Chromosomal microarray analysis

For pediatric disorders

When to consider testing

Chromosomal microarray (CMA) analysis examines a patient's genome for gains (duplications) and losses (deletions) of genetic material that can cause syndromic and non-syndromic conditions.

The American College of Medical Genetics,[1] the American Academy of Pediatrics,[2] and the American Academy of Neurology[3] recommend using CMA as a **first-tier diagnostic test** for the investigation of:

- developmental delay or intellectual disability (DD/ID)
- autism spectrum disorders (ASD)
- multiple congenital anomalies (MCA)

Chromosomal microarray analysis offers a high diagnostic yield and may prevent unnecessary, potentially invasive testing. A clinically significant chromosomal abnormality is detected in **approximately 15–20% of individuals** with unexplained DD/ID, ASD, or MCA.[4]

Why Invitae

Reliable results, fast

Invitae technology:

- provides genome-wide coverage of copy number gains and losses at a much higher resolution than conventional karyotyping
- evaluates >1.7 million SNP markers with a mean probe spacing of 1.8 kilobases
- reports copy number changes involving ≥16 probes and regions of homozygosity ≥5 megabases
- delivers results in 10 to 12 days, on average

Invitae offers a broad menu of additional testing including panels and exome sequencing for patients who remain undiagnosed after a negative CMA result.

Deep expertise

- Invitae has analyzed results from over 65,000 microarrays.
- Invitae's scientists, laboratory directors, and genetic counselors have decades of experience in interpreting microarray data and are available to assist you in understanding complex results.

65,000+
microarrays

Convenient buccal swab specimen option

When it is not possible to collect blood, you can opt to collect DNA from the inside of your patient's cheek for microarray analysis.

Affordable answers

- Invitae is in network for over 290 million individuals
- Patient-pay option: $450
- Medicaid accepted in many states

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How it works

Visit our Developmental Disorders (Cytogenetics) Online Test Catalog to learn more.

Order a test using the Invitae Online Portal, or download the paper order form.

Collect a specimen, either blood or buccal (cheek) swab.

Receive your chromosomal microarray analysis report in 10 to 12 days on average.

Transparent billing

At Invitae, we believe that high-quality genetic testing should be affordable and accessible. Our flexible billing options ensure that cost is not a barrier to accessing genetic information.

Insurance

Invitae is proud to be in network for over 290 million individuals and will work directly with your patient's insurance company to coordinate coverage and payment.

Patient pay

Patients have the option to pay $450 for Invitae chromosomal microarray analysis. This option requires upfront payment before test results are released.

Patient assistance program

Invitae is committed to making high-quality genetic testing affordable and accessible by removing financial and logistical barriers. Our patient assistance program is available to patients in the US who undergo testing with Invitae and meet income criteria. Please contact Client Services at 800-436-3037 to learn more about our interest-free payment plans and financial assistance program.

Hands-on support

For clinicians

Call Clinical Consult at 800-436-3037 to review patient cases, learn about the differences between Invitae's testing options, and discuss results. Contact Client Services at the same number to get help with ordering and billing.

For patients

Patients can contact Invitae Client Services with billing and insurance-related questions. For questions about genetic testing and test results, patients can talk to one of Invitae's genetic counselors. Both are available by calling 800-436-3037, Monday through Friday, 5:00 am to 5:00 pm Pacific.

For more information visit: www.invitae.com/clinical-support-services.

References:
1. Manning M, Hudgins L. Array-based technology and recommendations for utilization in medical genetics practice for detection of chromosomal abnormalities. *Genet Med.* 2010;12(11):742-5.
2. Moeschler JB, Shevell M, Committee on Genetics, 2014. Comprehensive evaluation of the child with intellectual disability or global developmental delays. *Pediatrics* 134, e903-18.
3. Michelson DJ, *et al.* Evidence report: Genetic and metabolic testing on children with global developmental delay: report of the Quality Standards Subcommittee of the American Academy of Neurology and the Practice Committee of the Child Neurology Society. *Neurology.* 2011;77(17):1629-35.
4. Miller DT, *et al.* Consensus Statement: Chromosomal microarray is a first-tier clinical diagnostic test for individuals with developmental disabilities or congenital anomalies. *Am J Hum Genet.* 2010;86(5):749-64.