ACTIA
Inherited Genetics

MEDGENOME

Chromosomal Micro Array (CMA)

CAP ACCREDITED
COLLEGE of AMERICAN PATHOLOGISTS

Actia

Inherited Genetics

The understanding of pathology at a molecular level is critical for identification of many diseases and their subtypes. Precision in diagnosis, including the identification of disease subtypes directly influences treatment and patient outcomes.

Actia from MedGenome provides an end-to-end integrated solution to clinical genomics in India and is highly focused on the Indian population. Actia has been delivering actionable genetic insights for inherited genetic conditions enabling happier outcomes.

What is a Chromosomal Microarray (CMA)?

- It is a microchip-based testing platform that allows automated analysis of many pieces of DNA at once.
- CMA analysis offers the capacity to examine the whole human genome in a single chip with high resolution. It offers unparalled 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 combination of CNV and Single Nucleotide Polymorphism's (SNP)

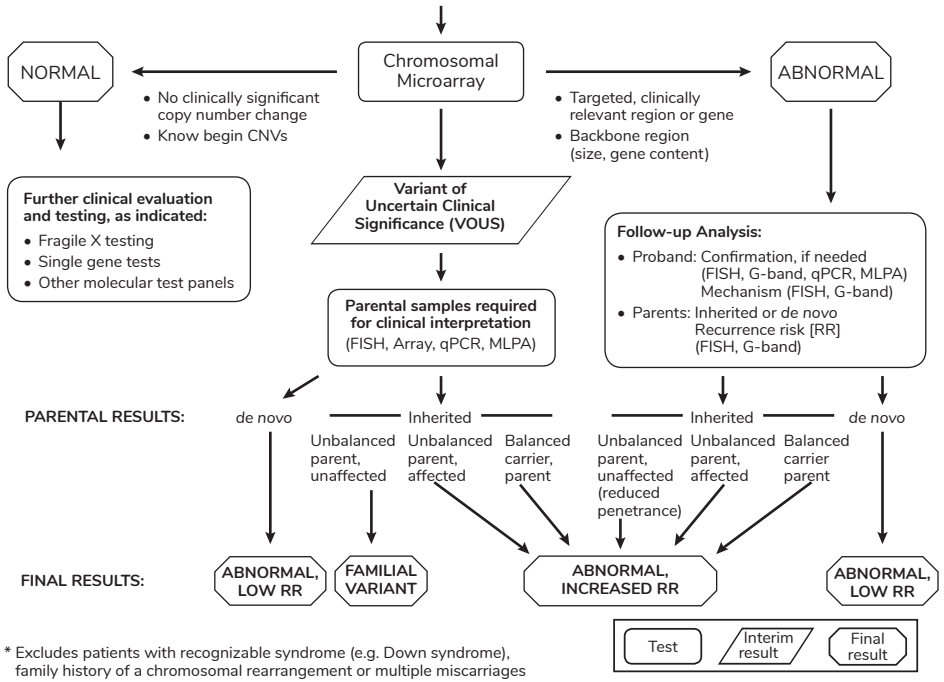
When does one use a CMA?

CMAs can be used for a wide variety of purposes from Product of Conception (POC) analysis to the various neurological conditions

- They are the first-tier test* for individuals with :
  - Developmental disabilities
  - Autism Spectrum disorders
  - Multiple Congenital anomalies
  - Mental Retardation
- For individuals with seizures and other developmental problems for which a chromosomal basis is suspected.
- Detection of CNVs at single exon level ensuring diagnosis of single gene diseases too.

The International Standard Cytogenetic Array (ISCA) consortium has developed the following algorithm for the use of CMAs **

*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)

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.*

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

What are the different types of CMAs?

1. Xon array
   - *Provides very high-resolution DNA copy number analysis to detect gains and losses at xon level. Also detects loss of heterozygosity (LOH), regions identical-by-descent, and uniparental isodisomy (UPD) on a single array*
   - *Extensive coverage for 453,076 xons is 17974 genes with increased sensitivity and specificity in 7000 clinically relevant genes*
2. 750K array
   - *Offers comprehensive gene-level coverage for known constitutional genes on a single array*
   - *Ideas for investigation of neurological disorders*
3. Optima array
   - *Ideal for genetic analysis of POC*
   - *Aneuploidy analysis*

Type	Number of CNV probes used	Number of SNPs Covered	Recommended for
Xon array	*6.55 Million*	*3,00,000*	*Exon level resolution for CNV detection*
750K	*750436*	*200436*	*Prenatal testing; First line screening for cases of Developmental Delay (DD), Mental Retardation (MR), Multiple Congenital Anoma lies (MCA) and Autism Spectrum Disorders (SAD)(ASD)*
Optima	*18018*	*148450*	*POC Testing/Aneuploidy*

Why choose MedGenome's CMA?

As the leader in genomic diagnostics in India, MedGenome offers cutting edge scientific offering coupled with world-class result interpretation and genetic counselling for CMA testing.

