CeGaT

Genetic Diagnostics

Revolution in Exome Diagnostics

CeGaT's ExomeXtra® with genome-wide CNV analysis

Exome Diagnostics

Introducing the New ExomeXtra®: One Single Test for Reliable Clinical Diagnoses

With the latest update of our ExomeXtra®, we offer you the most innovative solution in exome diagnostics. The integration of a **unique CNV backbone** now allows the genome-wide detection of copy-number variations (CNVs) within the scope of exome sequencing. Hence, with just one test, you get the combined benefits of whole genome sequencing and whole exome sequencing and the full diagnostic power of array CGH (Comparative Genomic Hybridization).

ExomeXtra® provides reliable clinical results to support you in accurately stating a diagnosis for your patients.

The new version of ExomeXtra® (version 5.0) includes:

- all protein-coding regions of the genome
- clinically relevant RNA genes
- > 38,000 intergenic and intronic positions associated with genetic disease according to ClinVar, HGMD, and internal databases
- high and uniform coverage of the entire mitochondrial genome to reliably detect different degrees of heteroplasmy
- pharmacogenetically relevant variants in selected genes
- the new backbone for genome-wide detection of CNVs

Benefit from Our Unique CNV Backbone

The new CNV backbone allows CNV calling in coding regions and CNV detection in non-coding regions. The latter is unique in exome diagnostics, providing more uniform coverage and increasing the probability of finding a disease-causing mutation.

Compared to the standard CNV determination approach, additional probes cover the non-coding regions in our genome-wide CNV calling. The CNV detection outside the coding exons has an array-like resolution of 50 kb.

Find Out More about Our
Exome Diagnostics Services
www.cegat.com/exome-diagnostics

About Us

CeGaT is a global provider of genetic analyses for a wide range of medical, research, and pharmaceutical applications.

Founded in 2009 in Tübingen, Germany, the company combines state-of-the-art sequencing technology with medical expertise – with the aim of identifying the genetic causes of diseases and supporting patient care. For researchers and pharmaceutical companies, CeGaT offers a broad portfolio of sequencing services and tumor analyses. CeGaT generates the data basis for clinical studies and medical innovations and drives science forward with its own insights.

The owner-managed company stands for independence, comprehensive personal customer service, and outstanding quality. CeGaT's laboratory is accredited according to CAP/CLIA, DIN EN ISO 15189, and DIN EN ISO/IEC 17025 and thus meets the highest international standards. To obtain first-class results, all processes are carried out in-house under scientific supervision.

DAkkS
Deutsche Akkreditierungsstelle
D-ML-13206-01-00

Accredited by DAkkS according to DIN EN ISO/IEC 15189:2014

CAP ACCREDITED
COLLEGE of AMERICAN PATHOLOGISTS

CLIA CERTIFIED ID: 99D2130225

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2023/11