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. ≥ 50ng/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.

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 (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: 9902130225

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

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 single gene analyses

Lipid metabolism disorder

- ❑ Familial defective apolipoprotein B-100 (partial sequence analysis of APOB exons 26 and 29)
- ❑ Autosomal dominant familial hypercholesterolemia (LDL receptor mutation)
- ❑ Autosomal dominant familal hypercholesterolemia type 3 (PCSK9 gene)
- ❑ Autosomal recessive familial hypercholesterolemia (ARH; LDLRAP1 gene)
- ❑ Lipoprotein lipase (LPL) deficiency
- ❑ Apolipoprotein C-II deficiency (hyperlipoproteinemia type Ib)
- ❑ Apolipoprotein A-V deficiency (late-onset hyperchylomicronemia)
- ❑ Cholesteryl ester storage disease and Wolman disease (LIPA gene)

Thrombophilia

- ❑ Thrombophilia due to activated protein C resistance (1691 G>A mutation exon 10 F5 gene)
- ❑ Thrombophilia due to thrombin defect (20210G>A mutation 3'-UTR F2 gene)

Liver and metabolic diseases

- ❑ Hemochromatosis type 1 (C282Y and H63D mutation; complete HFE gene analysis on request)
- ❑ Hemochromatosis type 4 (SLC40A1 gene)
- ❑ Hyperferritinemia-cataract syndrome (mutation in the IRE of the FTL gene)
- ❑ Wilson disease (ATP7B gene)
- ❑ Familial hyperbilirubinemia (UDP glucosyltransferase 1; TATA box mutation and p.Gly71Arg exchange UGT1A1 gene)
- ❑ Crigler-Najjar syndrome type 1 and 2 (UDP glucosyltransferase 1; UGT1A1 gene)
- ❑ Susceptibility to nonalcoholic fatty liver disease (PNPLA3 gene)
- ❑ Alagille syndrome (arteriohepatic dysplasia; JAG1 gene)

Pancreatitis

- ❑ Hereditary pancreatitis (cationic trypsinogen; PRSS1 gene)
- ❑ Hereditary pancreatitis (Carboxypeptidase A1; CPA1 gene)
- ❑ Hereditary pancreatitis (SPINK1 mutation as risk modifier)
- ❑ Hereditary pancreatitis (CTRC mutation as risk modifier)
- ❑ Cystic fibrosis (mono- or oligosymptomatic form; CFTR gene)

Exocrine pancreas insufficiency

- ❑ Shwachman-Bodian-Diamond syndrome (SBDS gene)

Angioedema / lymphedema

- ❑ Hereditary angioedema type I and II (deficiency of C1 esterase inhibitor; SERPING1 gene)
- ❑ Hereditary angioedema type III (mutation in exon 9 of the F12 gene)
- ❑ Hereditary lymphedema type IA (mutation in exons 17-25 of the FTL4 gene)

Lung disease

- ❑ Cystic fibrosis and mono- or oligosymptomatic presentations of the disease (e. g. bronchiectasis, CBAVD, pancreatitis; stepwise diagnostic: sequence analysis exon 11 → analysis of the 50 most common mutations found across populations of European origin plus poly-T tract intron 9 → complete sequence analysis of all 27 exons)
- ❑ Pulmonary surfactant metabolism dysfunction type 3 (ABCA3 deficiency)
- ❑ Pulmonary surfactant metabolism dysfunction type 1 (Surfactant protein B deficiency; SFTPB gene)
- ❑ Pulmonary surfactant metabolism dysfunction type 4 (mutation in the GM-CSF receptor α chain; CSF2RA gene)
- ❑ Pulmonary surfactant metabolism dysfunction type 2 (Surfactant protein C deficiency; SFTPC gene)
- ❑ Brain-thyroid-lung syndrome (NKX2-1 gene)

Muscle disease

- ❑ Myopathy due to myoadenylate deaminase deficiency (p.Q45* mutation; further stepwise diagnostic on request: 1) p.Q189H and p.K320I substitutions (exons 5 and 7); 2) complete sequence analysis of all 16 protein-coding exons of the AMPD1 gene)
- ❑ Chanarin-Dorfman syndrome (ABHD5 gene)
- ❑ Neutral lipid storage disease with myopathy (PNPLA2 gene)

Autoinflammatoery disease

- ❑ Familial Mediterranean fever (MEFV gene)
- ❑ Familial autosomal dominant periodic fever (TRAPS; mutation in exons 2, 3, 4, and 6 of the TNFRSF1A gene)
- ❑ Mevalonate kinase deficiency (Hyper-IgD syndrome, mevalonic aciduria, disseminated superficial actinic porokeratosis 3)
- ❑ Cryopyrin-associated periodic syndrome 1 (Familal cold-induced inflammatory syndrome 1, Muckle-Wells syndrome, CINCA syndrome; mutation in exons 3, 4, and 6 of the NLRP3 gene)
- ❑ Familial cold autoinflammatory syndrome 2 (exon 3 NLRP12 gene; complete gene analysis on request)
- ❑ Familial cold autoinflammatory syndrome 3 / autoinflammation, antibody deficiency, and immune dysregulation syndrome (PLCG2 gene)
- ❑ Familial cold autoinflammatory syndrome 4 / autoinflammation with infantile enterocolitis (NLRC4 gene)
- ❑ Pyogenic sterile arthritis, pyoderma gangrenosum, and acne syndrome (exons 10 and 11 PSTPIP1 gene; complete gene analysis on request)
- ❑ Progressive pseudorheumatoid arthropathy of childhood (PPAC; WISP3 gene)
- ❑ Interleukin 1 receptor antagonist deficiency (IL1RN gene)
- ❑ Blau syndrome/early-onset sarcoidosis (mutation in exon 4 of the NOD2 gene)

