ORDER FORM EXOME

For the diagnosis of syndromal complex phenotypes

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. 1-2 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:

VAT:

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

Invoice

❑ to sender / clinic

❑ to patient / other (KVA-No.: _____)

Surname:

First name:

Street:

Postcode/City:

Country:

Email:

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

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. Correct family relationships are assumed for comparative exome analysis using data from several family members (e.g. trio exome analysis).

This declaration of consent can be completely or partially withdrawn at any time.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

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.

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 have fulfilled the requirements to request this testing. For minors, I declare that I have the consent of all legal guardians. 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.

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 pseudonymous storage and use of surplus genetic material and/or test results for scientific research and in scientific literature. ❑ Yes ❑ No

With regard to secondary findings I would like to be informed: ❑ Yes ❑ No

Genetic variation may sometimes be identified, which does not fit within the scope of the requested genetic analysis (so-called secondary findings). The reporting of these variants is limited to pathogenic alterations (ACMG classes 4 and 5) 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; details on genes and associated diseases can be found at https://www.cegat.com/acmg-genes/). There is no claim of a comprehensive analysis of this gene set. An absence of secondary findings cannot be used to indicate a reduced disease risk.

Targeted analysis of the ACMG genes according to current recommendations can be requested as "additional analyses".

According to German Genetic Diagnostic Act (GenDG) we will issue the medical report to the counselling physician. Please indicate here the contact email of the counselling physician:

Email: _____

Physician's stamp / Barcode

Patient / Legal Guardian
(Block letters)

Physican
(Block letters)

X _____
Patient / Legal Guardian
(Date, Signature)

X _____
Physican
(Date, Signature)

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, the College of American Pathologists (CAP) and CLIA.

ORDER FORM EXOME

For the diagnosis of syndromal complex phenotypes

Indication & Clinical symptoms

Analysis type: ❑ Proband **is affected** ❑ Proband is **NOT affected** (predictive testing)

Indication / suspected diagnosis: _____

Preliminary genetic diagnostics: _____

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

Please include a copy of all existing reports of your patient (with pictures, if available).

Clinical symptoms

Please provide the following clinical information for your patient. Detailed clinical information will increase the likelihood of identifying causative alterations during exome analysis and significantly improve the interpretation of identified variants. The absence of phenotypic findings in organ systems, metabolic, or other molecular analyses is also valuable information for genetic analyses. Please take the opportunity to indicate 'no abnormalities' or 'not examined / unknown' in the relevant phenotype sections.

Prenatal medical history:
- ❑ Normal
- ❑ Prematurity
- ❑ Intrauterine growth restriction (IUGR)
- ❑ Poly- / Oligohydramnios
- ❑ Decreased fetal movement
- ❑ Other: _____

Developmental disorders
- ❑ Intellectual disability (❑ mild, ❑ moderate, ❑ severe)
- ❑ Global developmental delay
- ❑ Delayed motor milestones
- ❑ Delayed speech / language development
- ❑ Autism spectrum disorder
- ❑ Developmental regression
- ❑ Other: _____
- ❑ No intellectual disability
- ❑ No developmental disorder
- ❑ Not examined / unknown

Craniofacial anomalies
- ❑ Macrocephaly
- ❑ Microcephaly
- ❑ Craniosynostosis
- ❑ Broad forehead
- ❑ Cleft lip palate
- ❑ Hypertelorism
- ❑ Hypotelorism
- ❑ Abnormality of the nose (Please specify: _____)
- ❑ Abnormal ears (Please specify: _____)
- ❑ Micrognathia
- ❑ Oligodontia
- ❑ Other: _____
- ❑ No craniofacial anomalies
- ❑ Not examined / unknown

Brain abnormalities
- ❑ Lissencephaly
- ❑ Schizencephaly
- ❑ Porencephaly
- ❑ Pachygyria
- ❑ Polymicrogyria
- ❑ Band heterotopia
- ❑ Abnormality of corpus callosum (Please specify: _____)
- ❑ Hydrocephalus
- ❑ Holoprosencephaly
- ❑ Abnormality of basal ganglia
- ❑ Leukoencephalopathy
- ❑ Brain atrophy
- ❑ Ventriculomegaly
- ❑ Other: _____
- ❑ Normal brain MRI
- ❑ Not examined / unknown

