Genetic Diagnostics

ExomeFocus®

CeGaT

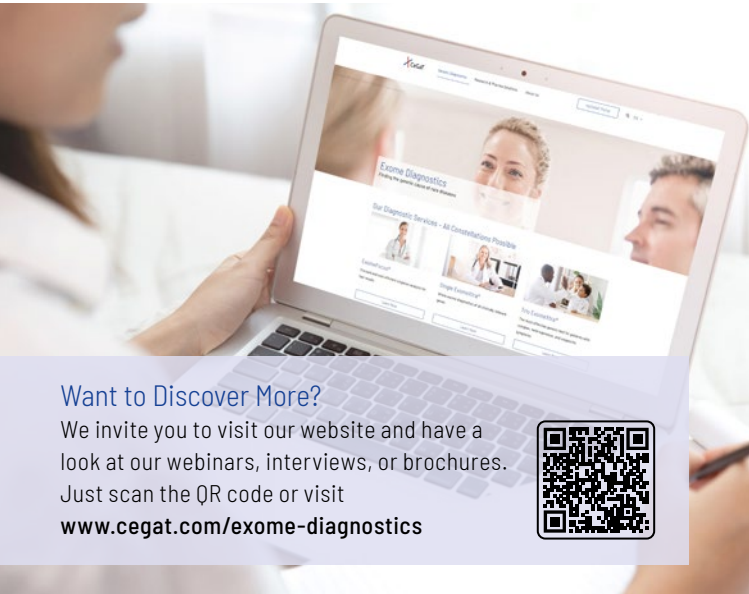
Exome Diagnostics

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.

ExomeFocus®

ExomeFocus® is the result of our many years of experience in genetic diagnostics and bioinformatics, representing 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 evaluates these high-impact variants to ensure clinical relevance for the patient's phenotype.

In case of unresolved cases, there is always an option to expand the analysis to Trio ExomeXtra®.



Want to Discover More?

We invite you to visit our website and have a look at our webinars, interviews, or brochures. Just scan the QR code or visit **www.cegat.com/exome-diagnostics**

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 HGMD and ClinVar. 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
- high 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 CNVs

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

About Us

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

Founded 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 and medical supervision.

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

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

Phone: +49 7071 56544-55
Fax: +49 7071 56544-56
Email: diagnostic-support@cegat.com
Web: www.cegat.com

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