Order Form
Prenatal ExomeXtra® – Inconspicuous Ultrasound

General Information

Fetus of

Surname of mother: _____

First name of mother: _____

Birthday 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

❑ Extracted DNA ____ µg (min. 1-2 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 (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 physican 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.com/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. 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.

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: _____

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

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Indication

Please attach copies of medical reports (including ultrasound or MRI reports, if available).

Previous genetic testing for fetus or parents?

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

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

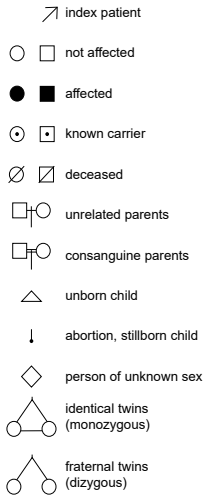
Other _____

Known genetic carrier status (incl. family members) - please attach copy of reports

_____

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

Do any family members currently have, or have previously had, a congenital or early onset disease or disorder with (suspected) genetic cause? ❑ 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

Request – Exome

❑ **Prenatal Trio Exome inconspicuous ultrasound findings:**
Comparative exome diagnostics between fetus and parents incl. medical report (EXM02PWU)

Additional analyses (additional fees may apply)

❑ **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**

❑ **Fetal sex determination**
I request that the report includes information on the chromosomal sex of the fetus. I confirm that disclosure of this information is permitted under the applicable legal and regulatory framework.

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Gene list for counselling

Please find below the list of genes associated with severe childhood-onset disease, which are analysed within the trio diagnostics pipeline for fetuses without abnormal ultrasound findings. Late-onset disease and carrier status will not be reported.