Respiratory symptoms
- ❑ Respiratory insufficiency
- ❑ Respiratory failure
- ❑ Apnea
- ❑ Recurrent infections
- ❑ Bronchiectasis
- ❑ Other: _____
- ❑ No respiratory symptoms
- ❑ Not examined / unknown

Neurological symptoms
- ❑ Seizures (❑ generalized / ❑ focal)
- ❑ Encephalopathy
- ❑ Abnormal nerve conduction velocity
- ❑ Neuropathy (❑ motor / ❑ sensory)
- ❑ Ataxia
- ❑ Tremor
- ❑ Dystonia
- ❑ Chorea
- ❑ Spasticity
- ❑ Gait disturbances
- ❑ Nystagmus
- ❑ Mood disturbances (❑ anxiety, ❑ depression, ❑ psychosis)
- ❑ Migraine, ❑ Headaches
- ❑ Sleep disturbances
- ❑ Unexplained pain
- ❑ Other: _____
- ❑ No neurological symptoms
- ❑ Not examined / unknown

Eye defects
- ❑ Visual impairment (bilateral? ❑ yes / ❑ no) (Please specify: _____)
- ❑ Anophthalmia / ❑ Microphthalmia (bilateral? ❑ yes / ❑ no)
- ❑ Strabismus (bilateral? ❑ yes / ❑ no)
- ❑ Congenital bilateral cataract
- ❑ Other: _____
- ❑ No eye defects
- ❑ Not examined / unknown

Clinical symptoms

Hearing defects and vestibular abnormalities
- ❑ Sensorineural hearing impairment (bilateral? ❑ yes / ❑ no)
- ❑ Conductive hearing impairment (bilateral? ❑ yes / ❑ no)
- ❑ Abnormality of vestibular system (❑ vertigo, ❑ dizziness, ❑ imbalance, ❑ spatial disorientation)
- ❑ Other: _____

- ❑ No hearing defects
- ❑ No vestibular abnormalities
- ❑ Not examined / unknown

Musculoskeletal symptoms
- ❑ Muscular hypotonia
- ❑ Muscular hypertonia
- ❑ Elevated creatine kinase
- ❑ Ptosis
- ❑ Flexion contracture
- ❑ Arthrogryposis (congenital? ❑ yes / ❑ no)
- ❑ Short stature (skeletal dysplasia? ❑ yes / ❑ no)
- ❑ Tall stature (overgrowth? ❑ yes / ❑ no)
- ❑ Joint Hypermobility
- ❑ Hand- / ❑ Foot polydactyly (bilateral? ❑ yes / ❑ no)
- ❑ Hand- / ❑ Foot syndactyly (bilateral? ❑ yes / ❑ no)
- ❑ Camptodactyly of finger
- ❑ Clubfoot (congenital? ❑ yes / ❑ no)
- ❑ Scoliosis
- ❑ Pectus excavatum
- ❑ Pectus carinatum
- ❑ Hemihypertrophy
- ❑ Abnormality of bone density (❑ increased / ❑ decreased)
- ❑ Exostosis
- ❑ Delayed bone age
- ❑ Other: _____

- ❑ No muscular abnormalities
- ❑ No skeletal abnormalities
- ❑ Not examined / unknown

Cardiovascular defects
- ❑ Atrial septal defect
- ❑ Ventricular septal defect
- ❑ Abnormality of cardiac ventricle
- ❑ Tetralogy of Fallot
- ❑ Cardiomyopathy
- ❑ Arrhythmia
- ❑ Aortic aneurysm
- ❑ Abnormality of vasculature (Please specify: _____)
- ❑ Pulmonary arterial hypertension
- ❑ Other: _____

