PEDIATRICS

Chromosomal Microarray Analysis

Array Comparative Genomic Hybridization

Baylor Genetics continues to provide high quality state of the art testing to the medical community through Chomosomal Microarray Analysis (CMA) products. We were one of the first to offer CMA for clinical applications and we remain a leader in the implementation of new technology for CMA. Baylor Genetics has several arrays designed to provide you with the most up to date technology and sophistication patients demand. Our arrays are custom designed to provide the highest detection rates possible. We have incorporated features such as exon coverage, whole genome coverage, SNP analysis for AOH/UPD, mitochondrial genome coverage, microRNAs, and other unique features that collectively yield the most comprehensive array analysis possible. In addition, Baylor Genetics custom designed arrays cover all subtelomere regions as well as pericentromic regions.

CMA-HR

- High resolution copy number analysis
- Custom BCM design-180K Agilent

Benefits

- Maximum sensitivity for detection of gains and losses
- 700 microRNAs
- Exon by exon coverage of over 1700 genes
- Tiling coverage of Mitochondrial Genome
- Whole genome coverage at a 30 kb resolution

Limitations: Does not detect absence of heterozygosity (AOH), uniparental disomy (UPD), or consanguinity.

CMA-SNP

- Excellent SNP coverage
- Affymetrix-CytoScan® HD

Benefits

- Detection of absence of heterozygosity (AOH) associated with uniparental disomy (UPD) or consanguinity
- Copy number coverage for classical deletion/duplication syndromes and detection of novel variants over 100 kb

Limitations: Does not detect exonic number changes.

CMA-HR + SNP SCREEN (COMPREHENSIVE)

- High resolution copy number analysis + SNPs (for detection of AOH & UPD)
- Custom BCM design - 400K Agilent

Benefits

- Maximum sensitivity for detection of gains and losses
- 700 microRNAs
- Exon by exon coverage of over 4,200 genes
- Tiling coverage of Mitochondrial Genome
- Whole genome coverage at a 30 kb resolution
- Detection of absence of heterozygosity (AOH) associated with uniparental disomy (UPD) or consanguinity
- 38 non-coding regulatory regions

Limitations: AOH less than 5Mb in size will not be reported. The detection rate of heterodisomies is currently not known for this assay.

With CMA-HR+SNP design, we can detect intragenic single exon deletions and duplications that would otherwise be missed as well as AOH events. This level of detail is ONLY available on the Baylor Genetics CMA-HR + SNP.

SUPERIOR COPY NUMBER DATA

- 280,000 oligos chosen for optimum data quality
- Exon by exon coverage for over 4,200 genes associated with MR, developmental delay, heart defects, etc.
- 38 non-coding regulatory regions

HIGH RESOLUTION WHOLE GENOME COVERAGE

- 30 kb resolution between known genes
- MicroRNAs
- Mitochondrial (for detection of deletions present in blood)

SNP COVERAGE FOR:

- 57,000 oligos used for SNP analysis
- Uniparental disomy (UPD)
- Detection of consanguinity
- Identification of regions of absence of heterozygosity (AOH)

For more information, or to request shipping kit, requisition forms, etc., please visit our website at www.bmgl.com.

FIGURE 1: CMA-HR + SNP SCREEN (COMPREHENSIVE) EXON 27 DELETION WITHIN CREBBP IN A PATIENT WITH RUBINSTEIN-TAYBI SYNDROME

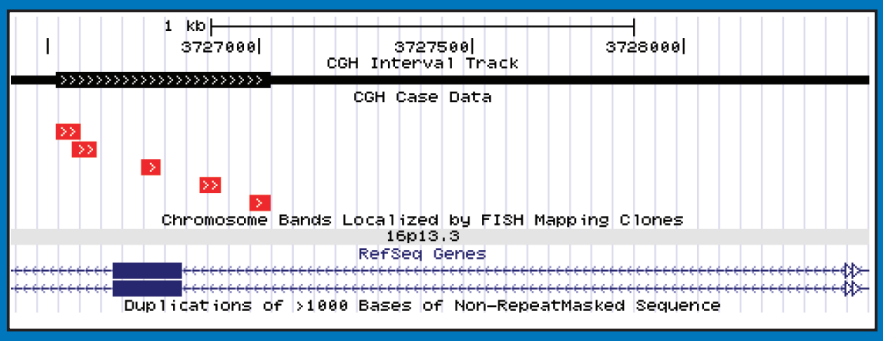


FIGURE 2: CHROMOSOMES 1-3 FOR A FIRST COUSIN OFFSPRING.

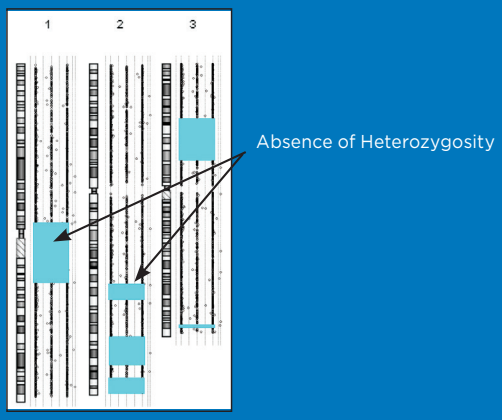
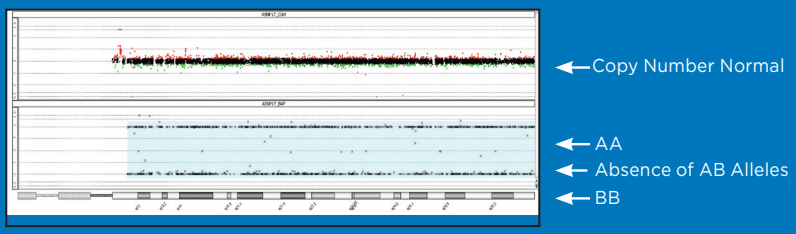


FIGURE 3: UPD ANALYSIS OF CHROMOSOME 15



For the first time ever, high resolution exon by exon copy number and SNP analysis is available from one array. The CMA-HR + SNP (Comprehensive) offers exon level deletion/duplication analysis for over 4,200 genes, and includes 57,000 probes used for SNP analysis. The SNP data will detect absence of heterozygosity (AOH) that is seen with consanguinity or uniparental disomy (UPD). Each patient will now benefit from both technologies, removing the obstacle of choosing one over the other. This combination of exon copy number and SNP data represents the most significant advance in array technology since the introduction of oligonucleotides.

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BAYLOR GENETICS
READY WITH ANSWERS