آغا خان یونیورسٹی ہسپتال
The Aga Khan University Hospital

THE AGA KHAN UNIVERSITY HOSPITAL CLINICAL LABORATORIES

UPDATE
Chromosomal Microarray for Constitutional genetics

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INTRODUCTION

Chromosomal microarray (CMA) is a high resolution genetic test to investigate chromosomal aberrations such as gains and losses (copy number variants, CNV) that could contribute to genetic defects. CMA can identify structural abnormalities in chromosomes at a much greater resolution than conventional karyotyping (G-banded karyotype) and allows identification of clinically significant copy number aberrations amongst patients with multiple constitutional disorders. CMA can identify genomic microduplications or microdeletions that are a few hundred basepair (bp) in size. High resolution CMA platforms that incorporate single-nucleotide polymorphism (SNP) genotyping enable even greater diagnostic yields. However, CMA cannot be used to determine balanced translocations.

Genomic microarrays are the first line screening test recommended by the American College of Medical Genetics for a number of conditions; 1) identification of congenital genetic defects, 2) characterization of acquired genetic changes 3) determination of genomic variations and polymorphisms. Therefore, CMA should be considered for clinical presentation of:

- Isolated autism spectrum disorder (ASD) or ASD plus other findings
- Isolated global developmental delay or intellectual disability
- Multiple congenital anomalies in the absence of a syndrome diagnosis
- Unusual physical features (dysmorphisms)

PRINCIPLE

The Applied Biosystems CytoScan 750K enables the detection of high resolution copy number across the genome, providing allelic imbalance information from SNPs. This high-density array contains greater than 750,000 markers for copy number and approximately 200,000 genotype-able SNPs, which provide high-resolution copy number, and accurate breakpoint estimation. The SNPs on this array are from the public SNP database (dbSNP) and maximize genomic coverage, genotyping accuracy and optimize detection of homozygosity. There are 550,000 unique non-polymorphic probes to cover constitutional and cancer genes which are covered by each assay.

REPORTING

CMA will be used to identify deletions greater than 400 bp and or duplications greater than 500 bp in regions which contain at least one known gene.

SPECIMEN COLLECTION:
5 ml whole blood in EDTA tube is required

SCHEDULE:
The test is performed 1st Monday of every month and report will be issued after 15 days.

PLEASE FILE FOR QUICK REFERENCE