

General Information

Patient

Surname, first name: _____

Date of birth: _____

Sex (assigned at birth): female male

Gender (if differs from sex assigned at birth):
 man non-binary woman self-described: _____

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 _____

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

Please Note: If you have previously had a stem cell transplant, please **do not send in EDTA blood**, but a **saliva sample**.

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 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



CLIA CERTIFIED ID: 99D2130225

CeGaT is accredited by DAKkS according to DIN EN ISO 15189, the College of American Pathologists (CAP) and CLIA.

Patient / Legal Guardian
 (Block letters)

Physician
 (Block letters)

X _____
Patient / Legal Guardian
 (Date, Signature)

X _____
Physician
 (Date, Signature)

Indication & Clinical symptoms

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

Indication / suspected diagnosis: _____

Previous genetic testing: _____
 (If performed, please specify test and results)

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.

| | | |
|---|--|---|
| <p>Prenatal medical history:</p> <input type="checkbox"/> Normal <input type="checkbox"/> Prematurity <input type="checkbox"/> Intrauterine growth restriction (IUGR) <input type="checkbox"/> Poly- / Oligohydramnios <input type="checkbox"/> Decreased fetal movement <input type="checkbox"/> Other: _____ <p>Developmental disorders</p> <input type="checkbox"/> Intellectual disability (<input type="checkbox"/> mild, <input type="checkbox"/> moderate, <input type="checkbox"/> severe) <input type="checkbox"/> Global developmental delay <input type="checkbox"/> Delayed motor milestones <input type="checkbox"/> Delayed speech / language development <input type="checkbox"/> Autism spectrum disorder <input type="checkbox"/> Developmental regression <input type="checkbox"/> Other: _____ <input type="checkbox"/> No intellectual disability <input type="checkbox"/> No developmental disorder <input type="checkbox"/> Not examined / unknown <p>Craniofacial anomalies</p> <input type="checkbox"/> Macrocephaly <input type="checkbox"/> Microcephaly <input type="checkbox"/> Craniosynostosis <input type="checkbox"/> Broad forehead <input type="checkbox"/> Cleft lip palate <input type="checkbox"/> Hypertelorism <input type="checkbox"/> Hypotelorism <input type="checkbox"/> Abnormality of the nose (Please specify: _____) <input type="checkbox"/> Abnormal ears (Please specify: _____) <input type="checkbox"/> Micrognathia <input type="checkbox"/> Oligodontia <input type="checkbox"/> Other: _____ <input type="checkbox"/> No craniofacial anomalies <input type="checkbox"/> Not examined / unknown | <p>Brain abnormalities</p> <input type="checkbox"/> Lissencephaly <input type="checkbox"/> Schizencephaly <input type="checkbox"/> Porencephaly <input type="checkbox"/> Pachygyria <input type="checkbox"/> Polymicrogyria <input type="checkbox"/> Band heterotopia <input type="checkbox"/> Abnormality of corpus callosum (Please specify: _____) <input type="checkbox"/> Hydrocephalus <input type="checkbox"/> Holoprosencephaly <input type="checkbox"/> Abnormality of basal ganglia <input type="checkbox"/> Leukoencephalopathy <input type="checkbox"/> Brain atrophy <input type="checkbox"/> Ventriculomegaly <input type="checkbox"/> Other: _____ <input type="checkbox"/> Normal brain MRI <input type="checkbox"/> Not examined / unknown <p>Respiratory symptoms</p> <input type="checkbox"/> Respiratory insufficiency <input type="checkbox"/> Respiratory failure <input type="checkbox"/> Apnea <input type="checkbox"/> Recurrent infections <input type="checkbox"/> Bronchiectasis <input type="checkbox"/> Other: _____ <input type="checkbox"/> No respiratory symptoms <input type="checkbox"/> Not examined / unknown | <p>Neurological symptoms</p> <input type="checkbox"/> Seizures (<input type="checkbox"/> generalized/ <input type="checkbox"/> focal) <input type="checkbox"/> Encephalopathy <input type="checkbox"/> Abnormal nerve conduction velocity <input type="checkbox"/> Neuropathy (<input type="checkbox"/> motor/ <input type="checkbox"/> sensory) <input type="checkbox"/> Ataxia <input type="checkbox"/> Tremor <input type="checkbox"/> Dystonia <input type="checkbox"/> Chorea <input type="checkbox"/> Spasticity <input type="checkbox"/> Gait disturbances <input type="checkbox"/> Nystagmus <input type="checkbox"/> Mood disturbances (<input type="checkbox"/> anxiety, <input type="checkbox"/> depression, <input type="checkbox"/> psychosis) <input type="checkbox"/> Migraine, <input type="checkbox"/> Headaches <input type="checkbox"/> Sleep disturbances <input type="checkbox"/> Unexplained pain <input type="checkbox"/> Other: _____ <input type="checkbox"/> No neurological symptoms <input type="checkbox"/> Not examined / unknown <p>Eye defects</p> <input type="checkbox"/> Visual impairment (bilateral? <input type="checkbox"/> yes/ <input type="checkbox"/> no) (Please specify: _____) <input type="checkbox"/> Anophthalmia/ <input type="checkbox"/> Microphthalmia (bilateral? <input type="checkbox"/> yes/ <input type="checkbox"/> no) <input type="checkbox"/> Strabismus (bilateral? <input type="checkbox"/> yes/ <input type="checkbox"/> no) <input type="checkbox"/> Congenital bilateral cataract <input type="checkbox"/> Other: _____ <input type="checkbox"/> No eye defects <input type="checkbox"/> 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
- Arthrogyrosis (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: _____

Request

Request

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.

1. _____ 2. _____ 3. _____ 4. _____ 5. _____ 6. _____ 7. _____ **S** Custom Geneset
8. _____ 9. _____

10. _____ 11. _____ 12. _____ 13. _____ 14. _____ 15. _____ 16. _____ **M** Custom Geneset
17. _____ 18. _____ 19. _____ 20. _____ 21. _____ 22. _____ 23. _____
24. _____ 25. _____ 26. _____ 27. _____ 28. _____ 29. _____

30. _____ 31. _____ 32. _____ 33. _____ 34. _____ 35. _____ 36. _____ **L** Custom Geneset
37. _____ 38. _____ 39. _____ 40. _____ 41. _____ 42. _____ 43. _____
44. _____ 45. _____ 46. _____ 47. _____ 48. _____ 49. _____ 50. _____

Additional analyses (additional fees may apply)

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 www.cegat.com/acmg-genes

Pharmacogenetics (PGX)

I would like to receive an additional report analyzing known variants that are involved in the metabolism of pharmaceutical products. Details can be found at www.cegat.com/pgx

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