CENTOGENE
THE RARE DISEASE COMPANY

Genome-Wide CNV Analysis

CentoArray®

For Early and Accurate Answers

CentoArray®
Chromosomal Microarray Analysis

Chromosomal Microarray Analysis (CMA) is a high-resolution genomic test that identifies chromosomal abnormalities such as deletions or duplications (CNVs) associated with a wide range of genetic conditions. Utilizing high-resolution CMA with Single Nucleotide Polymorphism (SNP) probes enables the detection of Loss / Absence Heterozygosity (LOH / AOH), which can indicate an elevated risk for autosomal recessive diseases or imprinting disorders resulting from Uniparental Disomy (UPD) or consanguinity.

CMA has been a key tool for diagnosing many genomic disorders and is recommended whenever a cytogenomic abnormality is suspected as an underlying cause of a patient's condition / disorder. Clinical guidelines advise using CMA in prenatal testing for pregnancies exhibiting structural abnormalities on ultrasound,[1,2] testing fetal tissue following pregnancy loss,[1,3,4] and conducting postnatal diagnostic testing for individuals displaying unexplained developmental delay, intellectual disability, autism spectrum disorder, and / or multiple congenital malformations.[5–7]

CENTOGENE's CMA solution, CentoArray®, is a powerful tool for fast and reliable cytogenomic diagnostics – offering enhanced coverage of disease-causing genes, detailed high-resolution copy number analysis, and UPD detection across various sample types – ensuring accurate medical interpretation at every stage of patient care, including fetal, pediatric, and adult diagnostics.

The CENTOGENE Advantage

Broad Coverage and High Resolution
Optimized probe spacing and resolution ensure the best detection of disease-related copy-number changes

Disease-Focused Content
Exon-by-exon coverage of over 4,800 clinically significant genes provides unparalleled disease-specific insight

Fast & Excellence in Medical Reporting
One test, rapid results, and excellence in every medical report, powered by our extensive experience in rare and neurodegenerative disease diagnostics

Key Features and Performance

CentoArray is a reliable and fast diagnostic tool for detecting chromosomal abnormalities, such as CNVs down to the exon level, large chromosomal imbalances, UPD, consanguinity, and mosaicism across diverse sample types. It is based on Illumina Infinium™ Global Diversity Array with Cytogenetics-8, clinically validated at CENTOGENE for use with various sample types to adapt to our customers' needs.[8,9] With over 1.8 million genomic markers, CentoArray is designed with dense probe coverage in regions of known genetic function, ensuring enhanced detection capabilities for pathogenic variants.[9]

Broad Coverage & High Resolution		• Illumina Infinium™ Global Diversity Array with Cytogenetics-8 • ~1.8 million markers genome-wide markers distributed • 160 K carefully selected SNP (single-nucleotide polymorphism) probes to enhance cytogenetic performance
Disease-Focused Content		• Exon-level resolution of relevant disease-associated genes • Coverage of >4,800 key genes across the genome
Standard Reporting Thresholds	**CNVs**	• >50 kb for copy number loss • >200 kb for copy number gain
	Mosaicism	≥30 %
	AOH/LOH	Regions >10 Mb
Flexible Specimen Types		CentoCard®, EDTA-blood, ready-to-use DNA, buccal swab, saliva, amniotic fluid, chorionic villi, cord blood, cultured cells, and tissue sample and biopsy material
Rapid Results		TAT: ≤15 business days

When to Recommend CentoArray

CentoArray offers comprehensive cytogenetic testing and is recommended for individuals suspected of chromosomal deletions or duplications. These abnormalities can explain conditions such as developmental delay, intellectual disability, or multiple congenital anomalies.[1–7] CENTOGENE provides CentoArray testing solutions for clinical diagnostics across all stages of life.

Postnatal Cases

CentoArray is the solution for postnatal diagnosis, often utilized in the pediatric setting, and is recommended for[5–7]:

- Patients with a history of multiple congenital anomalies, autism spectrum disorder, neurodevelopmental disorders, developmental delay, intellectual disability, failure to thrive, dysmorphic features, and/or epilepsy syndromes
- Diagnosis of UPD and regions exhibiting LOH/AOH
- Patients with unsuccessful prior genetic testing such as karyotype or Fluorescence In Situ Hybridization (FISH) studies

Prenatal Cases

CentoArray Prenatal is tailored for prenatal diagnosis (ongoing pregnancies) to assess the genetic health of a developing fetus. It is often recommended when there is concern about potential chromosomal abnormalities or genetic disorders in the unborn child[1,2], including:

- Prenatal ultrasound with abnormal findings
- Positive serum screen or noninvasive prenatal screening (NIPT)
- Advanced maternal age or higher a priori reproductive risk for a fetal chromosome abnormality
- A previous child, family history, or pregnancy loss with a confirmed chromosome abnormality

Products of Conception (POC)/Pregnancy Loss Cases

CentoArray POC is the solution for diagnostic testing of products of conception and is recommended for understanding the cause of pregnancy loss[1,3,4], to:

- Identify the underlying etiology of intrauterine fetal death or stillbirth
- Confirm a suspected clinical diagnosis

Best-in-Class Medical Reporting

Leveraging the Illumina Infinium Global Diversity Array and analyzed using Biomano Genomics NxClinical Software integrated into our diagnostic workflow, CentoArray provides the optimal combination of turnaround time, genomic coverage, and data quality across various specimen types.[8–10]

Our comprehensive medical reports are meticulously prepared by our interdisciplinary team of experts. Internal confirmatory testing, conducted when necessary to support a more accurate diagnosis, combined with our best-in-class medical interpretation can have a significant, positive impact on patient care.

Our medical reports are comprehensive, conclusive, and transparent. Key features of our medical reports include:

- Clear, structured, conclusive, and concise formats
- Integration of the patient's family history and clinical information
- Clear presentation of identified variations following international best-practice guidelines
- The classification of large CNVs is based on the American College of Medical Genetics and Genomics (ACMG) and ClinGen (Clinical Genome Resource) standards and guidelines for interpretation and reporting of constitutional CNVs[11]
- Recommendations for further differential diagnoses and testing or follow-up analyses for specific diseases
- References to publications supporting the medical and scientific results and interpretation, along with detailed method descriptions

For more infromation about our medical reporting, please visit centogene.com/reporting

References

1 American College of Obstetricians and Gynecologists (ACOG) 2016, PMID: 26938573
2 Society for Maternal-Fetal Medicine (SMFM) 2016, PMID: 27427470
3 Sahoo et al. 2016, PMID: 27337029
4 Rosenfeld et. 2015, PMID: 25846569
5 Manning et al. 2010, PMID: 20962661
6 Miller et al. 2010, PMID: 20466091
7 Shen et al. 2010, PMID: 20231187
8 Data on file at CENTOGENE
9 Illumina Infinium Global Diversity Array with Cytogenetics-8, https://emea.illumina.com/products/by-type/clinical-research-products/infinium-global-diversity-array-cytogenetics-8.html [15/04/2024]
10 Biomano Genomics NxClinical software, https://bionano.com/nxclinical-software [15/04/2024]
11 Riggs et al. 2020, PMID: 31690835

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