Gastroenterology

- ❑ Lactase persistence/nonpersistence (T/C exchange at position -13910 of the LCT gene)
- ❑ Hereditary fructose intolerance (aldolase B deficiency; ALDOB gene)
- ❑ Inflammatory bowel disease 1 (analysis of the three most frequent predisposing mutations in the NOD2 gene)

Amyloidosis

- ❑ Hereditary amyloidosis (FGA and TTR gene)
- ❑ Hereditary amyloidosis (APOA1, APOA2, and LYZ or GSN gene)
- ❑ Susceptibility to amyloidosis (SAA1 amino acids 70 and 75 encoded by exon 3)

Renal disease

- ❑ Renal glucosuria (SLC5A2 gene)
- ❑ Neurohypophyseal diabetes insipidus (AVP gene)
- ❑ Nephrogenic diabetes insipidus (AVPR2 gene)
- ❑ Nephrogenic diabetes insipidus (AQP2 gene)

Anemia / sickle cell disease

- ❑ Favism / Hemolytic anemia due to glucose-6-phosphate dehydrogenase deficiency (G6PD gene)
- ❑ Sickle cell anemia (HBB gene)

Blood vessel disease

- ❑ Hereditary hemorrhagic teleangiectasia of Rendu, Osler, and Weber (HHT; stepwise diagnostic: sequence analysis ENG gene → ACVRL1- gene → ENG-/ACVRL1-MLPA → SMAD4 gene)

Congenital neutropenia

- ❑ Autosomal dominant severe congenital neutropenia 1 (ELANE gene)
- ❑ Autosomal recessive severe congenital neutropenia 3 (HAX1 gene)
- ❑ Autosomal recessive severe congenital neutropenia 4 / Dursun syndrome (G6PC3 gene)
- ❑ Barth syndrome (TAZ gene)

Immunodeficiency /Autoimmune disease

- ❑ Autoimmune lymphoproliferative syndrome type IA (Canale-Smith syndrome; FAS gene)
- ❑ X-linked lymphoproliferative syndrome 1 (Duncan disease or Purtilo syndrome; SH2D1A gene)
- ❑ X-linked lymphoproliferative syndrome 2 (Duncan disease or Purtilo syndrome; XIAP gene)
- ❑ X-linked agammaglobulinemia 1 Bruton (BTK gene)
- ❑ X-linked hypogammaglobulinemia plus growth hormone deficiency (ELF4 gene)
- ❑ Autosomal dominant hyper-IgE recurrent infection syndrome (STAT3 gene)
- ❑ Cyclic neutropenia / autosomal dominant severe congenital neutropenia 1 (ELANE gene)
- ❑ X-linked chronic granulomatous disease (cgd91-phox/CYBB gene)

Pituitary hormone deficiency

- ❑ Combined pituitary hormone deficiency 1 (POU1F1 gene)
- ❑ Combined pituitary hormone deficiency 2 (PROP1 gene) and 5 (HESX1 gene)
- ❑ Combined pituitary hormone deficiency 3 (LHX3 gene)
- ❑ Combined pituitary hormone deficiency 4 (LHX4 gene)

Other endocrinopathies

- ❑ Hypocalciuric hypercalcemia type I / autosomal dominant hypocalcemia/ neonatal hyperparathyroidism (CASR gene)
- ❑ Autoimmune polyendocrine syndrome type I with or without reversible metaphyseal dysplasia (APS1; AIRE gene)

Skin disease

- ❑ Chanarin-Dorfman syndrome (ABHD5 gene)
- ❑ Chilblain lupus (TREX1 gene)
- ❑ Pustular psoriasis 14 (interleukin 36 receptor antagonist deficiency/DITRA; IL36RN gene)
- ❑ Oculocutaneous albinism type IA and IB (OCA1A+B; TYR gene)
- ❑ Disseminated superficial actinic porokeratosis (DSAP1 or POROK3; MVK gene)

Hereditary cancer predisposition syndrome

- ❑ Multiple endocrine neoplasia I (MEN1 gene)
- ❑ Multiple endocrine neoplasia IIA and IIB / medullary thyroid carcinoma / pheochromocytoma / renal agenesis (RET gene)
- ❑ von Hippel-Lindau syndrome (VHL gene)
- ❑ Li-Fraumeni syndrome (TP53 gene)
- ❑ Paragangliomas 1, 2, 3, 4, and/or 5 / pheochromocytoma (SDHD, SDHB, SDHC, SDHA, and/or SDH5/SDHAF2 gene)
- ❑ Familial gastrointestinal stromal tumor (GIST) / mast cell disease / piebaldism (KIT gene)

Additional analyses

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

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