THE UNIVERSITY OF CHICAGO | Genetic Services Laboratories

Postnatal Cytogenomic SNP Array Information Sheet

Cytogenomic SNP array is offered as a postnatal first-tier test appropriate for individuals with multiple anomalies that are not specific to well-delineated genetic syndromes, apparently nonsyndromic developmental delay/intellectual disability or autism spectrum disorders as recommended by the American College of Medical Genetics [1]. Copy number variations identified by cytogenomic SNP array are also identified in 5-30% of individuals with epilepsy [2]. Cytogenomic SNP array is also appropriate for: follow-up testing for individuals with the phenotypes listed above with a previously normal chromosome analysis result; clarification of size, precise breakpoints or gene content of abnormalities detected by routine chromosome analysis or FISH studies; to rule out cryptic copy number changes (imbalances) at the breakpoints of apparently balanced chromosome rearrangements and for the identification of long contiguous stretches of homozygosity. This testing may also be used to determine sex chromosome composition in patients with ambiguous genitalia or disorders of sex development.

Test methods

Cytogenomic SNP array uses the Affymetrix CytoScan HD platform. This array includes 2.67 million markers for copy number (CN) analysis including approximately 750,000 SNP probes, and 1.9 million non-polymorphic probes for whole genome coverage. This array provides an average intergenic probe density of one probe per 880 bases and an overall (gene and non-gene backbone) probe density of one probe every 1,148 bases.

Testing analysis

Results are analyzed using the Affymetrix Chromosome Analysis Suite (ChAS) software. Analysis of data is based on the most recent human genome build and is stated in the final report. All reported base pair coordinates are estimated. Deletions larger than 200 kb and duplications larger than 400 kb will generally be reported. Smaller copy number changes may also be reported if they are considered clinically significant based on genomic information available at the time the report is issued. Benign copy number variants will not be reported but will be kept on file in the laboratory. Copy number changes resulting in carrier status for autosomal recessive disorders may not be reported unless concern for a specific disorder is communicated to the laboratory. Copy number abnormalities reported within the validated parameters will not be confirmed by a separate method. However, when appropriate, follow up testing will be recommended if additional clinically relevant information can be obtained. Long continuous stretches of homology (LCSH), which can result from uniparental disomy (UPD) or common descent, can also be detected using the CytoScan HD array. Reported regions of LCSH will not be confirmed but recommendations for further molecular testing may be made to confirm UPD or to identify recessive alleles possibly associated with the patient's condition.

Limitations

This test will not detect balanced chromosome rearrangements such as Robertsonian or other reciprocal translocations, inversions or balanced insertions. This test will not detect imbalances of regions not represented on the array. This test does not detect all types and instances of uniparental disomy. This test is not designed to detect mosaicism at low levels. Normal findings do not rule out the diagnosis of any disorder since some genetic abnormalities may be undetectable with this assay. Specifically, this test does not detect point mutations, small deletions or insertions below the resolution of this assay, or other types of mutations such as epigenetic changes.

Cytogenomic SNP array (postnatal)

Sample specifications:	2-5cc of blood in a purple top EDTA tube, and 2-5cc in a green top sodium heparin tube
Cost:	$1500
CPT codes:	81229
Turn-around time:	4 weeks

Results

Results, along with an interpretive report, will be faxed to the referring physician. Additional reports will be provided as requested. All abnormal results will be reported by telephone. The results of this test may be of unclear clinical

significance. In such cases, additional family studies may be necessary to interpret the results. CNVs detected by this platform may not be investigated or reported if they are devoid of relevant gene content or reported as common findings in the general population based on available database searches or are gains smaller than 400 Kb or losses smaller than 200 Kb. A referral to a clinical geneticist or counselor is appropriate for individuals and families to discuss results of this test.

For more information about our cytogenomic SNP array, please contact our cytogenetic pathologist:
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For more information about our testing options, please visit our website at dnatesting.uchicago.edu or contact us at 773-834-0555.

References

1. Manning, M., L. Hudgins, and P.P.a.G. Committee, *Array-based technology and recommendations for utilization in medical genetics practice for detection of chromosomal abnormalities.* Genet Med, 2010. **12**(11): p. 742-5.
2. Spreiz, A., et al., *Chromosomal microaberrations in patients with epilepsy, intellectual disability, and congenital anomalies.* Clin Genet, 2014. **86**(4): p. 361-6.

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