Genetic Diagnostics

Exome Diagnostics

CeGaT

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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.

Benefit From our Unique Analysis Approach

Extra smart clinical design

Superior clinical relevance – One NGS test covers everything

CeGaT's exome diagnostics considers all known disease-causing regions including intronic, disease-relevant regions throughout the complete genome. It provides the ideal basis for genetic diagnostics and can be understood as a clinical genome providing the most comprehensive sequencing data, which includes:

- Coding sequences
- Mitochondrial genome
- Deep intronic variants
- > 100x highly uniform coverage

Extra thorough analysis

Superior clinical sensitivity – Walking the extra mile

CeGaT's data analysis goes beyond regular exome diagnostics and increases solution rates. All relevant variants are evaluated using our proprietary software, including pathogenicity scoring and in-silico predictions. We also assess copy number variants, including heterozygous compound combinations of sequence variants (SNV, INDELs) with CNVs.

Extra insightful results

Superior clinical usability – Reports that benefit patient care

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

- Written by multiple experts
- Reviewed by experienced medical doctors
- Based on the most recent literature
- VUS re-evaluation (Single ExomeXtra®; Trio ExomeXtra®)

Our Diagnostic Services

Parallel analysis of all coding regions of the genome

Trio ExomeXtra®:

Comparative exome diagnostics between a patient and his/her parents based on the CeGaT unique enrichment. Trio exome diagnostics is the most comprehensive NGS-based genetic test available. Trio ExomeXtra® offers in addition to classical trio analysis, increased identification of clinically relevant variants in genes with reduced penetrance, variable expressivity, and imprinting effects. The medical report includes a discussion of variants identified in the context of the patient's phenotype. Available for many family constellations.

Single Exome:

- **ExomeXtra®**

  Whole exome diagnostics, utilizing our unique enrichment, of all genes related to the patients phenotype and relevant for differential diagnoses. The medical report includes a discussion of variants identified in the context of the patient's phenotype. Single ExomeXtra® offers the most comprehensive analysis for cases where a Trio ExomeXtra® is not possible.

- **ExomeFocus®**

  Whole exome diagnostics based on our unique high-quality exome enrichment and an evaluation of prioritized high-impact variants. ExomeFocus® can be used modularly and can be extended to a trio exome analysis at any time.

Prenatal ExomeXtra®:

- **Diagnostics – suspicious ultrasound**
  The analysis of the fetus and both parents (trio exome diagnostics) allows the most efficient evaluation and increases the chance of identifying disease-causing variants. All prenatal analyses are automatically prioritized.

- **Diagnostics – without ultrasound findings**
  Following trio exome analysis and filtering, we screen an extensive panel of more than 2,000 genes for all pathogenic and likely pathogenic (ACMG class 4, 5) genetic variants associated with severe early-onset disease. This diagnostics aims to clarify the risk for early-onset genetic disease.

Want to Discover More?

If you are interested in learning more about our products, we invite you to visit our website and have a look at our webinars, interviews, or brochures. Just scan the QR code with your smartphone and find out more.

www.cegat.com/exome-diagnostics

CeGaT

Compare Our Different Diagnostic Products

Single ExomeFocus®	Single ExomeXtra®	Trio ExomeXtra®
CeGaT's own exome enrichment: all known disease-causing regions		
✓	✓	✓
In-house developed, enhanced bioinformatics, data analysis, variant calling, and variant pathogenicity scoring		
✓	✓	✓
2-3 weeks turnaround time		
✓	✓	✓
Reporting pathogenic and likely pathogenic variants (ACMG classes 4 & 5)		
✓	✓	✓
Reporting variants of unknown significance (ACMG class 3, VUS)		
✓	✓	✓
A detailed discussion of medical implications of reported variants in the context of the patient		
✗	✓	✓
VUS re-evaluation		
✗	✓	✓
Accounting for imprinting, variable expressivity, and reduced penetrance (IVERP)		
✗	✗	✓
Costs		
€	€ €	€ € €
Solving rate		
XXX	XXXX	XXXXX

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, 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. We would be pleased to provide you with our award-winning service.

CAP ACCREDITED
COLLEGE of AMERICAN PATHOLOGISTS

CLIA CERTIFIED ID: 99D2130225

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

Accredited by DAkkS according to DIN EN ISO 15189:2014

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2023/03