



Dayton Children's Cytogenetics Laboratory

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Test: Comparative Genome Hybridization by Microarray

aka: aCGH, array CGH, cytogenetic microarray, chromosome microarray, CMA, cytogenomic microarray

Test information:

Array Type: ISCA 105k oligo array v1.1, CytoChip™
 Average probe spacing: 35 kb overall (backbone), 5-8 kb in targeted regions

Analysis parameters:

Bckgr. corr.: Local
 Normalization: LOWESS, pin-wise, X&Y-adjusted, var. stab.
 Detection: CBS v2 (InfoQuant®)
 P-value: 0.001
 Min clones: 4
 Min Mbp: 0.005
 Threshold: 0.20

Coverage Summary: (Unique probes per chromosome; array contains ~105,000 targets)

Chr	Clones	Chr	Clones
1	7932	14	3169
2	8285	15	3092
3	6843	16	2781
4	6327	17	3044
5	6100	18	2584
6	5770	19	2058
7	5876	20	2145
8	4921	21	1283
9	3962	22	1501
10	4643	X	5492
11	4809	Y	636
12	4630	All	101201
13	3318	Autosome	95073

Description of aCGH analysis

Genes typically come in two copies. Microarray comparative genome hybridization (aCGH) looks for imbalances such as extra or missing copies of DNA. aCGH is conducted by combining equal amounts of patient DNA, labeled fluorescent green, with control DNA, known to have two copies of each genomic target, labeled fluorescent red. DNAs are hybridized simultaneously to the entire array over 100,000 (100k) individual oligonucleotide targets. Results are determined by comparing the ratio of red (control) and green (patient) DNA matching each individual target. Changes in the normal ratio reflect gains or losses of copies of DNA, or genomic imbalance in the patient that could explain a medical condition.

This test measures gains and losses of copies of DNA. It will detect genomic imbalances such as aneuploidy, chromosomal deletions, or chromosomal duplications of the specific loci located on the array. It will not detect balanced rearrangements such as inversions, reciprocal translocations, Robertsonian translocations or insertions. This array has an average spacing of about one probe per 35 kb across the genome (backbone) and one probe per 5-8 kb at loci where gains and losses may have clinical significance. This array will not detect epigenetic alterations such as imprinting defects, uniparental disomy, or loss of heterozygosity; these are rare causes of genomic disorders. It will not detect changes in copy number of loci not represented on the array. It will not reliably detect mosaicism. It will not reliably detect imbalances of less than 100 kb. It will not detect point mutations – changes of one or a few base pairs of DNA - within individual genes. A negative aCGH test result does not rule out a genetic diagnosis, and such diagnoses may require additional, targeted genetic testing.

Consultation with genetics experts at Dayton Children's Medical Center is available to assist in determining the suitability of aCGH testing in specific situations, or to assist in interpretation of test results. Additionally, clinical evaluation and genetic counseling are available to patients before or after genetic testing. Call 937-641-3800 for information.

This test platform was developed by the International Standard Cytogenomic Array (ISCA) Consortium. Analysis is performed with parameters reported above. The performance characteristics of this platform were established by the Cytogenetics Laboratory at Dayton Children's Medical Center. It has not been cleared by the US Food and Drug Administration (FDA). The FDA has determined that such approval or clearance is not necessary. This test is used for clinical purposes; it should not be regarded as investigational or for research. This laboratory is certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high-complexity testing. Pursuant to the requirements of CLIA, this laboratory has established and verified this test's accuracy and precision. Additional information is available upon request.