INSTITUTE OF IMMUNOLOGY AND GENETICS

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Requesting physician:	**Patient information:**

Request Form Developmental disorders

Cause of examination: ☐ diagnostic ☐ predictive for familial mutation _____

Clinical information:

Family history:

(Check all that apply)

☐ Patient clinically affected

☐ No previous molecular genetic examinations existent

☐ The following previous molecular genetic examinations have been done:

_____

Date / Time of sampling:	**Signature:**

Specimen requirements and logistics:

- **Patient Consent Form for Genetic Testing according to the German Law and Request Form**
- **Billing information (Insurance-, Institutional- or Selfpay-Billing)**
- **5 ml EDTA blood collection tube labeled with patient name and date of birth**

Patient name: _____ **DOB:** _____

☐ **Aicardi-Goutieres-syndrome**
ADAR, IFIH1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TREX1

☐ **Autism**
ADNP, ADSL, ALDH5A1, AP1S2, ARID1B, ARX, ASH1L, ASXL3, ATRX, AUTS2, BRAF, CACNA1C, CASK, CDKL5, CHD2, CHD7, CHD8, CNOT3, CNTNAP2, CUL3, DHCR7, DPP&, DYRK1A, EHMT1, EIF4E, FGD1, FOXG1, FOXP1, GNAI1, GRIN2B, HPRT1, IL1RAPL1, KATNAL2, KDM5C, KIAA0442, KMT2A, KMT5B, L1CAM, MAGEL2, MAOA, MBD5, MECP2, MED12, MID1, MYT1L, NAA15, NEXMIF, NHS, NIPBL, NLGN4X, NLGN1, NLGN3, NLGN4, NOVA2, NRXN1, NSD1, OPHN1, PCDH19, PHF6, PNKP, POGZ, PQBP1, PTCHD1, PTEN, PTPN11, RAB39B, RAL1, RELN, RPL10, SCN1A, SCN2A, SETD5, SETD1B, SHANK2, SHANK3, SLC9A6, SLC9A9, SMARCB1, SMC1A, SMC3, SYN1, SYNGAP1, TANC2, TBL1XR1, TBR1, TCF4, TMLHE, TRP12, TRRAP, TSC1, TSC2, UBE2A, UBE3A, VAMP2, VPS13B, ZEB2, ZSWIM6

☐ **Bardet-Biedl-syndrome**
ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, C8orf37, CCDC28B, CEP164, CEP290, CEP41, IFT172, IFT27, IFT74, KIF7, LZTFL1, MKKS, MKS1, NPHP1, PTHB1, SDCCAG8, TMEM67, TRIM32, TTC21B, TTC8, WDPCP

☐ **CDG-syndrome**
ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG9, B4GALT1, CAD, CCDC115, COG1, COG2, COG4, COG5, COG6, COG7, COG8, DDOST, DHDDS, DOLK, DPAGT1, DPM1, DPM2, DPM3, FUT8, GMPPA, MAN1B1, MGAT2, MOGS, MPDU1, MPI, NGLY1, NUS1, PGM1, PMM2, RFT1, SLC35A1, SLC35C1, SLC39A8, SRD5A3, SSR4, STT3A, STT3B, TMEM165, TUSC3,

☐ **Coffin-Siris-syndrome**
ARID1A, ARID1B, ARID2, DPF2, SMARCA4, SMARCB1, SMARCE1, SOX11, SMARCC2, SMARCD1, SOX4, PHF6, KMT2A, DOCK6, GRIN2B, KMT2D, SHANK3, SMARCA2

☐ **Cornelia-de-Lange syndrome**
ANKRD11, ASXL1, BRD4, HDAC8, NIPBL, RAD21, SMC1A, SMC3, EP300, TAF1

☐ **Cortical brain malformation**
ACTB, ACTG1, ADAR, ADGRG1, AKT1, AKT3, ARID1A, ARID1B, ARFGEF2, ARID2, ARX, ASPM, B3GALNT2, CCND2, CDK5, CDK13, CECR1, COL4A1, COL4A2, CSNK2A1, CTC1, DAG1, DCX, DYNC1H1, FKRP, FKTN, FLNA, GPSM2, IFIH1, ISPD, JAM3, KATNB1, KIF1BP, KIF2A, KIF5C, LAMB1, LAMC3, LARGE1, MTOR, NDE1, NEDD4L, NSDHL, OCLN, OSGEP, PAFAH1B1, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX3, PEX5, PEX6, PEX7, PHF6, PIK3CA, PIK3R2, POMGNT1, POMGNT2, POMT1, POMT2, RELN, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RTTN, RXYLT1, SAMHD1, SMARCA4, SMARCB1, SMO, CMARCE1, SRPX2, TREX1, TUBA1A, TUBB, TUBB2B, TUBB3, TUBA8, TUBG1, VLDLR, WDR62

