TRANSFERRING CODE TO DIAGNOSIS

Order Form 'Focused-Panel'

General Information

CeGaT Center for Genomics and Transcriptomics

Patient

Surname: _____

First name: _____

Date of birth: _____

Sex: ❑ male ❑ female

Material

❑ Blood ____ ml (min. 3 ml EDTA-blood)

❑ Dried blood spot cards (10 spots per patient)

❑ 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 anonymised 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 specialised 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 incidental 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 incidental findings report. An absence of incidental findings cannot be used to indicate a reduced disease risk.

With regard to incidental findings I would like:

❑ to be informed

❑ to NOT be informed

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.

_____ Patient / legal representative (Block letters)

_____ Doctor (Surname, First name)

X _____ Patient / legal representative (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.

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Indication

CeGaT
Center for Genomics and Transcriptomics

For targeted and effective processing, please complete the medical history form with as much detail as possible and include a copy of all existing reports.

Indication / Suspected Diagnosis

Pedigree

Consanguinity: ❑ Yes ❑ No Ethnic origin: _____

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.

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 (optional)	Relationship to the patient (e.g. mother)	Age of onset	Diagnosis / Symptoms

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Inquiry

- ❑ **Afibrinogenemia/Dysfibrinogenemia (3 Genes, SSP01)**
  FGA, FGB, FGG
- ❑ **Common variable immune deficiency (CVID) (13 Genes, SSP02)**
  CD19, CD81, CR2, CXCR4, ICOS, LRBA, MS4A1, NFKB1, NFKB2, PRKCD, TNFRSF13B, TNFRSF13C, TNFSF12
- ❑ **Tuberous sclerosis (2 Genes, SSP03)**
  TSC1, TSC2
- ❑ **Hereditary breast and ovarian cancer (small) (5 Genes, SSP04)**
  BRCA1, BRCA2, CHEK2, RAD51C, PALB2
- ❑ **Hereditary breast and ovarian cancer (large) (11 Genes, SSP05)**
  ATM, BRCA1, BRCA2, CDH1, CHEK2, PALB2, PTEN, RAD51C, RAD51D, STK11, TP53
- ❑ **Lynch syndrome (5 Genes, SSP06)**
  EPCAM, MLH1, MSH2, MSH6, PMS2
- ❑ **Hereditary hemorrhagic telangiectasia (HHT) (5 Genes, SSP07)**
  ACVRL1, ENG, GDF2, RASA1, SMAD4
- ❑ **Neurofibromatosis (3 Genes, SSP08)**
  NF1, NF2, SPRED1
- ❑ **Hyperekplexia (3 Genes, SSP09)**
  GLRA1, GLRB, SLC6A5
- ❑ **Holoprosencephaly (9 Genes, SSP10)**
  CDON, FGFR1, GLI2, PTCH1, SHH, SIX3, TDGF1, TGIF1, ZIC2
- ❑ **Refsum disease (8 Genes, SSP11)**
  AMACR, PEX1, PEX2, PEX26, PEX3, PEX5, PEX7, PHYH
- ❑ **Episodic ataxia (9 Genes, SSP12)**
  ATP1A3, CACNA1A, CACNB4, FGF14, KCNA1, KCNQ2, SCN2A, SLC1A3, SLC2A1
- ❑ **Dopa-responsive dystonia (3 Genes, SSP13)**
  GCH1, TH, SPR
- ❑ **Neuropathic pain syndromes (4 Genes, SSP14)**
  SCN9A, SCN10A, SCN11A, TRPA1
- ❑ **Malignant hyperthermia (3 Genes, SSP15)**
  RYR1, CACNA1S, STAC3
- ❑ **Familial intrahepatic cholestasis (4 Genes, SSP16)**
  ABCB11, ABCB4, ATP8B1, MYO5B
- ❑ **Maple syrup urine disease (4 Genes, SSP17)**
  BCKDHA, BCKDHB, DBT, DLD
- ❑ **Maturity onset diabetes of the young (MODY) (12 Genes, SSP18)**
  ABCC8, BLK, CEL, GCK, HNF1A, HNF1B, HNF4A, INS, KLF11, NEUROD1, PAX4, PDX1
- ❑ **Kabuki syndrome (5 Genes, SSP19)**
  KDM6A, KMT2D, CHD7, EYA1, IRF6
- ❑ **Craniosynostoses (7 Genes, SSP20)**
  FGFR1, FGFR2, FGFR3, TCF12, TWIST1, ERF, MSX2

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

www.cegat.de/en/diagnostic-support
diagnostic-support@cegat.de
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