Genetic Diagnostics

Exome Diagnostics

CeGaT

Finding the Genetic Cause of Rare Diseases

Exome diagnostics is the genetic testing approach of choice for patients with complex, heterogeneous, and unspecific symptoms. It supports physicians in stating a diagnosis, often after their patients have experienced years of uncertainty.

The exome comprises all protein-coding regions (exons) of the approximately 23,000 genes in the human genome. Although the exome accounts for only about 1%-2% of the whole genome, about 89% of all known disease-causing variants are located within the exons. Whole exome sequencing allows the simultaneous analysis of genes in any combination. If the exomes of additional family members are also sequenced, as in trio exome diagnostics, an exome-wide segregation analysis can be performed. This approach significantly increases the chances of finding the genetic cause of complex phenotypes in a shorter time compared to genetic testing of small gene sets.

We are committed to identifying the genetic cause of disease for every patient. Therefore, we have developed an innovative diagnostic approach that goes beyond the possibilities of regular exome diagnostics: ExomeFocus® and ExomeXtra® (Single & Trio) offer an efficient solution for every patient scenario and family constellation.

Benefit From our Unique Analysis Approach

Extra Smart Clinical Design

CeGaT's exome design considers all known disease-causing regions including more than 38,000 intronic and intergenic variants described as disease-relevant in the Human Gene Mutation Database (HGMD) and ClinVar, the largest public database of genotype-phenotype relationships. CeGaT's ExomeXtra® provides the ideal basis for genetic diagnostics and can be understood as a clinical genome providing the most comprehensive sequencing data, which 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
- backbone for genome-wide detection of copy number variants (CNVs)

Watch Our Latest Webinar in the Field of Exome Diagnostics.

Scan the QR code and learn how we can help you solve complex patient cases.

www.cegat.com/webinars

Extra Thorough Analysis

CeGaT's data analysis goes beyond normal exome diagnostics and increases solution rates. We address copy number variants (CNVs), including compound heterozygous combinations of sequence variants (SNVs, indels) with CNVs. Sequencing data is routinely screened for repeat expansions related to reported phenotypes. For Trio ExomeXtra®, we consider variants in genes with reduced penetrance, variable expressivity, or imprinting effects.

Extra Insightful Results

CeGaT combines human know-how with bioinformatic analysis. The in-house developed software generates data that are evaluated by multiple scientific experts – creating the best possible medical report:

- written by PhDs
- reviewed by medical doctors and geneticists
- based on the most recent literature
- VUS re-evaluation (Single ExomeXtra®; Trio ExomeXtra®)

Our Diagnostic Services

Single Exome:

- **ExomeXtra®**

  The optimal whole exome diagnostics test for an individual patient. Utilizing our proprietary exome enrichment Single ExomeXtra® ensures comprehensive coverage of all known disease-causing genome regions. Tailored to the patient's phenotype, a medical report with a discussion of variants is generated.

- **ExomeFocus®**

  The most efficient approach for singleton exome diagnostics. Our in-house developed software uses our extensive in-house database of genetic variants as well as all publicly available databases to data-mine the patient's exome for high-impact variants. Our scientific team then evaluates these variants to ensure clinical relevance for the patient's phenotype.

Trio ExomeXtra®:

Used for diagnosing affected patients with unaffected parents. The analysis includes both parents, significantly improving the chances of a successful diagnosis. Beyond IVERP*, the test encompasses detection of uniparental disomies (UPD), covering both isodisomies and heterodisomies affecting whole or partial chromosomes.

Prenatal ExomeXtra®:

- **Diagnostics – with conspicuous ultrasound findings**
  Exome analysis to determine the genetic cause of a disease in a fetus with abnormal ultrasound findings.

- **Diagnostics – without ultrasound findings**
  Trio Exome analysis and screening of over 2,000 genes to clarify the risk of severe, early-onset diseases.

* Accounting for variants in genes with known imprinting, variable expressivity and reduced penetrance.

Compare Our Diagnostic Services

Single ExomeFocus®	Single ExomeXtra®	Trio ExomeXtra®	Prenatal ExomeXtra®
CeGaT's own exome enrichment: all known disease-causing regions			
✓	✓	✓	✓
In-house developed, enhanced bioinformatics, data analysis, variant calling, and variant pathogenicity scoring			
✓	✓	✓	✓
Reported variants of the ACMG classes			
3/4/5	3/4/5	3/4/5	4/5
Variant classification with details on ACMG criteria and scale			
✓	✓	✓	✓
IVERP variants*			
✗	✗	✓	✓**
VUS re-evaluation			
✗	✓	✓	✗
Costs			
€	€ €	€ € €	€ € €
Solving rate			
✗✗✗	✗✗✗✗	✗✗✗✗✗	✗✗✗✗✗

* Accounting for variants in genes with known imprinting, variable expressivity and reduced penetrance.
** Prenatal diagnostics with conspicuous ultrasound findings.

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 and medical supervision.

CAP ACCREDITED
COLLEGE of AMERICAN PATHOLOGISTS

CLIA CERTIFIED ID: 99D2130225

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Deutsche Akkreditierungsstelle
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Accredited by DAkkS according to DIN EN ISO 15189:2014

CeGaT GmbH
Paul-Ehrlich-Str. 23
72076 Tübingen
Germany

Phone: +49 7071 565544-55
Fax: +49 7071 565544-56
Email: diagnostic-support@cegat.com
Web: www.cegat.com

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