Order Form
Family Planning Panel

General Information Patient 1

Patient 1

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. 1-2 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 (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 agree to receiving a joint report with my partner, as potential parents.

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. Comparative exome analysis is performed assuming the correct family relations and using the data from the family members. The report will include the reportable variants from all members that are included in the scope of the chosen 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 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 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 https://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

Patient 1
(Block letters)

Physican
(Block letters)

X _____
Patient 1
(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 15189:2014, the College of American Pathologists (CAP) and CLIA.

Order Form
Family Planning Panel

Indication Patient 1

Consanguinity: ❑ No ❑ Yes

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

Inquiry

☑ **Family Planning Panel (1943 Genes, FPP) including FMR1 Repeat and SMN1 MLPA**

Both persons seeking advice will receive a joint report listing variants in genes that could lead to severe diseases in their offspring, if disease onset is expected to occur during childhood. Analysis of the FMR1 repeat is performed only in female probands.

Additional analysis (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 https://www.cegat.com/acmg-genes/

❑ **Pharmacogenetics (PGX) (22 genes)**

ABCG2, CACNA1S, CYP2B6, CYP2C19, CYP2C9, CYP2D6, CYP3A4, CYP3A5, CYP4F2, DPYD, G6PD, HLA-A, HLA-B, IFNL3, MT-RNR1, NUDT15, POR, RYR1, SLCO1B1, TPMT, UGT1A1, VKORC1

I would like to receive an additional report analyzing known variants that are involved in the metabolism of pharmaceutical products.

Order Form
Family Planning Panel

General Information Patient 2

Patient 2

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. 1-2 µg DNA, concentr. ≥ 50 ng/µl) DNA-No.: _____

❑ Other specimen _____

External ID: _____

Date of sample collection: _____

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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:

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Country:

Email:

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I agree to receiving a joint report with my partner, as potential parents.

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Email: _____

Physician's stamp / Barcode

Patient 2
(Block letters)

Physican
(Block letters)

X _____
Patient 2
(Date, Signature)

X _____
Physican
(Date, Signature)

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

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CLIA CERTIFIED ID: 99D2130225

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Order Form
Family Planning Panel

Indication Patient 2

Consanguinity: ❑ No ❑ Yes

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

Inquiry

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Order Form
Family Planning Panel

Inquiry

Gene list (1943 Genes, FPP)

