

- 1) South ST, Lee C, Lamb AN, Higgins AW, Kearney HM, Working Group for the American College of Medical Genetics and Genomics Laboratory Quality Assurance Committee. ACMG standards and guidelines for constitutional cytogenomic microarray analysis, including postnatal and prenatal applications: revision 2013. *Genet Med.* 2013; 15(11):901-9.
- 2) Shao L, Akkari Y, Cooley LD, et al. Chromosomal microarray analysis, including constitutional and neoplastic disease applications, 2021 revision: a technical standard of the American College of Medical Genetics and Genomics (ACMG). *Genet Med.* 2021;23(10):1818-1829.
- 3) Rehder CW, David KL, Hirsch B, Toriello HV, et al. American College of Medical Genetics and Genomics: standards and guidelines for documenting suspected consanguinity as an incidental finding of genomic testing. *Genet Med.* 2013; 15(2):150-2.
- 4) Clinical and Laboratory Standards Institute. *Genomic Copy Number Microarrays for Constitutional Genetic and Oncology Applications*. 1st ed. CLSI guideline MM21-Ed1. Clinical and Laboratory Standards Institute, Wayne, PA; 2015.

CYG.49600 DNA Copy Number Array Reports Elements

Phase I

In addition to all relevant items outlined in CYG.31875, reports for DNA copy number analysis using arrays include the following elements:

1. Platform used
2. Genome build used
3. Analysis and/or reporting strategy: Detection and/or reporting criteria for copy number and homozygosity (if applicable), including number of probes and/or size limitations
4. ISCN-compliant nomenclature for reported findings
5. References to any databases used
6. A statement on the need for genetic counseling when indicated
7. A statement recommending further testing when indicated
8. Clinical significance of DNA copy number changes, when applicable

NOTE: Platform information includes, but is not limited to manufacturer, array version, number of probes, average probe spacing, SNP content, and targeted regions. Analysis strategy includes the copy number controls methodology: comparative/competitive or in silico.

Complex findings should be reported using ISCN-compliant nomenclature (eg, table format, "cth" for chromothripsis), when possible.

REFERENCES

- 1) South ST, Lee C, Lamb AN, Higgins AW, Kearney HM, Working Group for the American College of Medical Genetics and Genomics Laboratory Quality Assurance Committee. ACMG standards and guidelines for constitutional cytogenomic microarray analysis, including postnatal and prenatal applications: revision 2013. *Genet Med.* 2013; 15(11):901-9.
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- 3) Vermeesch JR, Fiegler H, de Leeuw N, Szuhai K, Schoumans J, Ciccone R, Speleman F, Rauch A, Clayton-Smith J, Van Ravenswaaij C, Sanlaville D, Patsalis PC, Firth H, Devriendt K, Zuffardi O. Guidelines for molecular karyotyping in constitutional genetic diagnosis. *Eur J Hum Genet.* 2007 Nov;15(11):1105-14
- 4) McGowan-Jordan J, Hastings R, Moore S, eds; International Standing Committee on Human Cytogenomic Nomenclature. *ISCN: An International System for Human Cytogenomic Nomenclature (2020)*. Basel, New York: Karger; 2020.
- 5) Department of Health and Human Services, Centers for Medicare and Medicaid Services, Clinical laboratory improvement amendments of 1988; final rule. *Fed Register.* 2003(Jan 24):1043-1044 [42CFR493.1276], 1047-1048 [42CFR493.1291]
- 6) Clinical and Laboratory Standards Institute. *Genomic Copy Number Microarrays for Constitutional Genetic and Oncology Applications*. 1st ed. CLSI guideline MM21-Ed1. Clinical and Laboratory Standards Institute, Wayne, PA; 2015.

PERSONNEL

NOTE: For purposes of CAP accreditation, the "laboratory director" is that individual who oversees all sections of the laboratory, and in whose name accreditation is granted. Specific requirements for that person are found in the Director Assessment Checklist. The section director (technical supervisor) refers to the person responsible for the medical, technical and/or scientific oversight of the cytogenetics laboratory section.