- ❑ No cardiac abnormalities
- ❑ Not examined / unknown

Immunological and hematological abnormalities
- ❑ Autoinflammatory disease
- ❑ Immunodeficiency (Please specify: _____)
- ❑ Recurrent infections
- ❑ Anemia (Erythrocytes)
- ❑ Neutropenia
- ❑ Thrombocytopenia
- ❑ Abnormal coagulation
- ❑ Megaloblastic anemia
- ❑ Bone marrow failure
- ❑ Hemochromatosis
- ❑ Other: _____

- ❑ No immunological abnormalities
- ❑ No hematological abnormalities
- ❑ Not examined / unknown

Metabolic and endocrine defects
- ❑ Failure to thrive
- ❑ Obesity
- ❑ Suspected mitochondriopathy
- ❑ Lactic acidosis
- ❑ Proteinuria
- ❑ Hyperglycemia
- ❑ Hypoglycemia
- ❑ Ketosis
- ❑ Hypercalcemia
- ❑ Diabetes mellitus
- ❑ Diabetes insipidus
- ❑ Hypothyroidism
- ❑ Hypoparathyroidism
- ❑ Exocrine pancreatic insufficiency
- ❑ Other: _____

- ❑ No metabolic abnormalities
- ❑ No endocrine abnormalities
- ❑ Not examined / unknown
- ❑ Copy of laboratory findings attached

Renal and genitourinary tract abnormalities
- ❑ Renal cysts
- ❑ Renal agenesis
- ❑ Horseshoe kidney
- ❑ Hypercalciuria
- ❑ Hematuria
- ❑ Proteinuria
- ❑ Hypospadias
- ❑ Cryptorchidism
- ❑ Ambiguous genitalia
- ❑ Other: _____

- ❑ No renal abnormalities
- ❑ No genitourinary abnormalities
- ❑ Not examined / unknown

Hepatic dysfunction
- ❑ Liver dysfunction (Please specify: _____)
- ❑ Recurrent acute liver failure
- ❑ Hepatic cysts
- ❑ Cholestasis
- ❑ Hypercholanemia
- ❑ Hepatomegaly
- ❑ Other: _____

- ❑ No hepatic abnormalities
- ❑ Not examined / unknown

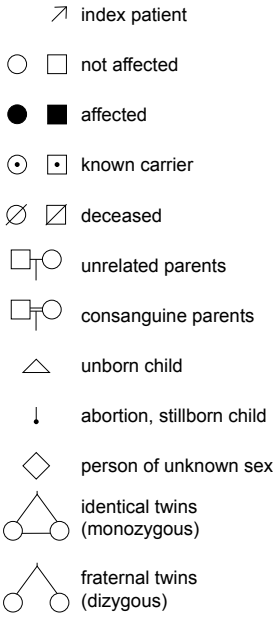
Skin, nails and hair
- ❑ Abnormality of connective tissue (Please specify: _____)
- ❑ Multiple cafe-au-lait spots
- ❑ Port-wine stain
- ❑ Albinism
- ❑ Progeroid appearance
- ❑ Skin lesions
- ❑ Eczema
- ❑ Edema
- ❑ Ichthyosis
- ❑ Dysplastic nails
- ❑ Anhidrosis
- ❑ Hyperhidrosis
- ❑ Alopecia
- ❑ Hypertrichosis (Where? _____)
- ❑ Other: _____

- ❑ No abnormalities of skin, nails and hair
- ❑ Not examined / unknown

Other
- ❑ Organomegaly (which? _____)
- ❑ Neoplasm / ❑ Cancer
- ❑ Pancreatitis
- ❑ Episodic fever
- ❑ Hyperthermia
- ❑ Hypothermia
- ❑ Constipation, ❑ Obstipation
- ❑ Diarrhea
- ❑ Episodic vomiting
- ❑ Other: _____

Indication

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

Inquiry

Single Exome Diagnostics:

- ❑ **ExomeFocus®:** Exome diagnostics based on our proprietary, high-quality exome enrichment. The medical report focuses on pathogenic and likely-pathogenic variants in the context of the patient's phenotype (ACMG classes 3, 4, 5) (EXM01FOCUS).
- ❑ **ExomeXtra®:** Exome diagnostics based on our proprietary, high-quality exome enrichment. The medical report focuses on pathogenic, likely-pathogenic variants and VUS in the context of the patient's phenotype (ACMG classes 3, 4, 5) and comprises a discussion of these variants (EXM01).

