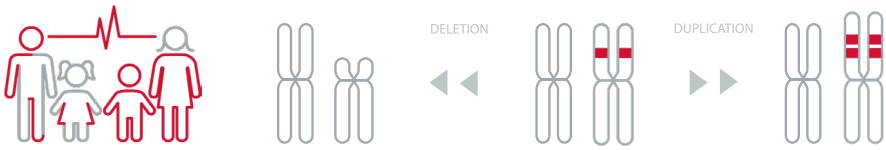
CENTOGENE
THE RARE DISEASE COMPANY

CentoLCV



CentoLCV – Our versatile platform for detecting chromosomal imbalances

As part of our commitment to helping end the diagnostic odyssey of rare disease patients, CENTOGENE has developed CentoLCV – a comprehensive Copy Number Variation (CNV) analysis using genome-wide Next Generation Sequencing (NGS). CNVs, which represent a large component of structural variations in the human genome, consist of gains or losses of genomic regions ranging from a few thousand to several million DNA bases pairs in size.[1-2] CNVs are involved with a wide variety of genetic disorders and have a significant impact on human health and disease.[2-4] NGS, and more specifically WGS, are powerful tools to detect CNVs.[6,7]

Our new CNV detection platform has a higher genome coverage and resolution than conventional tests using karyotyping or microarrays[5-7] and can detect large CNVs, such as full and partial chromosomal aneuploidies, microdeletions/microduplications, and partial or complete single-gene related CNVs of clinical relevance. CentoLCV ultimately allows for a more accurate molecular diagnosis, leading to better, more informed outcomes, and potentially also reduced reproductive risks.

Who should consider CentoLCV?

Geneticists, neonatologists, pediatricians, and neurologists providing diagnoses and treatments for patients matching any of the following criteria:

- Suspected chromosomal imbalances (e.g., Down syndrome and Turner syndrome), including microdeletion/microduplication syndromes (e.g., DiGeorge syndrome and Williams syndrome)
- Multiple congenital anomalies, including global developmental delay (e.g., Phelan-McDermid syndrome), intellectual disability (e.g., 17q21.31 microdeletion in learning disability), and many more
- Autism or autism spectrum disorders (e.g., 16p11.2 microdeletion in autism)

Key features of CentoLCV

CentoLCV is based on high throughput genome sequencing technology and covers the complete genome at the sequence level. It confidently detects CNVs with high sensitivity and resolution, providing a fast and precise diagnostic test.[5]

CATEGORY	FEATURES
COVERAGE	• Full genome coverage (>20,000 genes) with a mean depth ≥3x • Coding (exonic) and non-coding regions (intronic, regulatory regions and splice sites)
VARIANTS	• Whole and partial chromosomal aneuploidies • Unbalanced translocations • Microdeletions and microduplications • Deletions and duplications within single genes
DETECTION RANGE AND SENSITIVITY	• ≥50 kb, and lower for homo/hemizygous deletions • 4x higher sensitivity than microarray technologies
OPTIONS	• Order individually or as add-on to CentoXome®
TAT	• ≤15 business days
MATERIAL	• 1 CentoCard®*

*Please check our web page for further options (https://www.centogene.com/diagnostics/how-to-order.html)

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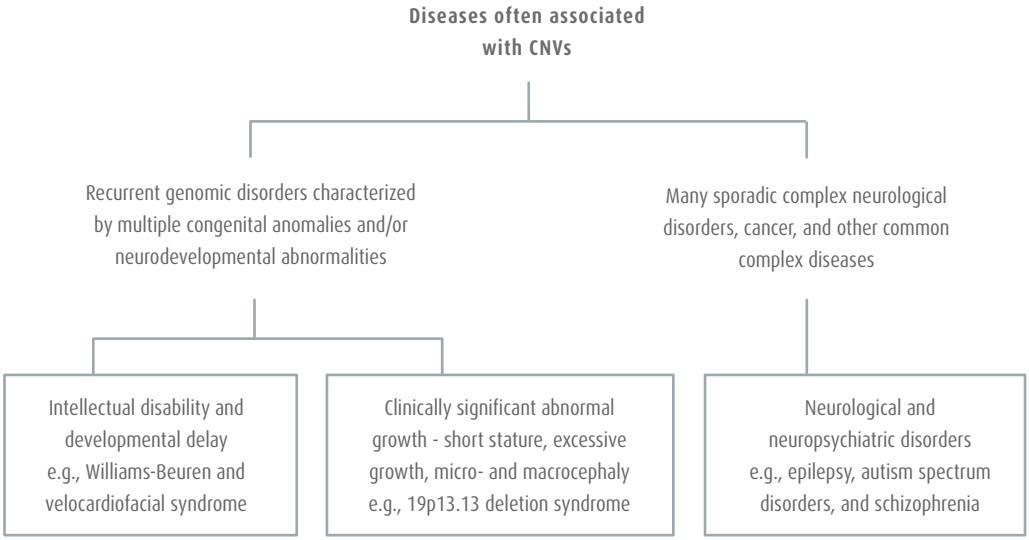
What disorders are targeted by CentoLCV?

CNVs are involved in a wide variety of disorders, ranging from pediatric disorders and congenital birth defects to adult-onset neuropsychiatric and neurodegenerative disorders.[2-4]



Why is CentoLCV a superior alternative to conventional karyotyping and microarrays?

- It provides robust detection of CNV changes throughout the entire genome with higher resolution and precision than conventional karyotyping and microarrays[5-7]
- Many hereditary disorders are also caused by novel aberrations, which may be difficult to detect by conventional karyotyping and microarrays[5-7]
- It exceeds the diagnostic yield of conventional karyotyping and microarrays[5-8]

Features	CentoLCV	Microarrays*	Karyotyping
RESOLUTION RANGE[5-7]	Exon/gene- to chromosome level	Sub- to chromosome level	Chromosome level
COVERAGE AND TARGET RANGE[5-7]	Unbiased across whole genome	Biased by probe spacing and density across the genome	Narrowed to large chromosome changes across the genome
DIAGNOSTIC YIELD[5-8]	>15%	10-15%	3-7%

*Details depends on the platform used

REFERENCES

[1] Freeman *et al.* 2006, PMID: 16809666
[2] Zhang *et al.* 2009, PMID: 19715442
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[4] Lew *et al.* 2018, PMID: 30258274
[5] CENTOGENE data on file
[6] Zhou *et al.* 2018, PMID: 30061371
[7] Dong *et al.* 2016, PMID: 26820068
[8] Miller *et al.* 2010, PMID: 20466091

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