Order Form
Exome & Custom Panel

General Information

Patient

Surname: _____

First name: _____

Date of birth: _____

Sex: ❑ male ❑ female

Material

❑ Blood ____ ml (min. 3 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

A 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
All genes, including the complete mtDNA are sequenced when exome diagnostics is performed. The diagnostic evaluation is limited to variants in genes relevant to the provided phenotypic information.

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
Exome & Custom Panel

Indication

Indication / Suspected Diagnosis:

Clinical Major Symptoms:

Preliminary genetic diagnostics:

Transplants (bone marrow, tissue, stem cells) ❑ No ❑ Yes, (please specify) _____

Please include a copy of all existing reports of your patient.

Pedigree

Consanguinity: ❑ Yes ❑ No Ethnic origin: _____

Family medical history

Are there other family members who currently have or have had the same or a similar disease as the patient?

❑ Yes ❑ No

If yes, please list the affected family members:

Name (not required)	Relationship to the patient (e.g. mother)	Age of onset	Diagnosis / Symptoms

Order Form
Exome & Custom Panel
Inquiry

Inquiry

- ❑ **Single exome: Exome diagnosis of the patient including medical findings (EXM01)**
- ❑ **Trio Exome: Exome diagnostics (coverage >100x) of patient and parents, incl. medical report (EXM03)**
- ❑ **Individual panel:** Please enter your desired genes below. We are happy to assist you with its compilation.

1. _____ 2. _____ 3. _____ 4. _____ 5. _____ 6. _____ 7. _____

8. _____ 9. _____ 10. _____ 11. _____ 12. _____ 13. _____ 14. _____

The analysis of the patient and both non-affected parents (trio exome) allows a more efficient evaluation of the variants identified in the index patient and leads to an increased chance of positive identification of the disesase causing variant(s).

Additional analysis

- ❑ **Genes to be considered in the context of exome diagnosis:** _____
- ❑ **Please perform Array-CGH diagnostics before exome diagnostics**
- ❑ **Deletion / duplication analysis (MLPA), Gene:** _____
- ❑ **Repeat expansion:** _____
- ❑ **ACMG genes:** I would like to be informed of relevant pathogenic variants within genes listed by the American College of Medical Genetics and Genomics (ACMG) according to current guidelines (ACMG SF V2.0; Kalia et al., 2017, PMID: 27854360). The analysis is limited to the sequence data generated for the primary request, and re-sequencing of regions with poor sequence coverage will not typically be performed. A negative “ACMG genes” report cannot be used to rule out disease risk. According to German legislation, predictive tests for minors may not be performed for diseases which have an onset in adulthood.

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

Order Form
Exome & Custom Panel
Declaration of consent Parent 1

Personal data (Family member)

Surname: _____ First name: _____

Date of birth: _____ Sample ID: _____

Relationship to the patient

❑ Father ❑ Mother ❑ Other; please state: _____

Does the family member suffer from the same or similar illness as the index patient?

❑ No ❑ Yes, symptoms are:

❑ **"ACMG genes":** I would like to be informed of relevant pathogenic variants within genes listed by the American College of Medical Genetics and Genomics (ACMG) according to current guidelines (ACMG SF V2.0; Kalia et al., 2017, PMID: 27854360). The analysis is limited to the sequence data generated for the primary request, and re-sequencing of regions with poor sequence coverage will not typically be performed. A negative "ACMG genes" report cannot be used to rule out disease risk. According to German legislation, predictive tests for minors may not be performed for diseases which have an onset in adulthood.

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❑ to NOT be informed

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_____ _____
Patient / Legal Guardian (Block letters) Doctor (Surname, First name)

X _____ **X** _____
Patient / Legal Guardian (Signature) Doctor (Date, Signature)

Doctor's stamp / Barcode

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Order Form
Exome & Custom Panel
Declaration of consent Parent 2

Personal data (Family member)

Surname: _____ First name: _____

Date of birth: _____ Sample ID: _____

Relationship to the patient

❑ Father ❑ Mother ❑ Other; please state: _____

Does the family member suffer from the same or similar illness as the index patient?

❑ No ❑ Yes, symptoms are:

❑ **"ACMG genes":** I would like to be informed of relevant pathogenic variants within genes listed by the American College of Medical Genetics and Genomics (ACMG) according to current guidelines (ACMG SF V2.0; Kalia et al., 2017, PMID: 27854360). The analysis is limited to the sequence data generated for the primary request, and re-sequencing of regions with poor sequence coverage will not typically be performed. A negative "ACMG genes" report cannot be used to rule out disease risk. According to German legislation, predictive tests for minors may not be performed for diseases which have an onset in adulthood.

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X _____ **X** _____
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