AAAS, AARS1, AARS2, ABAT, ABCA12, ABCA3, ABCB11, ABCB4, ABCB7, ABCC6, ABCC8, ABCC9, ABCD1, ABCD4, ABHD12, ABHD5, ACACA, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAN, ACAT1, ACD, ACE, ACO2, ACOX1, ACOX2, ACP5, ACSL4, ACTA1, ACTL6B, ACY1, ADA, ADA2, ADAM17, ADAM22, ADAMTS13, ADAMTS19, ADAMTS2, ADAMTSL2, ADAR, ADARB1, ADAT3, ADCY1, ADCY5, ADCY6, ADGRG1, ADGRG6, ADGRV1, ADK, ADPRS, ADSL, AFF2, AFG3L2, AGA, AGK, AGL, AGPAT2, AGPS, AGRN, AGT, AGTPBP1, AGTR1, AGXT, AHCY, AHI1, AIFM1, AIMP1, AIMP2, AIPL1, AIRE, AK2, AKR1D1, 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, ANK3, ANKLE2, ANKS6, ANO10, ANO5, ANOS1, ANTXR1, ANTXR2, AP1B1, AP1S1, AP1S2, AP3B1, AP3B2, AP3D1, AP4B1, AP4E1, AP4M1, AP4S1, APC2, APTX, AQP2, AR, ARFGEF2, ARG1, ARHGDIA, ARHGEF9, ARL13B, ARL3, ARL6, ARL6IP1, ARMC9, ARNT2, ARPC1B, ARSA, ARSB, ARSL, ARV1, ARX, ASAH1, ASCC1, ASL, ASNS, ASPA, ASPH, ASPM, ASS1, ATAD1, ATAD3A, ATCAY, ATIC, ATM, ATOH7, ATP13A2, ATP1A2, ATP2B3, ATP5F1D, ATP5MK, ATP6AP1, ATP6AP2, ATP6V0A2, ATP6V0A4, ATP6V1A, ATP6V1B1, ATP6V1E1, ATP7A, ATP7B, ATP8A2, ATP8B1, ATPAF2, ATR, ATRX, AUH, AVIL, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT1, B4GALT7, B4GAT1, B9D1, B9D2, BANF1, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCKDHA, BCKDHB, BCKDK, BCOR, BCS1L, BGN, BHLHA9, BIN1, BLM, BLNK, BLTP1, BMP1, BMP2, BMPER, BMPR1B, BOLA3, BPNT2, BRAT1, BRCA1, BRCA2, BRF1, BRWD3, BSCL2, BSND, BTD, BTK, BUB1B, C12orf57, C19orf12, C1QBP, C2CD3, C2orf69, CA2, CA5A, CA8, CABP2, CACNA1D, CAD, CAMK2A, CANT1, CAPN3, CARD11, CARMIL2, CARS2, CASK, CASQ2, CASR, CAV1, CAVIN1, CBS, CC2D1A, CC2D2A, CCBE1, CCDC103, CCDC115, CCDC22, CCDC39, CCDC40, CCDC47, CCDC65, CCDC8, CCDC88A, CCDC88C, CCN6, CCNO, CCNQ, CCT5, CD19, CD247, CD27, CD3D, CD3E, CD3G, CD40, CD40LG, CD55, CD70, CD79A, CD79B, CDC14A, CDC45, CDH11, CDH2, CDH23, CDH3, CDIN1, CDK10, CDK5RAP2, CDKL5, CDSN, CDT1, CENPF, CENPJ, CEP104, CEP120, CEP135, CEP152, CEP164, CEP290, CEP41, CEP55, CEP57, CEP63, CEP78, CEP83, CERS1, CERS3, CFAP298, CFAP300, CFAP410, CFAP418, CFL2, CFP, CFTR, CHAT, CHKB, CHM, CHMP1A, CHRDL1, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CHST3, CHSY1, CHUK, CIB2, CIITA, CILK1, CISD2, CIT, CKAP2L, CLCN1, CLCN2, CLCN4, CLCN5, CLCN7, CLCNKB, CLDN1, CLDN10, CLDN14, CLDN16, CLDN19, CLIC5, CLMP, CLN3, CLN5, CLN6, CLN8, CLP1, CLPB, CLPP, CLRN1, CNKSR2, CNNM2, CNPY3, CNTNAP1, CNTNAP2, COA6, COA8, COASY, COCH, COG1, COG2, COG4, COG5, COG6, COG7, COL11A1, COL11A2, COL13A1, COL17A1, COL18A1, COL1A2, COL27A1, COL3A1, COL4A3, COL4A4, COL4A5, COL6A1, COL6A2, COL6A3, COL7A1, COL9A2, COLEC10, COLEC11, COLQ, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ8B, COQ9, CORO1A, COX10, COX14, COX15, COX20, COX6A2, COX6B1, COX7B, COX8A, CPLANE1, CPLX1, CPS1, CPT1A, CPT2, CRADD, CRB1, CRB2, CRBN, CREB3L1, CRIPT, CRLF1, CRPPA, CRTAP, CRYAA, CRYAB, CSF1R, CSF2RB, CSF3R, CSPP1, CSTA, CSTB, CTC1, CTDP1, CTNNA2, CTNS, CTPS1, CTSA, CTSD, CTSK, CTU2, CUL4B, CUL7, CWC27, CWF19L1, CYB5R3, CYBA, CYBB, CYC1, CYP11A1, CYP11B1, CYP11B2, CYP17A1, CYP24A1, CYP27A1, CYP27B1, CYP2R1, CYP2U1, CYP4F22, CYP7B1, D2HGDH, DAG1, DARS1, DARS2, DBT, DCAF17, DCDC2, DCHS1, DCLRE1C, DCX, DDB2, DDC, DDHD1, DDHD2, DDR2, DDX11, DDX3X, DDX59, DEAF1, DEGS1, DENND5A, DGAT1, DGKE, DGUOK, DHCR24, DHCR7, DHDDS, DHH, DHODH, DHTKD1, DHX37, DIAPH1, DIS3L2, DKC1, DLAT, DLD, DLG3, DLL3, DLX5, DMD, DMP1, DMXL2, DNA2, DNAAF11, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH11, DNAH5, DNAH9, DNAJC12, DNAJC19, DNAJC21, DNAJC3, DNAJC6, DNM1L, DNM2, DNMT3B, DOCK2, DOCK6, DOCK7, DOCK8, DOK7, DOLK, DONSON, DPAGT1, DPH1, DPM1, DPM2, DPYD, DRC1, DSE, DSG1, DSP, DST, DSTYK, DUOX2, DUOXA2, DYM, DYNC2H1, DYNC2I1, DYNC2I2, DYNC2LI1, DYSF, EARS2, EBP, ECEL1, ECHS1, EDA, EDAR, EDARADD, EDN3, EDNRB, EFEMP2, EFL1, EFNB1, EGR2, EIF2AK3, EIF2AK4, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, EIF4A3, ELAC2, ELMO2, ELMOD3, ELOVL4, ELP1, ELP2, EMC1, EMC10, EMD, EMG1, EML1, ENPP1, ENTPD1, EOGT, EPCAM, EPG5, EPM2A, EPRS1, EPS8, EPS8L2, ERAL1, ERBB3, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC6L2, ERCC8, ERLIN1, ERLIN2, ESCO2, ESPN, ESRRB, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOC3L2, EXOSC3, EXOSC8, EXOSC9, EXPH5, EXT2, EXTL3, F10, F13A1, F2, F7, F8 (intronic inversions not covered), F9, FA2H, FADD, FAH, FAM126A, FAM149B1, FAM20A, FAM20C, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAR1, FARS2, FASTKD2, FAT4, FBLN5, FBP1, FBXL4, FBXO7, FCSK, FERMT3, FEZF1, FGA, FGB, FGD1, FGD4, FGF3, FGFR3, FGG, FH, FHL1, FIG4, FITM2, FKBP10, FKBP14, FKRP, FKTN, FLAD1, FLNA, FLNB, FLVCR1, FLVCR2, FOLR1, FOXE1, FOXE3, FOXL2, FOXN1, FOXP3, FOXRED1, FRAS1, FREM1, FREM2, FRMPD4, FRRS1L, FSHB, FTCD, FTL, FTO, FTSJ1, FUCA1, FUT8, FXN, G6PC1, G6PC3, GAA, GAD1, GALC, GALE, GALK1, GALNS, GALT, GAMT, GAN, GAS8, GATA1, GATM, GBA1, GBA2, GBE1, GCDH, GCH1, GCK, GCSH, GDAP1, GDF1, GDF5, GDF6, GDI1, GEMIN4, GFER, GFM1, GFM2, GFPT1, GHR, GIPC3, GJA1, GJB2, GJB3, GJB6, GJC2, GK, GLA, GLB1, GLDC, GLDN, GLE1, GLIS3, GLRX5, GLS, GLUL, GLYCTK, GM2A, GMPPA, GMPPB, GNB5, GNPAT, GNPTAB, GNPTG, GNRH1, GNRHR, GNS, GOLGA2, GORAB, GOSR2, GOT2, GPAA1, GPC3, GPC6, GPHN, GPSM2, GPT2, GPX4, GRHL2, GRHPR, GRIA3, GRID2, GRIK2, GRIN1, GRIP1, GRM1, GRM7, GRXCR1, GSS, GTF2H5, GTPBP3, GUCY1A1, GUCY2C, GUF1, GUSB, GYS1, GYS2, GZF1, HACD1, HACE1, HADH, HADHA, HADHB, HAMP, HARS1, HARS2, HAX1, HBB, HCCS, HCFC1, HDAC8, HEPACAM, HERC1, HERC2, HES7, HESX1, HEXA, HEXB, HFE, HGF, HGSNAT, HIBCH, HIKESHI, HINT1, HJV, HK1, HLCS, HMGCL, HMGCS2, HMX1, HNRNPH2, HOGA1, HOXA1, HOXC13, HPD, HPDL, HPGD, HPRT1, HPS1, HPSE2, HSD11B2, HSD17B10, HSD17B3, HSD17B4, HSD3B2, HSD3B7, HSPA9, HSPD1, HSPG2, HTRA2, HUWE1, HYAL1, HYDIN, HYLS1, IARS1, IARS2, IBA57, ICOS, IDS, IDUA, IER3IP1, IFIH1, IFNGR1, IFNGR2, IFT122, IFT140, IFT172, IFT27, IFT43, IFT52, IFT74, IFT80, IFT81, IGBP1, IGF1, IGF1R, IGFBP7, IGHMBP2, IGSF1, IHH, IKBKB, IKBKG, IL10RA, IL11RA, IL12RB1, IL1RAPL1, IL1RN, IL21R, IL2RA, IL2RB, IL2RG, IL7R, ILDR1, INPP5E, INPP5K, INPPL1, INS, INSR, INTU, INVS, IPO8, IQCB1, IQSEC1, IQSEC2, IRAK4, IRF8, IRX5, ISCA1, ISCA2, ITCH, ITGA3, ITGA6, ITGA7, ITGA8, ITGB4, ITK, ITPA, ITPR1, IVD, JAGN1, JAK3, JAM2, JAM3, JUP, KARS1, KATNB1, KATNIP, KCNE1, KCNJ1, KCNJ10, KCNJ11, KCNMA1, KCNQ1, KCTD7, KDELR2, KDM5B, KDM5C, KDM6A, KIAA0586, KIAA0753, KIDINS220, KIF14, KIF1A, KIF1C, KIF7, KIFBP, KISS1R, KLHL15, KLHL40, KLHL41, KLHL7, KNL1, KPTN, KRT10, KRT14, KRT18, KRT5, KRT8, KY, L1CAM, L2HGDH, LAGE3, LAMA1, LAMA2, LAMA3, LAMB1, LAMB2, LAMB3, LAMC2, LAMC3, LAMP2, LARGE1, LARP7, LARS2, LAS1L, LAT, LBR, LDHA, LDLR, LFNG, LGI4, LHB, LHFPL5, LHX3, LIAS, LIFR, LIG4, LIMS2, LINS1, LIPA, LIPT1, LMBR1, LMBRD1, LMNA, LMOD3, LNPK, LONP1, LOXHD1, LPIN1, LPIN2, LPL, LRBA, LRP2, LRP4, LRP5, LRPPRC, LRRC56, LRTOMT, LTBP2, LTBP3, LTBP4, LYRM4, LYRM7, LYST, LZTFL1, LZTR1, MAB21L2, MAG, MAGI2, MAGT1, MALT1, 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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, NEK1, NEK8, NEK9, NEMF, NEU1, NEUROG3, NEXMIF, NFASC, NFU1, NGF, NGLY1, NHEJ1, NHLRC1, NHP2, NHS, NIPAL4, NKAP, NKX3-2, NKX6-2, NMNAT1, NNT, NODAL, NONO, NOP10, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NPR2, NR0B1, NR1H4, NRROS, NRXN1, NSDHL, NSMCE2, NSMCE3, NSUN2, NT5C2, NT5C3A, NTNG2, NTRK1, NUBPL, NUDT2, NUP107, NUP133, NUP188, NUP62, NUP88, NUP93, NYX, OBSL1, OCLN, OCRL, ODAD1, ODAD2, OFD1, OGDH, OPA1, OPA3, OPHN1, ORAI1, ORC1, ORC4, ORC6, OSGEP, OSTM1, OTC, OTOA, OTOF, OTOG, OTOGL, OTUD5, OTUD6B, OTULIN, OXCT1, OXR1, P3H1, PAH, PAK3, PAM16, PANK2, PAPSS2, PARN, PARS2, PAX3, PC, PCBD1, PCCA, PCCB, PCDH12, PCDH15, PCDH19, PCK1, PCNT, PCSK1, PCYT1A, PCYT2, PDE10A, PDE6D, PDE6G, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PDZD7, PEPD, PERCC1, PET100, PEX1, PEX10, PEX11B, PEX12, 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Inquiry

