no: 7991602001, 7991602002, 7991602003

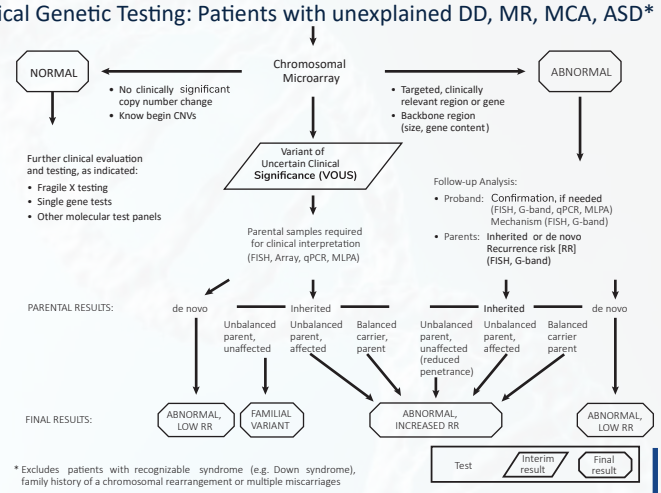
Which all conditions can CMA detect?

Conditions characterized by mental retardation such as:

- Angelman Syndrome
- Wolf-Hirschhorn Syndrome
- Williams Syndrome
- DiGeorge Syndrome
- Prader-Willi Syndrome

- Accurate identification of a number of chromosome disorders early in pregnancy, including virtually all known microdeletion and microduplication syndromes.
- Detection of a variety of gains or losses towards the ends (telomeres) of the chromosomes, the important causes of many developmental disability syndromes.

Clinical Genetic Testing: Patients with unexplained DD, MR, MCA, ASD*



* Excludes patients with recognizable syndrome (e.g. Down syndrome), family history of a chromosomal rearrangement or multiple miscarriages

*International standard cytogenetic array (ISCA) consortium

** David T. Miller, Margaret P. Adam, Swaroop Aradhya, et al. "Consensus Statement: Chromosomal Microarray Is a First-Tier Clinical Diagnostic Test for Individuals with Developmental Disabilities or Congenital Anomalies" The American Journal of Human Genetics, Volume 86, Issue 5, Pages 749-764 (May 2010)

How the test is done?

A blood sample is preferred for microarray analysis. Sometimes spit (saliva), tissue sample, POC, Amniotic Fluid and chorionic villi may be used. This test compares the patient's sample to a normal control sample to find very small missing or extra chromosome pieces that cannot be seen under a microscope. The test does not show structural changes in chromosomes.

Test sample requirements

Blood (3-5 ml in EDTA tubes)
Amniotic Fluid (AF)
Chorionic Villi (CVS)
Product of Conception (POC) or (minimum 200mg of POC)

Required forms:

- Relevant clinical information including all the clinical presentations and symptoms
- Test request form

Registered Office: A-21A, Nirala Nagar, Lucknow-226020 | Corporate Office : B-171, Nirala Nagar, Lucknow
Email : rmllabs@hotmail.com | Mob.no: 7991602001, 7991602002, 7991602003