AAAS, AARS1, AARS2, ABAT, ABCA12, ABCA3, ABCA4, ABCB11, ABCB4, ABCB7, ABCC6, ABCC8, ABCC9, ABCD1, ABCD4, ABHD12, ABHD5, ACACA, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACADVL,DLG4, ACAN, ACAT1, ACD, ACE, ACO2, ACOX1, ACOX2, ACP5, ACSF3, ACSL4, ACTA1, ACTA2, ACTB, ACTG1, ACTG2, ACTL6B, ACTN2, ACVR2B, ACVRL1, ACY1, ADA, ADA2, ADAM17, ADAM22, ADAMTS13, ADAMTS19, ADAMTS2, ADAMTSL2, ADAR, ADARB1, ADAT3, ADCY1, ADCY5, ADCY6, ADGRG1, ADGRG6, ADGRV1, ADK, ADNP, ADPRS, ADSL, AFF2, AFG2A, AFG3L2, AGA, AGK, AGL, AGPAT2, AGPS, AGRN, AGT, AGTPBP1, AGTR1, AGXT, AHCY, AHDC1, AHI1, AIFM1, AIMP1, AIMP2, AIPL1, AIRE, AK2, AKR1D1, AKT1, AKT2, AKT3, AKT3,SDCCAG8, ALAD, ALDH18A1, ALDH1A3, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALMS1, ALOX12B, ALOXE3, ALPL, ALS2, ALX3, ALX4, AMACR, AMER1, AMN, AMPD1, AMPD2, AMT, ANK2, ANK3, ANKH, ANKLE2, ANKRD11, ANKS6, ANO10, ANO5, ANOS1, ANTXR1, ANTXR2, AP1B1, AP1S1, AP1S2, AP3B1, AP3B2, AP3D1, AP4B1, AP4E1, AP4M1, AP4S1, APC2, APTX, AQP2, AR, ARCN1, ARFGEF2, ARG1, ARHGAP31, ARHGDIA, ARHGEF9, ARID1A, ARID1B, ARID2, ARL13B, ARL3, ARL6, ARL6IP1, ARMC9, ARNT2, ARPC1B, ARSA, ARSB, ARSL, ARV1, ARX, ASAH1, ASCC1, ASH1L, ASL, ASNS, ASPA, ASPH, ASPM, ASS1, ASXL1, ASXL2, ASXL3, ATAD1, ATAD3A, ATCAY, ATG7, ATIC, ATL1, ATM, ATOH7, ATP13A2, ATP1A1, ATP1A2, ATP1A3, ATP2B3, ATP5F1D, ATP5MK, ATP6AP1, ATP6AP2, ATP6V0A2, ATP6V0A4, ATP6V1A, ATP6V1B1, ATP6V1B2, ATP6V1E1, ATP7A, ATP7B, ATP8A2, ATP8B1, ATPAF2, ATR, ATRX, AUH, AUTS2, AVIL, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT1, B4GALT7, B4GAT1, B9D1, B9D2, BAG3, BANF1, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCKDHA, BCKDHB, BCKDK, BCL11A, BCOR, BCS1L, BGN, BHLHA9, BICD2, BIN1, BLM, BLNK, BLTP1, BMP1, BMP2, BMP4, BMPER, BMPR1B, BOLA3, BPNT2, BPTF, BRAF, BRAT1, BRCA1, BRCA2, BRD4, BRF1, BRIP1, BRPF1, BRWD3, BSCL2, BSND, BTD, BTK, BUB1B, C12orf57, C19orf12, C1QBP, C2CD3, C2orf69, CA2, CA5A, CA8, CABP2, CACNA1A, CACNA1C, CACNA1D, CACNA1E, CACNA1G, CAD, CAMK2A, CAMK2B, CAMTA1, CANT1, CAPN3, CARD11, CARMIL2, CARS2, CASK, CASP10, CASQ2, CASR, CAV1, CAVIN1, CBL, CBS, CC2D1A, CC2D2A, CCBE1, CCDC22, CCDC39, CCDC40, CCDC47, CCDC78, CCDC8, CCDC88A, CCDC88C, CCN6, CCND2, CCNK, CCNO, CCNQ, CCT5, CD19, CD247, CD27, CD2AP, CD3D, CD3E, CD3G, CD40, CD40LG, CD55, CD70, CD79A, CD79B, CD96, CDC14A, CDC42, CDC42BPB, CDC45, CDH1, CDH11, CDH2, CDH23, CDH3, CDIN1, CDK10, CDK13, CDK19, CDK5RAP2, CDKL5, CDKN1C, CDON, CDSN, CDT1, CENPF, CEP104, CEP120, CEP135, CEP152, CEP164, CEP290, CEP41, CEP55, CEP57, CEP63, CEP78, CEP83, CERS1, CERS3, CERT1, CFAP298, CFAP300, CFAP410, CFAP418, CFL2, CFP, CFTR, CHAMP1, CHAT, CHD2, CHD3, CHD4, CHD7, CHD8, CHKB, CHM, CHMP1A, CHRDL1, CHRNA1, CHRNA2, CHRNA4, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CHST3, CHSY1, CHUK, CIB2, CIITA, CILK1, CISD2, CIT, CKAP2L, CLCN1, CLCN2, CLCN4, CLCN5, CLCN7, CLCNKA, CLCNKB, CLDN1, CLDN10, CLDN14, CLDN16, CLDN19, CLIC5, CLMP, CLN3, CLN5, CLN6, CLN8, CLP1, CLPB, CLPP, CLRN1, CLTC, CNKSR2, CNNM2, CNOT1, CNOT3, CNPY3, CNTNAP1, CNTNAP2, COA6, COA8, COASY, COCH, COG1, COG2, COG4, COG5, COG6, COG7, COG8, COL10A1, COL11A1, COL11A2, COL13A1, COL17A1, COL18A1, COL1A1, COL1A2, COL27A1, COL2A1, COL3A1, COL4A1, COL4A2, COL4A3, COL4A4, COL4A5, COL5A1, COL6A1, COL6A2, COL6A3, COL7A1, COL9A1, COL9A2, COL9A3, COLEC10, COLEC11, COLQ, COMP, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ8B, COQ9, CORO1A, COX10, COX14, COX15, COX20, COX6A2, COX6B1, COX7B, COX8A, CPAP, CPLANE1, CPLX1, CPS1, CPT1A, CPT2, CRADD, CRB1, CRB2, CRBN, CREB3L1, CREBBP, CRELD1, CRIPT, CRKL, CRLF1, CRPPA, CRTAP, CRX, CRYAA, CRYAB, CSF1R, CSF3R, CSNK1E, CSNK2A1, CSNK2B, CSPP1, CSTA, CSTB, CTC1, CTCF, CTDP1, CTNNA2, CTNNB1, CTNND1, CTNS, CTPS1, CTSA, CTSD, CTSK, CTU2, CUL3, CUL4B, CUL7, CWC27, CWF19L1, CXCR4, CYB5R3, CYBA, CYBB, CYC1, CYP11A1, CYP11B1, CYP11B2, CYP17A1, CYP24A1, CYP27A1, CYP27B1, CYP2R1, CYP2U1, CYP4F22, CYP7B1, D2HGDH, DAG1, DARS1, DARS2, DBT, DCAF17, DCC, DCDC2, DCHS1, DCLRE1C, DCX, DDB2, DDC, DDHD1, DDHD2, DDR2, DDX11, DDX3X, DDX59, DDX6, DEAF1, DEGS1, DENND5A, DEPDC5, DGAT1, DGKE, DGUOK, DHCR24, DHCR7, DHDDS, DHH, DHODH, DHTKD1, DHX16, DHX30, DHX37, DIAPH1, DIS3L2, DKC1, DLAT, DLD, DLG3, DLG4, DLL1, DLL3, DLL4, DLX5, DMD, DMP1, DMXL2, DNA2, DNAAF11, DNAAF19, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH11, DNAH5, DNAH9, DNAJC12, DNAJC19, DNAJC21, DNAJC3, DNAJC5, DNAJC6, DNM1, DNM1L, DNM2, DNMT3A, DNMT3B, DOCK2, DOCK6, DOCK7, DOCK8, DOK7, DOLK, DONSON, DPAGT1, DPF2, DPH1, DPM1, DPM2, DPYD, DRC1, DRC2, DRC4, DSE, DSG1, DSP, DST, DSTYK, DTNA, DUOX2, DUOXA2, DVL1, DVL3, DYM, DYNC1H1, DYNC2H1, DYNC2I1, DYNC2I2, DYNC2LI1, DYRK1A, DYSF, EARS2, EBF3, EBP, ECEL1, ECHS1, EDA, EDAR, EDARADD, EDN3, EDNRA, EDNRB, EED, EEF1A2, EFEMP2, EFL1, EFNB1, EFTUD2, EGR2, EHMT1, EIF2AK3, EIF2AK4, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, EIF4A3, ELAC2, ELANE, ELMO2, ELMOD3, ELN, ELOVL4, ELP1, ELP2, EMC1, EMC10, EMD, EMG1, EML1, EMX2, ENG, ENPP1, ENTPD1, EOGT, EP300, EPB42, EPCAM, EPG5, EPHB4, EPM2A, EPRS1, EPS8, EPS8L2, ERAL1, ERBB3, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC6L2, ERCC8, ERF, ERLIN1, ERLIN2, ESCO2, ESPN, ESRRB, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOC3L2, EXOSC3, EXOSC8, EXOSC9, EXPH5, EXT1, EXT2, EXTL3, EYA1, EZH2, F10, F13A1, F2, F7, F8 (intronic inversions not covered), *F9, FA2H, FADD, FAH, FAM111A, FAM149B1, FAM161A, FAM20A, FAM20C, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAR1, FARS2, FAS, FASLG, FASTKD2, FAT4, FBLN5, FBN1, FBN2, FBP1, FBXL4, FBXO11, FBXO7, FBXW11, FBXW4, FCSK, FERMT3, FEZF1, FGA, FGB, FGD1, FGD4, FGF10, FGF12, FGF3, FGF8, FGF9, FGFR1, FGFR2, FGFR3, FGG, FH, FHL1, FIG4, FITM2, FKBP10, FKBP14, FKRP, FKTN, FLAD1, FLNA, FLNB, FLT4, FLVCR1, FLVCR2, FN1, FOLR1, FOXC1, FOXC2, FOXE1, FOXE3, FOXF1, FOXG1, FOXL2, FOXN1, FOXP1, FOXP3, FOXRED1, FRAS1, FREM1, FREM2, FRMPD4, FRRS1L, FSHB, FTCD, FTL, FTO, FTSJ1, FUCA1, FUT8, FXN, FZD2, G6PC1, G6PC3, GAA, GABBR2, GABRA1, GABRB1, GABRB2, GABRB3, GABRG2, GAD1, GALC, GALE, GALK1, GALNS, GALT, GAMT, GAN, GATA1, GATA2, GATA3, GATA4, GATA6, GATAD2B, GATM, GBA1, GBA2, GBE1, GCDH, GCH1, GCK, GCSH, GDAP1, GDF1, GDF3, GDF5, GDF6, GDI1, GEMIN4, GFAP, GFER, GFM1, GFM2, GFPT1, GHR, GIPC3, GJA1, GJA8, GJB2, GJB3, GJB6, GJC2, GK, GLA, GLB1, GLDC, GLDN, GLE1, GLI2, GLI3, GLIS2, GLIS3, GLRX5, GLS, GLUD1, GLUL, GLYCTK, GM2A, GMNN, GMPPA, GMPPB, GNA11, GNAI3, GNAO1, GNAQ, GNAS, GNB1, GNB5, GNPAT, GNPTAB, GNPTG, GNRH1, GNRHR, GNS, GOLGA2, GORAB, GOSR2, GOT2, GPAA1, GPC3, GPC6, GPHN, GPSM2, GPT2, GPX4, GREB1L, GRHL2, GRHL3, GRHPR, GRIA2, GRIA3, GRID2, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRIP1, GRM1, GRM7, GRXCR1, GSS, GTF2H5, GTPBP3, GUCY1A1, GUCY2C, GUF1, GUSB, GYS1, GYS2, GZF1, H1-4, HACD1, HACE1, HADH, HADHA, HADHB, HAMP, HARS1, HARS2, HAX1, HBB, HCCS, HCFC1, HCN1, HDAC4, HDAC8, HECW2, HEPACAM, HERC1, HERC2, HES7, HESX1, HEXA, HEXB, HGF, HGSNAT, HIBCH, HIKESHI, HINT1, HIVEP2, HJV, HK1, HLCS, HMGCL, HMGCS2, HMX1, HNF1B, HNF4A