Order Form
Prenatal Exome Diagnostics

General Information

Fetus of

Surname of mother: _____

First name of mother: _____

Sex of Fetus: ❑ male ❑ female ❑ unknown

Has MCC testing been performed? ❑ Yes ❑ No

Material

❑ Amniotic fluid ❑ Chorionic villi ❑ Starting material has been cultivated

❑ Abortion material

❑ Extracted DNA ____ µg (min. 5 µg DNA, concentr.. ≥ 50 ng/µl)

from DNA-Nr.: _____

❑ Other specimen _____

External ID: _____

Date of sample collection: _____

Pregnancy week and estimated due date: _____

Samples can be sent by mail in a cardbox 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

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.

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.

Order Form
Prenatal Exome Diagnostics

Indication

Please attach copies of medical reports (including ultrasound or MRI reports, if available). The variant interpretation is based on clinical information available at the time of analysis.

Indication / Suspected diagnosis:

Ultrasound medical report available? ❑ Yes (please attach copy) ❑ No

Clinical symptoms:

Preliminary genetic diagnostics for fetus or parents?

Chromosome / Karyotype analysis: ❑ Yes (please attach copy) ❑ No

Array-CGH: ❑ Yes (please attach copy) ❑ No

Other

Pedigree

Consanguinity: ❑ Yes ❑ No Ethnic origin: _____

Family medical history

If the mother of the fetus has been pregnant in the past, were there any anomalies during pregnancy? ❑ Yes ❑ No

Are there other family members who currently have or have had a disease or disorder relevant for the clinical indication of the fetus? ❑ Yes ❑ No

If yes, please list the affected family members:

Name (not required)	Relationship to the fetus (e.g. mother)	Age of onset	Diagnosis / Symptoms

Order Form
Prenatal Exome Diagnostics
Inquiry

Inquiry – Exome

❑ **Single Exome:** Exome diagnostics of the fetus including medical report (EXM01)

❑ **Maternal cell contamination (MCC) testing (please provide material from the mother of the fetus, EDTA blood recommended)**

❑ **Trio Exome:** Comparative exome diagnostics between fetus and parents incl. medical report (EXM02)

❑ **Genes to be considered in the context of exome diagnostics:**

The analysis of the fetus and both non-affected parents (Trio Exome) allows a more efficient evaluation of the variants identified in the fetus and leads to an increased chance of positive identification of the disease causing variants.

Additional analysis (additional fees may apply)

❑ Please perform array-CGH diagnostics
- ❑ prior **or**
- ❑ parallel

to exome diagnostics.

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

Order Form
Prenatal Exome Diagnostics

Additional Information

Please use this space to provide any additional relevant information.

Order Form
Prenatal Exome Diagnostics

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 an illness or disorder with (suspected) genetic cause?

❑ No ❑ Yes, symptoms are:

❑ **ACMG genes diagnostics for adults (59 genes)/ for minors (52 genes)**

ACTA2, ACTC1, APC, APOB, ATP7B, BMPR1A, BRCA1*, BRCA2*, CACNA1S, COL3A1, DSC2, DSG2, DSP, FBN1, GLA, KCNH2, KCNQ1, LDLR, LMNA, MEN1, MLH1*, MSH2*, MSH6*, MUTYH*, MYBPC3, MYH11, MYH7, MYL2, MYL3, NF2, OTC, PCSK9, PKP2, PMS2*, PRKAG2, PTEN, RB1, RET, RYR1, RYR2, SCN5A, SDHAF2, SDHB, SDHC, SDHD, SMAD3, SMAD4, STK11, TGFBR1, TGFBR2, TMEM43, TNNI3, TNNT2, TP53, TPM1, TSC1, TSC2, VHL, WT1

I would like to be informed of relevant alterations within the above listed genes that have been selected by the American College of Medical Genetics and Genomics (ACMG). The analysis is performed according to current guidelines (ACMG SF V2.0; Kalia et al., 2017, PMID: 27854360) and restricted to the sequence data generated for the primary request. 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, the genes BRCA1, BRCA2, MLH1, MSH2, MSH6, MUTYH and PMS2 will not be analyzed for minors, unless the phenotypic spectrum is within the scope of the primary medical indication of the patient.

Declaration of consent

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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.

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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:

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Patient
(Block letters)

Doctor
(Surname, First name)

X _____
Patient
(Date, Signature)

X _____
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Doctor's stamp / Barcode

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Order Form
Prenatal Exome Diagnostics

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 an illness or disorder with (suspected) genetic cause?

❑ No ❑ Yes, symptoms are:

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