Order Form
Custom Panel

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).

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:

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

In some cases, all genes, including the complete mtDNA, are sequenced when molecular genetic 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This declaration of consent can be completely or partially withdrawn at any time.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.

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.

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. ❑ Yes ❑ No

With regard to secondary findings I would like:

❑ to be informed ❑ to NOT be informed

Incidental findings are pathogenic or likely pathogenic alterations (class V and IV) according to the ACMG classification criteria (Richards et al., 2015, PMID: 25741868). These findings may sometimes be identified in next-generation sequencing data, and may not fit within the scope of the requested genetic analysis. The reporting of these variants (so-called 'incidental findings report') is limited to alterations within genes for which a treatment or course of action exists for you or your family (based on the current guidelines of the American College of Medical Genetics and Genomics; ACMG SF V2.0; Kalia et al., 2017, PMID: 27854360). An absence of incidental findings cannot be used to indicate an absence of (genetic) disease risk. Incidental findings are identified by chance during the analysis of the primary request, and no targeted analysis of the ACMG genes is performed.

Patient / Legal Guardian
(Block letters)

Doctor
(Surname, First name)

X _____
Patient / Legal Guardian
(Date, Signature)

X _____
Doctor
(Date, Signature)

Doctor's stamp / Barcode

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.

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Indication

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.

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

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Major 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: _____

Additional Information

Please use this space to provide any additional relevant information.

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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

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Inquiry

Inquiry

❑ **Custom panel:** Please enter your desired genes below (or copy genes into next page). Please feel free to contact us for advice regarding the selection of relevant genes.

S Custom Geneset

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

8. _____ 9. _____

M Custom Geneset

10. _____ 11. _____ 12. _____ 13. _____ 14. _____ 15. _____ 16. _____

17. _____ 18. _____ 19. _____ 20. _____ 21. _____ 22. _____ 23. _____

24. _____ 25. _____ 26. _____ 27. _____ 28. _____ 29. _____

L Custom Geneset

30. _____ 31. _____ 32. _____ 33. _____ 34. _____ 35. _____ 36. _____

37. _____ 38. _____ 39. _____ 40. _____ 41. _____ 42. _____ 43. _____

44. _____ 45. _____ 46. _____ 47. _____ 48. _____ 49. _____ 50. _____

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

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Additional Information

Please use this space to provide any additional relevant information.