

### General Information

**Patient**

Surname: \_\_\_\_\_

First name: \_\_\_\_\_

Date of birth: \_\_\_\_\_

Sex:  male  female

**Material**

Blood \_\_\_\_ ml (min. 1-2 ml EDTA-blood)

Dried blood spot cards (at least 5 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).

#### Declaration of consent

By signing this form, I declare that I have received comprehensive information regarding 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 for genetic analyses.

I have been informed, and agree, that my personal data and the data obtained in the analysis will be recorded, evaluated or stored in an pseudonymized form in scientific databases, and that further, in accordance with data protection and medical confidentiality, the request, or parts thereof, may be transmitted to a specialized cooperating laboratory.

I consent to the re-evaluation of my test results within the data storage period. If significant alterations become apparent, my doctor will be informed by e-mail.

I have been informed, and agree to the electronic storage, processing, use, and transmission of all data collected by CeGaT GmbH.

For more detailed information on data privacy as well as your rights please refer to [www.cegat.de/en/privacy-policy](http://www.cegat.de/en/privacy-policy)

#### Please Note

Our panels are regularly updated to reflect current scientific research. It should therefore be recognized that there is the possibility that the list of genes on the order form may have changed slightly (genes added or removed) by the time the sample is analyzed in the laboratory. By signing this form, the patient accepts that the list of genes actually analyzed may be slightly different from what is currently listed. When NGS is utilized more than the requested genes are sequenced for each sample.

**This declaration of consent can be completely or partially withdrawn at any time.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

I, the referring physician, confirm that I am authorized to request genetic testing for the above-mentioned patient. For predictive testing, I confirm that I am authorized, and that I have fulfilled the requirements, to request this testing.

**If the patient did not sign this order form:** I, the referring physician, confirm that the patient received genetic counseling and agrees with the genetic testing. The patient's consent has been obtained in writing.

**Sender / Clinic**

Surname: \_\_\_\_\_

First name: \_\_\_\_\_

Institution: \_\_\_\_\_

Street: \_\_\_\_\_

Postcode/City: \_\_\_\_\_

Country: \_\_\_\_\_

Phone: \_\_\_\_\_

Email: \_\_\_\_\_

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

**Invoice**  to sender / clinic  to patient / other:

Surname: \_\_\_\_\_

First name: \_\_\_\_\_

Street: \_\_\_\_\_

Postcode/City: \_\_\_\_\_

Country: \_\_\_\_\_

Email: \_\_\_\_\_

**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 time-span of 10 years (as required by German law).  Yes  No

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

**With regard to secondary findings I would like:**

to be informed  to NOT be informed

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.

**Doctor's stamp / Barcode**



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

_____	_____
<b>Patient / Legal Guardian</b> (Block letters)	<b>Doctor</b> (Surname, First name)
<b>X</b> _____	<b>X</b> _____
<b>Patient / Legal Guardian</b> (Date, Signature)	<b>Doctor</b> (Date, Signature)

## 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: \_\_\_\_\_

Transplants (bone marrow, tissue, stem cells)     No     Yes, (please specify) \_\_\_\_\_

### Pedigree

-  index patient
- not affected
- affected
- known carrier
- deceased
-  unrelated parents
-  consanguine parents
-  unborn child
-  abortion, stillborn child
-  person of unknown sex
-  identical twins (monozygous)
-  fraternal twins (dizygous)

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

### Additional comments

**Inquiry****Inquiries**

A full list of more than 650 genes currently available for testing are listed under [www.cegat.com](http://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**

- |   |  |
|---|--|
| <input type="checkbox"/> <b>Achromatopsia (EYE04):</b> CNGB3, c.1148delC; p.Thr383Ilefs*13                                      | <input type="checkbox"/> <b>Pulmonary alveolar proteinosis:</b> ABCA3, c.875A>T; p.Glu292Val             |
| <input type="checkbox"/> <b>Optic atrophy (EYE17, LHON-Hotspots):</b> MT-ND1, m.3460G>A; MT-ND4, m.11778G>A; MT-ND6, m.14484T>C | <input type="checkbox"/> <b>Hemochromatosis Type 1:</b> HFE, c.187C>G; p.His63Asp; c.845G>A; p.Cys282Tyr |
| <input type="checkbox"/> <b>Hereditary pancreatitis:</b> PRSS1, c.365G>A; p.Arg122His; c.86A>T; p.Asn29Ile; c.47C>T; p.Ala16Val |  |

**Additional analyses**

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

**[www.cegat.de/en/diagnostic-support](http://www.cegat.de/en/diagnostic-support)  
[diagnostic-support@cegat.de](mailto:diagnostic-support@cegat.de)  
Phone +49 7071 56544-55**