Comparative Exome Diagnostics:

- ❑ **Duo ExomeXtra®:** Comparative exome diagnostics between patient and a family member, incl. a medical report with a discussion of variants in the context of the patient's phenotype; reporting ACMG classes 3 , 4, 5 (EXM02Duo)
- ❑ **Trio ExomeXtra®:** Comparative exome diagnostics between patient and his/her parents, incl. a medical report with a discussion of variants in the context of the patient's phenotype; reporting ACMG classes 3 , 4, 5 (EXM02)
- ❑ **Upgrade ExomeFocus® to Trio ExomeXtra®:** Expansion of a Single ExomeFocus® analysis to a Trio ExomeXtra®. Please provide in addition to the parental samples, CeGaT-ID of the previously analysed index: _____

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 disease causing variants.

Additional analyses (additional fees may apply)

❑ Genes to be considered in the context of Single Exome Diagnostics (ExomeXtra® only): _____

_____

_____

❑ Please perform array-CGH diagnostics

- ❑ prior **or**
- ❑ parallel

to exome diagnostics.

❑ Deletion / Duplication analysis (MLPA), gene(s) of interest: _____

❑ Repeat expansion: _____

❑ **HLA-Typing (HLA01)**
I would like to receive an additional report stating the HLA alleles (HLA class I (Gene A, B, C) and HLA class II (Gene DPA1, DPB1, DQA1, DQB1, DRB1, DRB3, DRB4, DRB5)).

❑ **ACMG genes diagnostics**
I would like to be informed of relevant alterations within the list of recommended genes for secondary analysis, according to the current guidelines of the American College of Medical Genetics and Genomics. The analysis is restricted to the sequence data, re-sequencing of regions with poor sequence coverage will not typically be performed. A negative "ACMG genes" report cannot be used to rule out (genetic) disease risk. Additional fees may apply. According to German legislation, predictive tests for minors may not be performed for diseases which have an onset in adulthood. Therefore, some genes will not be analyzed for minors, unless the phenotypic spectrum is within the scope of the primary medical indication of the patient.
Details on genes and associated diseases can be found at https://www.cegat.com/acmg-genes/

❑ **Pharmacogenetics (PGX) (22 genes)**
ABCG2, CACNA1S, CYP2B6, CYP2C19, CYP2C9, CYP2D6, CYP3A4, CYP3A5, CYP4F2, DPYD, G6PD, HLA-A, HLA-B, IFNL3, MT-RNR1, NUDT15, POR, RYR1, SLCO1B1, TPMT, UGT1A1, VKORC1

I would like to receive an additional report analyzing known variants that are involved in the metabolism of pharmaceutical products.

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

www.cegat.com/diagnostic-support | sales@cegat.com | Phone +49 7071 565 44-55

Additional Information

Please use this space to provide any additional relevant information.

ORDER FORM EXOME

For the diagnosis of syndromal complex phenotypes

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: _____

❑ **HLA-Typing (HLA01)**
I would like to receive an additional report stating the HLA alleles (HLA class I (Gene A, B, C) and HLA class II (Gene DPA1, DPB1, DQA1, DQB1, DRB1, DRB3, DRB4, DRB5)).

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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.

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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. For minors, I declare that I have the consent of all legal guardians.

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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

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With regard to secondary findings I would like to be informed: ❑ Yes ❑ No

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(Block letters)

Doctor
(Block letters)

X _____
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(Date, Signature)

X _____
Doctor
(Date, Signature)

Doctor's stamp / Barcode

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ORDER FORM EXOME

For the diagnosis of syndromal complex phenotypes

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?

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