

Patient

Surname: _____

First name: _____

Date of birth: _____

Sex: male female

Material

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

Dried blood spot cards (at least 10 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: _____

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. 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 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

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 Guardian (Block letters)

Doctor (Surname, First name)

X _____
Patient / Legal Guardian (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



CeGaT is accredited by DAKKS according to DIN EN ISO 15189:2014, by the College of American Pathologists (CAP) and CLIA.

Indication / Suspected Diagnosis:

Clinical Major Symptoms:

Preliminary genetic diagnostics:

Please include a copy of all existing reports of your patient.

Pedigree

Consanguinity: Yes No

Ethnic origin: _____

-  index patient
- not affected
- affected
- known carrier
- deceased
-  unrelated parents
-  consanguine parents
-  unborn child
-  abortion, stillborn child
-  person of unknown sex
-  identical twins (monozygous)
-  fraternal twins (dizygous)

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

Clinical Features

1. Relevant clinical history

	Yes	No
Jaundice	<input type="checkbox"/>	<input type="checkbox"/>
Liver transplantation	<input type="checkbox"/>	<input type="checkbox"/>
Elevated levels aminotransferases	<input type="checkbox"/>	<input type="checkbox"/>
Normal γ -GT activity	<input type="checkbox"/>	<input type="checkbox"/>
Elevated levels of bilirubin	<input type="checkbox"/>	<input type="checkbox"/>
Malformation of bile ducts	<input type="checkbox"/>	<input type="checkbox"/>
a) Hepatomegaly	<input type="checkbox"/>	<input type="checkbox"/>
b) Splenomegaly	<input type="checkbox"/>	<input type="checkbox"/>
Pruritus	<input type="checkbox"/>	<input type="checkbox"/>

other symptoms: _____

2. Signs of dysmorphism

No Yes; which?: _____

3. Genetic analyses

Not performed Yes, please attach copy of results if patient agrees

Array-CGH: No Yes

Sequencing: No Yes

Other: _____

4. Metabolic diseases

No Yes, please attach copy of results if patient agrees

5. Further comments

For babies and infants

6. Pregnancy history

Normal Bleedings

Medication: _____

Infection: _____

Jaundice: _____

Pruritus: _____

Other: _____

7. Birth data

Size: _____ Weight: _____ Head circumference: _____

Umbilical cord pH-value: _____

Date of birth: _____ On time Abnormal: _____

Noticeable problems: _____

Additional comments

Inquiry

Large Panel Diagnostic Option (118 Genes, LIV-D45)

In addition to the full analysis of the requested gene set(s), we will expand the analysis to all pathogenic and likely pathogenic variants (ACMG class 4 and 5) in genes which are related to the indication described for the proband on the entire diagnostic panel (118 genes in total).

Familial cholestasis (8 Genes, LIV01)

ABCB11, ABCB4, ABCG5, ABCG8, ATP8B1, DCDC2, MYO5B, NR1H4

Hypercholanemia and defects of bile acid synthesis (11 Genes, LIV02)

ABCD3, AKR1D1, AMACR, BAAT, CYP27A1, CYP7A1, CYP7B1, EPHX1, HSD3B7, SLC10A2, TJP2

Impairments of transport in hepatocytes and cholangiocytes (8 Genes, LIV03)

ABCC2, ATP7B, CFTR, CIRH1A, SERPINA1, VIL1, VIPAS39, VPS33B

Impairments of organogenesis (10 Genes, LIV04)

CC2D2A, CLDN1, INVS, JAG1, NOTCH2, PKD2, PKHD1, RPGRIP1L, TMEM67, ZIC3

Metabolic disorders of hepatocytes, including Tyrosinemia, Glycogen storage diseases, Hyperammonemia, Shwachman-Diamond syndrome, Disorder of fatty acid oxidations and Peroxisomal diseases (59 Genes, LIV05)

ACADM, ACADVL, ADK, AGL, AGPAT2, ALAD, ALDOB, ASL, ASS1, BSCL2, CCDC115, COG1, COG6, COG7, CPS1, ETFA, ETFB, ETFDH, FAH, G6PC, GAA, GALE, GALT, GBE1, GYS2, IARS, KRT18, KRT8, LMNA, MPI, NGLY1, NR1H4, OTC, PEX1, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PKLR, PMM2, PTRF, PYGL, SBDS, SLC37A4, SLC39A8, STT3B, TTC37

Lysosomal storage disorders (5 Genes, LIV06)

GBA, LIPA, NPC1, NPC2, SMPD1

Recurrent acute liver failure (6 Genes, LIV07)

DLD, EIF2AK3, HMGCL, LARS, NBAS, TRMU

Hepatic mitochondrialriopathies (17 Genes, LIV08)

BCS1L, C10orf2, CPT1A, CPT2, DGUOK, FARS2, GFM1, HADHA, MPV17, MRPS16, POLG, SCO1, SLC25A20, SUCLG1, TRMU, TSFM, TYMP

Additional analyses

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