, HNRNPH2, HNRNPK, HNRNPU, HOGA1, HOXA1, HOXA13, HOXC13, HOXD13, HPD, HPDL, HPGD, HPRT1, HPS1, HPSE2, HRAS, HSD11B2, HSD17B10, HSD17B3, HSD17B4, HSD3B2, HSD3B7, HSPA9, HSPD1, HSPG2, HTRA2, HUWE1, HYAL1, HYCC1, HYLS1, IARS1, IARS2, IBA57, ICOS, IDS, IDUA, IER3IP1, IFIH1, IFITM5, IFNGR1, IFNGR2, IFT122, IFT140, IFT172, IFT27, IFT43, IFT52, IFT56, IFT74, IFT80, IFT81, IGBP1, IGF1, IGF1R, IGF2, IGFBP7, IGHMBP2, IGSF1, IHH, IKBKB, IL10RA, IL11RA, IL12RB1, IL1RAPL1, IL1RN, IL21R, IL2RA, IL2RB, IL2RG, IL7R, ILDR1, INPP5E, INPP5K, INPPL1, INS, INSR, INTS1, INTU, INVS, IPO8, IQCB1, IQSEC1, IQSEC2, IRAK4, IRF2BPL, IRF6, IRF8, IRX5, ISCA1, ISCA2, ITCH, ITGA3, ITGA6, ITGA7, ITGA8, ITGB4, ITK, ITPA, ITPR1, IVD, JAG1, JAGN1, JAK3, JAM2, JAM3, JUP, KANK1, KANSL1, KARS1, KAT5, KAT6A, KAT6B, KAT8, KATNB1, KATNIP, KBTBD13, KCNA2, KCNB1, KCNC1, KCNC3, KCNE1, KCNH1, KCNJ1, KCNJ10, KCNJ11, KCNJ6, KCNMA1, KCNQ1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCTD1, KCTD7, KDELR2, KDM5B, KDM5C, KDM6A, KIAA0586, KIAA0753, KIDINS220, KIF11, KIF14, KIF1A, KIF1C, KIF22, KIF2A, KIF4A, KIF5A, KIF5C, KIF7, KIFBP, KIRREL3, KISS1R, KLF1, KLHL15, KLHL40, KLHL41, KLHL7, KMT2A, KMT2B, KMT2C, KMT2D, KMT2E, KMT5B, KNL1, KPTN, KRAS, KRIT1, KRT10, KRT14, KRT18, KRT5, KRT8, KY, L1CAM, L2HGDH, LAGE3, LAMA1, LAMA2, LAMA3, LAMB1, LAMB2, LAMB3, LAMC2, LAMC3, LAMP2, LARGE1, LARP7, LARS2, LAS1L, LAT, LBR, LDB3, LDHA, LDLR, LEMD3, LETM1, LFNG, LGI4, LHB, LHCGR, LHFPL5, LHX3, LHX4, LIAS, LIFR, LIG4, LIMS2, LINS1, LIPA, LIPT1, LMBR1, LMBRD1, LMNA, LMOD3, LMX1B, LNPK, LONP1, LOXHD1, LPIN1, LPIN2, LPL, LRBA, LRP2, LRP4, LRP5, LRPPRC, LRRC56, LRTOMT, LTBP2, LTBP3, LTBP4, LYRM4, LYRM7, LYST, LZTFL1, LZTR1, MAB21L2, MACF1, MAF, MAFB, MAG, MAGEL2, MAGI2, MAGT1, MALT1, MAMLD1, MAN1B1, MAN2B1, MANBA, MAOA, MAP1B, MAP2K1, MAP2K2, MAP3K1, MAP3K20, MAP3K7, MAPK8IP3, MAPKBP1, MAPRE2, MARS1, MARVELD2, MASP1, MAT1A, MATN3, MBD5, MBOAT7, MBTPS2, MC2R, MCCC1, MCCC2, MCEE, MCM4, MCOLN1, MCPH1, MDH2, MECOM, MECP2, MECR, MED12, MED12L, MED13, MED13L, MED17, MED23, MED25, MEF2C, MEGF10, MEGF8, MEIS2, MEOX1, MESD, MESP2, MET, METTL23, METTL5, MFN2, MFRP, MFSD2A, MFSD8, MGAT2, MGME1, MGP, MICOS13, MICU1, MID1, MIPEP, MIR17HG, MITF, MKKS, MKS1, MLC1, MLPH, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMP13, MMP2, MMP21, MMUT, MN1, MNX1, MOCS1,*