SCARF2, SCN1B, SCN4A, SCN9A, SCNN1A, SCNN1B, SCNN1G, SCO1, SCO2, SCYL1, SCYL2, SDCCAG8, SDHA, SDHAF1, SDHD, SEC23A, SEC23B, SEC24D, SELENOI, SELENON, SEPSECS, SERAC1, SERPINB6, SERPINF1, SERPINH1, SETX, SFTPB, SFXN4, SGCA, SGCB, SGCD, SGCG, SGO1, SGPL1, SGSH, SH2D1A, SH3PXD2B, SH3TC2, SHOX, SHROOM4, SIL1, SKIC2, SKIC3, SLC10A7, SLC12A1, SLC12A3, SLC12A5, SLC12A6, SLC13A5, SLC16A1, SLC16A2, SLC17A5, SLC18A3, SLC19A2, SLC19A3, SLC1A4, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A26, SLC25A3, SLC25A38, SLC25A4, SLC25A42, SLC25A46, SLC26A2, SLC26A3, SLC26A4, SLC26A5, SLC27A4, SLC29A3, SLC2A1, SLC2A10, SLC2A2, SLC30A10, SLC33A1, SLC34A1, SLC34A3, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC35D1, SLC37A4, SLC39A13, SLC39A14, SLC39A4, SLC39A8, SLC3A1, SLC46A1, SLC4A1, SLC4A4, SLC52A2, SLC52A3, SLC5A1, SLC5A5, SLC5A6, SLC5A7, SLC6A3, SLC6A5, SLC6A8, SLC6A9, SLC7A7, SLC9A1, SLC9A3, SLC9A6, SLX4, SMAD4, SMARCAL1, SMC1A, SMOC1, SMPD1, SMPD4, SMS, SNAP29, SNORD118, SNX10, SNX14, SOD1, SOST, SOX3, SP110, SP7, SPAG1, SPARC, SPART, SPATA5, SPEG, SPG11, SPINK5, SPINT2, SPR, SPTBN2, SPTBN4, SQSTM1, SRD5A2, SRD5A3, SSR4, ST14, ST3GAL3, ST3GAL5, STAC3, STAG2, STAMBP, STAR, STAT1, STAT2, STAT5B, STIL, STIM1, STN1, STRA6, STRADA, STS, STT3A, STUB1, STX11, STXBP2, SUCLA2, SUCLG1, SUFU, SUMF1, SUOX, SURF1, SVBP, SYN1, SYNE1, SYNE4, SYNJ1, SYP, SZT2, TAC3, TACO1, TACR3, TAF1, TAF13, TAF2, TAF6, TAFAZZIN, TALDO1, TANGO2, TAP1, TAPT1, TARS2, TASP1, TAT, TBC1D20, TBC1D23, TBC1D24, TBC1D8B, TBCD, TBCE, TBCK, TBX15, TBX19, TBX22, TBX4, TBXAS1, TCAP, TCF12, TCIRG1, TCN2, TCTN2, TCTN3, TDP2, TECPR2, TECTA, TELO2, TENM3, TENT5A, TERT, TF, TFR2, TGDS, TGFB1, TGM1, TH, THOC2, THOC6, TIMM50, TIMM8A, TIMMDC1, TJP2, TK2, TMC1, TMCO1, TMEM107, TMEM126A, TMEM126B, TMEM132E, TMEM138, TMEM165, TMEM199, TMEM216, TMEM231, TMEM237, TMEM260, TMEM38B, TMEM67, TMEM70, TMEM94, TMIE, TMPRSS3, TMPRSS6, TMTC3, TMX2, TNFRSF11A, TNFRSF11B, TNFRSF13B, TNFSF11, TNNT1, TOE1, TOP3A, TP53RK, TPI1, TPK1, TPM3, TPP1, TPRKB, TPRN, TRAF3IP1, TRAIP, TRAK1, TRAPPC11, TRAPPC12, TRAPPC2, TRAPPC4, TRAPPC9, TRDN, TREX1, TRIM2, TRIM32, TRIM37, TRIOBP, TRIP11, TRIP13, TRIP4, TRIT1, TRMT1, TRMT10A, TRMT10C, TRMT5, TRMU, TRNT1, TRPM6, TRPV6, TSEN15, TSEN2, TSEN54, TSFM, TSHB, TSHR, TSPAN7, TSPEAR, TSPYL1, TTC19, TTC21B, TTC26, TTC7A, TTC8, TTI2, TTN, TTPA, TUBGCP2, TUBGCP4, TUBGCP6, TUFM, TUSC3, TWIST2, TWNK, TXN2, TXNDC15, TXNL4A, TYK2, TYMP, TYR, TYRP1, UBA1, UBA5, UBE2A, UBE2T, UBE3B, UBR1, UCHL1, UFC1, UFM1, UGDH, UGP2, UGT1A1, UMPS, UNC13D, UNC80, UPB1, UPF3B, UQCC2, UQCRB, UQCRC2, UQCRFS1, UQCRQ, UROC1, UROS, USB1, USH1C, USH1G, USH2A, USP18, USP53, USP9X, UVSSA, VAC14, VAMP1, VARS1, VARS2, VDR, VIPAS39, VLDLR, VMA21, VPS11, VPS13B, VPS13D, VPS33A, VPS33B, VPS37A, VPS41, VPS45, VPS51, VPS53, VRK1, VSX2, WARS2, WAS, WASHC5, WBP2, WDPCP, WDR19, WDR35, WDR4, WDR45, WDR45B, WDR62, WDR73, WDR81, WFS1, WHRN, WNK1, WNT1, WNT10A, WNT10B, WNT2B, WNT3, WNT4, WNT7A, WRAP53, WRN, WWOX, XIAP, XPA, XPC, XRCC2, XRCC4, XYLT1, XYLT2, YARS2, YIF1B, ZAP70, ZBTB24, ZC3H14, ZC4H2, ZDHHC9, ZFYVE26, ZIC3, ZMPSTE24, ZNF335, ZNF711, ZNHIT3

Disease-relevant changes (SNV/CNV) in the genes listed above will be reported (diseases underlying a recessive/X-chromosomal inheritance pattern). In addition, disease-causing alterations (SNV/CNV) may be reported in other genes known to be subject to genetic imprinting. Furthermore, it may be possible to report variants in other genes associated with dominantly inherited diseases if these are present as a genetic mosaic in one of the persons seeking advice. Only variants in genes are reported, which, within the framework of the respective underlying inheritance processes, can potentially lead to serious diseases in the planned offspring of the couple seeking advice, with an assumed onset of disease in childhood. Single variants of a parent, that might lead to the potential offspring being an inherited carrier without expected disease onset, are not reported. The findings are limited to variants of ACMG classes 4(LP)/5(P), that are confirmed to be disease-relevant according to current data. Individual ACMG class 3(VUS) variants may be additionally considered in individual cases at a physician's discretion.