TRANSFERRING CODE TO DIAGNOSIS

Order Form Molecular Diagnostics
General Information

CeGaT Center for Genomics and Transcriptomics

Patient

Surname: _____

First name: _____

Date of birth: _____

Sex: ❑ male ❑ female

Material

❑ Blood ____ ml (min. 5 ml EDTA-blood)

❑ Dried blood spot cards (at least 10 spots)

❑ DNA ____ µg (min. 5 µg DNA, concentr. ≥ 50 ng/µl) DNA-No.: _____

❑ Other specimen _____

External ID: _____

Date of sample collection: _____

Samples can be sent by mail in a cardboard box or air cushion envelope. Samples should not be exposed to direct sunlight. Dried blood spot cards can be ordered for free (info@cegat.com).

Sender / Clinic

Surname:

First name:

Institution:

Street:

Postcode/City:

Country:

Phone:

Email:

If applicable, please include a VAT number or a copy of your business registration certificate.

VAT: _____

Invoice

❑ to patient ❑ to sender / clinic

Declaration of consent

By signing this form, I declare that I have received comprehensive information about the genetic background related to the disease in question as well as the possibilities and limitations of molecular genetic testing. I understand that I have the right to withdraw my consent to genetic analyses.

I have been informed, and agree, that the data obtained in the analysis will be recorded, evaluated, or stored in an anonymized form in scientific databases, and further, in accordance with data protection and medical confidentiality, that the request, or parts thereof, may be transmitted to a specialized cooperating laboratory. I have been informed, and agree, that all data collected by CeGaT GmbH is electronically stored, processed, and used. I also consent to the data being transmitted electronically (e.g. by e-mail or fax).

If you do not check these boxes, your answer will be recorded as "No".

I consent to the storage of my genetic material for additional tests and/or quality control (for max. 10 years). ❑ Yes ❑ No

I consent to the storage of my test results beyond the timespan of 10 years (as required by German law). ❑ Yes ❑ No

I consent to the anonymous storage and use of surplus genetic material and/or test results for scientific research. ❑ Yes ❑ No

Genetic variation may sometimes be identified, which does not fit within the scope of the requested genetic analysis (so-called secondary findings report). The reporting of these variants is limited to pathogenic alterations within selected genes, for which a treatment or course of action exists for you or your family (according to the current guidelines of the American College of Medical Genetics and Genomics; ACMG SF V2.0; Kalia et al., 2017, PMID: 27854360). There is no claim of a comprehensive analysis of the genes included within the secondary findings report. An absence of secondary findings cannot be used to indicate a reduced disease risk.

With regard to secondary findings I would like:

❑ to be informed

❑ to NOT be informed

Please Note

We reserve the right to decide individually about the sequencing technology (Sanger / NGS).

This declaration of consent can be completely or partially withdrawn at any time. I have had sufficient time to consider giving my consent.

_____ Patient / Legal Guardian (Block letters)

_____ Doctor (Surname, First name)

X _____ Patient / Legal Guardian (Date, Signature)

X _____ Doctor (Date, Signature)

Doctor's stamp / Barcode

Contact

To discuss the diagnostic strategy please do not hesitate to contact us.
Phone: +49 7071 565 44-55
Email: diagnostic-support@cegat.de

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

CAP ACCREDITED COLLEGE of AMERICAN PATHOLOGISTS CLIA CERTIFIED ID: 99D2130225

CeGaT is accredited by DAkkS according to DIN EN ISO 15189:2014, by the College of American Pathologists (CAP) and CLIA.

Order Form
Molecular Diagnostics
Indication & Inquiry

Predictive genetic diagnosis

If you inquire a predictive diagnosis, please fill out and print the additional form „Predictive Genetic Diagnosis“.

According to German Gendiagnostikgesetz (GenDG, §7,1), the “predictive genetic examinations may only be conducted by medical doctors who are certified specialists in human genetics or by other medical doctors who within the framework of their own area of expertise were also able to obtain certification, specialization or additional qualification to conduct genetic examinations.“

By signing the „Predictive Genetic Diagnosis“ form, the physician submitting the request confirms that they have this qualification.

X _____
Doctor (signature)

Indication / Suspected Diagnosis

Further information

❑ autosomal dominant ❑ sporadic ❑ familial ❑ segregation to: _____

❑ autosomal recessive ❑ X-chromosomal ❑ consanguine Ethnic origin: _____

Pedigree

For a better description and illustration of the suspected familiy history, CeGaT offers a free Pedigree Chart Designer (PCD). You can find the PCD on our website or http://pedigree.cegat.de/.

Additional comments

Order Form
Molecular Diagnostics
Inquiry

Inquiries

A full list of more than 650 genes currently available for testing are listed under www.cegat.com. If your gene of interest is not included on the list, please do not hesitate to contact us.

Genes / OMIM No.

Inquiry for selected hotspot analyses

- ❑ **Achromatopsia (EYE04): CNGB3, c.1148delC; p.Thr383Ile*fs**13**
- ❑ **Optic atrophy (EYE17, LHON-Hotspots): MT-ND1, m.3460G>A; MT-ND4, m.11778G>A; MT-ND6, m.14484T>C**
- ❑ **Hereditary pancreatitis: PRSS1, c.365G>A; p.Arg122His; c.86A>T; p.Asn29Ile; c.47C>T; p.Ala16Val**
- ❑ **Pulmonary alveolar proteinosis: ABCA3, c.875A>T; p.Glu292Val**
- ❑ **Hemochromatosis Type 1: HFE, c.187C>G; p.His63Asp; c.845G>A; p.Cys282Tyr**

Additional analyses

For further information and advice please do not hesitate to contact our Diagnostic Support team.

www.cegat.de/en/diagnostic-support
diagnostic-support@cegat.de
Phone +49 7071 565 44-55