Gene list for counselling (continued)

MOCS2, MOGS, MORC2, MPDU1, MPDZ, MPI, MPL, MPLKIP, MPV17, MPZ, MPZL2, MRE11, MRPL3, MRPL44, MRPS14, MRPS16, MRPS2, MRPS22, MRPS34, MSL3, MSMO1, MSN, MSRB3, MSTO1, MSX1, MSX2, MT-ATP6, MTFMT, MTHFD1, MTHFR, MTM1, MTMR2, MTO1, MTOR, MTR, MTRFR, MTRR, MTTP, MUSK, MUTYH, MUTYH,TOE1, MVK, MYBPC1, MYBPC3, MYCN, MYD88, MYH11, MYH11,NDE1, MYH3, MYH6, MYH7, MYH8, MYH9, MYL3, MYMK, MYO15A, MYO18B, MYO3A, MYO5A, MYO5B, MYO6, MYO7A, MYO9A, MYOD1, MYPN, MYRF, MYSM1, MYT1L, NAA10, NAA15, NACC1, NADSYN1, NAGA, NAGLU, NAGS, NALCN, NANS, NARS1, NARS2, NAXD, NAXE, NBAS, NBEA, NBN, NCAPD3, NCF1, NCF2, NCF4, NCKAP1L, NDE1, NDN, NDP, NDRG1, NDST1, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA6, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF8, NDUFB3, NDUFB8, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NEB, NECAP1, NECTIN1, NECTIN4, NEDD4L, NEK1, NEK8, NEK9, NEMF, NEU1, NEUROG3, NEXMIF, NF1, NFASC, NFIA, NFIB, NFIX, NFKBIA, NFU1, NGF, NGLY1, NHEJ1, NHLRC1, NHP2, NHS, NIPAL4, NIPBL, NKAP, NKX2-1, NKX2-5, NKX2-6, NKX3-2, NKX6-2, NLRC4, NLRP3, NMNAT1, NNT, NODAL, NOG, NONO, NOP10, NOTCH1, NOTCH2, NOTCH3, NOVA2, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NPR2, NR0B1, NR1H4, NR2F1, NR2F2, NR3C2, NR5A1, NRAS, NRROS, NRXN1, NRXN3, NSD1, NSDHL, NSMCE2, NSMCE3, NSUN2, NT5C2, NT5C3A, NTNG2, NTRK1, NTRK2, NUBPL, NUDT2, NUP107, NUP133, NUP188, NUP62, NUP88, NUP93, NYX, OBSL1, OCLN, OCRL, ODAD1, ODAD2, ODAD3, OFD1, OGDH, OPA1, OPA3, OPHN1, ORAI1, ORC1, ORC4, ORC6, OSGEP, OSTM1, OTC, OTOA, OTOF, OTOG, OTOGL, OTUD5, OTUD6B, OTULIN, OTX2, OXCT1, OXR1, P3H1, P4HB, PACS1, PACS2, PAFAH1B1, PAH, PAK3, PALB2, PAM16, PANK2, PAPSS2, PARN, PARS2, PAX2, PAX3, PAX6, PAX8, PC, PCARE, PCBD1, PCCA, PCCB, PCDH12, PCDH15, PCDH19, PCGF2, PCK1, PCNT, PCSK1, PCYT1A, PCYT2, PDCD10, PDE10A, PDE4D, PDE6D, PDE6G, PDGFRB, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PDX1, PDZD7, PEPD, PERCC1, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PGAP1, PGAP2, PGAP3, PGK1, PGM1, PGM3, PHACTR1, PHEX, PHF21A, PHF6, PHF8, PHGDH, PHIP, PHKG2, PHOX2B, PHYH, PI4KA, PIBF1, PIEZO1, PIEZO2, PIGA, PIGB, PIGG, PIGK, PIGL, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGT, PIGV, PIGY, PIK3CA, PIK3CD, PIK3R1, PIK3R2, PIP5K1C, PISD, PITX1, PITX2, PITX3, PJVK, PKD1L1, PKHD1, PKLR, PLA2G6, PLAA, PLCB1, PLCB4, PLCE1, PLCG2, PLEC, PLEKHG2, PLEKHG5, PLG, PLK4, PLOD1, PLOD2, PLOD3, PLP1, PLPBP, PLS3, PLVAP, PMM2, PMP22, PMPCA, PMPCB, PNKP, PNP, PNPLA1, PNPLA6, PNPLA8, PNPO, PNPT1, POC1A, POC1B, POGZ, POLA1, POLD1, POLE, POLG, POLG2, POLR1A, POLR1B, POLR1C, POLR1D, POLR3A, POLR3B, POMC, POMGNT1, POMGNT2, POMK, POMP, POMT1, POMT2, POP1, POR, PORCN, POU1F1, POU3F4, PPA2, PPIB, PPIP5K2, PPM1D, PPP1CB, PPP1R12A, PPP1R15B, PPP1R21, PPP2CA, PPP2R1A, PPP2R5D, PPP3CA, PPT1, PQBP1, PRDM12, PRDM15, PRDM16, PRDM5, PRDX1, PREPL, PRF1, PRG4, PRICKLE1, PRKAG2, PRKAR1A, PRKCD, PRKD1, PRKDC, PRKRA, PRMT7, PROC, PRODH, PROP1, PROS1, PRPS1, PRRT2, PRRX1, PRSS12, PRSS56, PRUNE1, PRX, PSAP, PSAT1, PSMB8, PSMD12, PSPH, PTCH1, PTCHD1, PTDSS1, PTEN, PTF1A, PTH1R, PTHLH, PTPN11, PTPN14, PTPN23, PTPRC, PTPRQ, PTRH2, PTS, PUF60, PURA, PUS1, PUS7, PXDN, PYCR1, PYCR2, PYGL, PYGM, PYROXD1, QARS1, QDPR, QRICH1, RAB11A, RAB11B, RAB18, RAB23, RAB27A, RAB33B, RAB39B, RAB3GAP1, RAB3GAP2, RAC1, RAC2, RAD21, RAD50, RAD51C, RAF1, RAG1, RAG2, RAI1, RALGAPA1, RAPSN, RARB, RARS1, RARS2, RASA1, RAX, RB1, RBBP8, RBCK1, RBM10, RBM8A, RBPJ, RDH11, RDX, RECQL4, RELN, REN, RERE, RET, RETREG1, RFT1, RFX5, RFX6, RFXANK, RFXAP, RHOA, RHOBTB2, RIC1, RIMS2, RIN2, RINT1, RIPK1, RIPK4, RIPOR2, RIT1, RLIM, RMND1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF113A, RNF13, RNF135, RNF168, RNU2-2, RNU4-2, RNU4ATAC, RNU5B-1 ROBO3, ROGDI, ROR1, ROR2, RORA, RPE65, RPGRIP1, RPGRIP1L, RPIA, RPL10, RPL11, RPL13, RPL26, RPL5, RPS19, RPS26, RPS28, RPS29, RPS6KA3, RPS7, RRM2B, RSPH1, RSPH3, RSPO2, RSPO4, RSPRY1, RTEL1, RTN4IP1, RTTN, RUNX2, RUSC2, RXYLT1, RYR1, S1PR2, SACS, SALL1, SALL4, SAMD9, SAMD9L, SAMHD1, SAR1B, SARS2, SASS6, SATB2, SBDS, SBF1, SBF2, SC5D, SCAMP5, SCAPER, SCARB2, SCARF2, SCN11A, SCN1A, SCN1B, SCN2A, SCN3A, SCN4A, SCN8A, SCN9A, SCNN1A, SCNN1B, SCNN1G, SCO1, SCO2, SCYL1, SCYL2, SDCCAG8, SDHA, SDHAF1, SDHD, SEC23A, SEC23B, SEC24D, SELENOI, SELENON, SEPSECS, SERAC1, SERPINB6, SERPINF1, SERPINH1, SET, SETBP1, SETD1A, SETD1B, SETD2, SETD5, SETX, SF3B4, SFTPB, SFTPC, SFXN4, SGCA, SGCB, SGCD, SGCE, SGCG, SGO1, SGPL1, SGSH, SH2D1A, SH3PXD2B, SH3TC2, SHANK1, SHANK2, SHH, SHOC2, SHOX, SIK1, SIL1, SIM1, SIN3A, SIX1, SIX3, SIX5, SKI, SKIC2, SKIC3, SLC10A7, SLC12A1, SLC12A3, SLC12A5, SLC12A6, SLC13A5, SLC16A1, SLC16A2, SLC17A5, SLC18A3, SLC19A2, SLC19A3, SLC1A2, SLC1A4, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A24, SLC25A26, SLC25A3, SLC25A38, SLC25A4, SLC25A42, SLC25A46, SLC26A2, SLC26A3, SLC26A4, SLC26A5, SLC26A7, SLC27A4, SLC29A3, SLC2A1, SLC2A10, SLC2A2, SLC30A10, SLC33A1, SLC34A1, 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TAF1, TAF13, TAF2, TAF6, TAFAZZIN, TALDO1, TANGO2, TAOK1, TAP1, TAPT1, TARS2, TASP1, TAT, TBC1D20, TBC1D23, TBC1D24, TBC1D8B, TBCD, TBCE, TBCK, TBL1XR1, TBR1, TBX1, TBX15, TBX18, TBX19, TBX20, TBX22, TBX3, TBX4, TBX5, TBXAS1, TCAP, TCF12, TCF20, TCF4, TCIRG1, TCN2, TCOF1, TCTN1, TCTN2, TCTN3, TDP2, TECPR2, TECTA, TEK, TELO2, TENM3, TENT5A, TERT, TF, TFAP2A, TFAP2B, TFR2, TGDS, TGFB1, TGFB2, TGFB3, TGFBR1, TGFBR2, TGIF1, TGM1, TH, THAP1, THOC2, THOC6, THRA, TIMM50, TIMM8A, TIMMDC1, TINF2, TJP2, TK2, TKT, TLK2, TMC1, TMCO1, TMEM107, TMEM126A, TMEM126B, TMEM132E, TMEM138, TMEM165, TMEM216, TMEM231, TMEM237, TMEM260, TMEM38B, TMEM43, TMEM67, TMEM70, TMEM94, TMIE, TMPRSS3, TMPRSS6, TMTC3, TMX2, TNFRSF11A, TNFRSF11B, TNFRSF13B, TNFSF11, TNNT1, TOE1, TOP3A, TP53, TP53RK, TP63, TPI1, TPK1, TPM2, TPM3, TPO, TPP1, TPRKB, TPRN, TRAF3IP1, TRAIP, TRAK1, TRAPPC11, TRAPPC12, TRAPPC2, TRAPPC4, TRAPPC9, TRDN, TREX1, TRIM2, TRIM32, TRIM37, TRIO, TRIOBP, TRIP11, TRIP12, TRIP13, TRIP4, TRIT1, TRMT1, TRMT10A, TRMT10C, TRMT5, TRMU, TRNT1, TRPM1, TRPM6, TRPS1, TRPV3, TRPV4, TRPV6, TRRAP, TSC1, TSC2, TSEN15, TSEN2, TSEN54, TSFM, TSHB, TSHR, TSPAN7, TSPEAR, TSPYL1, TTC19, TTC21B, TTC7A, TTC8, TTI2, TTN, TTPA, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP2, TUBGCP4, TUBGCP6, TUFM, TUSC3, TWIST1, TWIST2, TWNK, TXN2, TXNDC15, TXNL4A, TYK2, TYMP, TYR, TYRP1, UBA1, UBA5, UBE2A, UBE2T, UBE3A, UBE3B, UBR1, UBTF, UCHL1, UFC1, UFM1, UGDH, UGP2, UGT1A1, UMPS, UNC13D, UNC80, UPF3B, UPK3A, UQCC2, UQCRB, UQCRC2, UQCRFS1, UQCRQ, UROC1, UROS, USB1, USH1C, USH1G, USH2A, USP18, USP53, USP7, USP9X, UVSSA, VAC14, VAMP1, VAMP2, VANGL1, VARS1, VARS2, VDR, VIPAS39, VLDLR, VMA12, VMA21, VMA22, VPS11, VPS13B, VPS13D, VPS33A, VPS33B, VPS37A, VPS41, VPS45, VPS51, VPS53, VRK1, VSX2, WAC, WARS2, WAS, WASF1, WASHC5, WBP2, WDFY3, WDPCP, WDR11, WDR19, WDR26, WDR35, WDR37, WDR4, WDR45, WDR45B, WDR62, WDR73, WDR81, WFS1, WHRN, WNK1, WNT1, WNT10A, WNT10B, WNT2B, WNT3, WNT4, WNT5A, WNT7A, WRAP53, WRN, WT1, WWOX, XIAP, XPA, XPC, XRCC2, XRCC4, XYLT1, XYLT2, YAP1, YARS2, YIF1B, YWHAG, YY1, ZAP70, ZBTB18, ZBTB20, ZBTB24, ZC3H14, ZC4H2, ZDHHC9, ZEB2, ZFPM2, ZFYVE26, ZIC1, ZIC2, ZIC3, ZMIZ1, ZMPSTE24, ZMYND10, ZMYND11, ZNF335, ZNF423, ZNF462, ZNF469, ZNF711, ZNHIT3

Order Form
Prenatal ExomeXtra® – Inconspicuous Ultrasound

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:

Additional analyses (additional fees may apply)

❑ **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

Declaration of consent

By signing this form, I declare that I have received comprehensive information regarding the genetic background related to the requested analysis, 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 physican 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.com/privacy-policy.

Please Note

All genes, including the complete mtDNA are sequenced when exome diagnostics is performed. 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. 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".

Patient
(Block letters)

Physican
(Block letters)

X _____
Patient
(Date, Signature)

X _____
Physican
(Date, Signature)

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

Order Form
Prenatal ExomeXtra® – Inconspicuous Ultrasound

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:

Additional analyses (additional fees may apply)

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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

Declaration of consent

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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 physican 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.com/privacy-policy.

Please Note

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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 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".

Patient
(Block letters)

Physican
(Block letters)

X _____
Patient
(Date, Signature)

X _____
Physican
(Date, Signature)

Physican's stamp / Barcode

DAkkS Deutsche Akkreditierungsstelle D-ML-13206-01-00

CAP ACCREDITED COLLEGE of AMERICAN PATHOLOGISTS
CLIA CERTIFIED ID: 99D2130225

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