At MedGenome we have the following CMA offerings:

Type	Xon array	750K	Optima
Number of CNV probes used	6.55 Million	750436	18018
Number of SNPs covered	3,00,000	200436	148450
Probe density	>500Kb	>100Kb	>200Kb
Deletion	>25Kb	>100kb	> 1 Mb
Duplication	>200Kb	>400kb	> 2 Mb
Loss of Heterozygosity	>100Kb	>5 Mb	> 5 Mb
Absence of Heterozygosity	> 3% of total autosomal (>3Mb LCSH)	> 3% of total autosomal (>3Mb LCSH)	Not Reported
Long continuous stretches of Heterozygosity	LCSH >8-15 Mb	LCSH >8-15 Mb	Not Reported
Recommended for	First line screening for cases of Developmental Delay (DD), Mental Retardation (MR), Multiple Congenital Anomalies (MCA) and Autism Spectrum Disorders (ASD)		POC Testing
TAT	14 Working Days	12 Working Days	12 Working Days

Test sample requirements

Blood (3-5 ml in EDTA tubes)

Extracted DNA samples (1µg high quality DNA)

Product of Conception (POC) or (minimum 200mg of POC)

Required forms:

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

Turn around time (TAT):

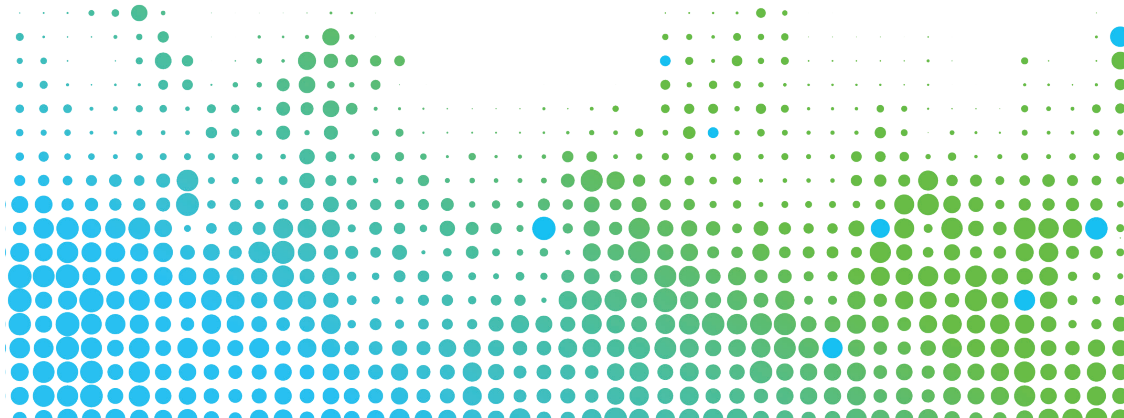
- 14 working days

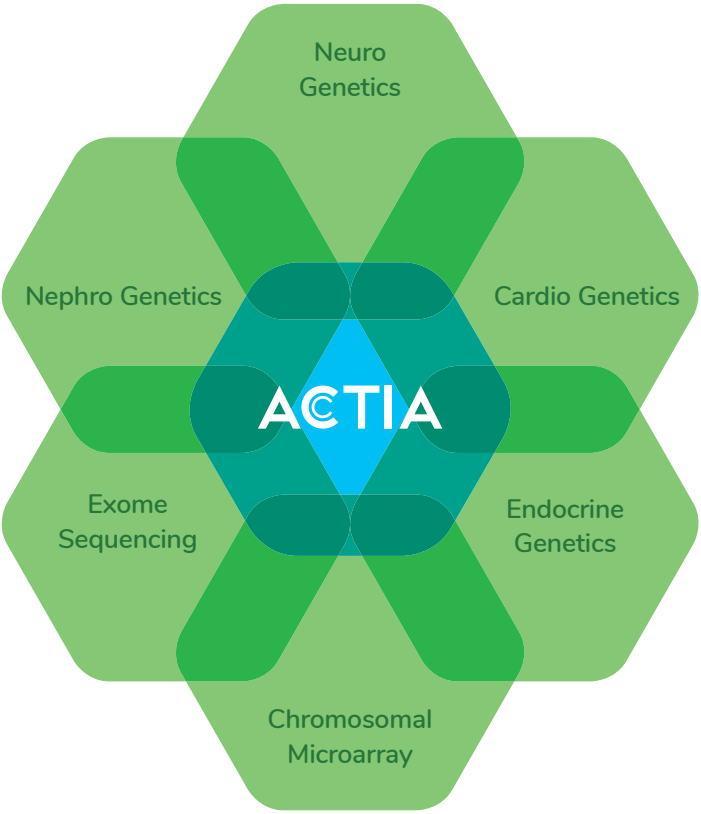
Free Genetic Counselling

ACTIA offers all your patients FREE pre & post-test genetic counselling with our expert and certified genetic counsellors.

Best available support for your patients and families via:

- Latest technologies
- Helpful customer service
- Clear result interpretation
- Counselling sessions with our Genetic Counsellors





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