☐ **Craniosynostosis**
ALPL, ALX4, ASXL1, CDC45, COLEC11, CTSK, EFNB1, ERF, FGFR1, FGFR2, FGFR3, FLNA, GLI3, GNPTAB, IDS, IDUA, IHH, IL11RA, KAT6A, KMT2D, KRAS, MEGF8, MSX2, PHEX, POR, RAB23, RECQL4, RUNX2, SKI, SMAD6, SMO, STAT3, TCF12, TGFBR1, TGFBR2, TLK2, TWIST1, WDR35, ZEB2, ZIC1

☐ **Developmental disorders**
(Gene panel with 978 genes)*

☐ **Holoprosencephaly**
CDON, DHCR7, DLL1, EYA4, FBXW11, FGF8, FGFR1, GAS1, GLI2, GLI3, PTCH1, SHH, SIX3, SMAD2, TDGF1, TGIF1, ZIC2

☐ **Joubert-syndrome**
AHI1, ARL13B, ARL3, ARMC9, B9D1, B9D2, C2CD3, CC2D2A, CELSR2, CEP104, CEP120, CEP164, CEP190, CEP41, CPLANE1, CSPP1, EXOC8, FAM149B1, HYLS1, IFT172, INPP5E, KIAA0556, KIAA0586, KIAA0753, KIF7, MKS1, NPHP1, OFD1, PDE6D, PDPR, PIBF1, POC1B, RPGRIP1L, SUFU, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TTC21B, ZNF423

☐ **Kallmann-syndrome**
CHD7, FGF8, FGFR1, KAL1, PROK2, PROKR2

☐ **Leukodystrophy und Leukoencephalopathy**
AARS1, AARS2, ABCD1, ACOX1, ADAR, AIMP1, AIMP2, ALDH3A2, ARSA, ASPA, BCAP31, CLCN2, COL4A1, CSF1R, CTC1, CYP27A1, DARS1, DARS2, EARS2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EPRS1, EXOSC8, FAM126A, FOLR1, FUCA1, GALC, GBE1, GCDH, GFAP, GJA1, GJC2, HEPACAM, HIKESHI, HSD17B4, HSPD1, HTRA1, IFIH1, ISCA2, KCNT1, L2HGDH, LAMA2, LMNB1, MCOLN1, L`MLC1, NAXE, NKX6-2, NOTCH3, PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PLAA, PLEKHG2, PLP1, POLR1C, POLR3A, POLR3B, POLR3K, PSAP, PYCR2, RARS1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, SAMHD1, SCP2, SLC16A2, SLC17A5, SNORD118, SOX10, SPTAN1, STN1, SUmf1, TMEM106B, TREX1, TUB4A, TUBB4A, UFM1, VPS11, ZNHIT3

☐ **Lissencephaly**
ARX, DCX, FKRP, ISPD, NDE1, NUDC, PAFAH1B1, POMGNT1, POMT1, POMT2, RELN, TK2, TUBA1A, VLDLR

☐ **Lysosomal disease**
AGA, ARSA, ARSB, CTNS, CTSA, FUCA1, GALC, GALNS, GBA, GLA, GLB1, GM2A, GNTAB, GNPTG, GNS, GUSB, HEXA, HEXB, HGSNAT, HYAL1, IDS, IDUA, LIPA, MAN1B1, MAN2B1, MANBA, MCOLN1, NAGA, NAGLU, NEU1, NPC1, NPC2, PSAP, SGSH, SLC17A5, SMPD1, SUMF1, VPS33A

☐ **Macrocephaly**
ABCC9, AKT1, AKT3, AMER1, ASPA, BRWD3, CCDC88C, CCDCC22, CDKN1C, CHD8, CUL4B, DIS3L2, DNMT3A, DVL1, DVL3, EIF2B5, EZH2, FOXP1, GCDH, GFAP, GLI3, GPSM2, GPC3, GRIA3, HEPACAM, HERC1, HRAS, HUWE1, IGF2, KIAA0196, KPTN, KRAS, KIF7, LZTR1, L1CAM, MED12, MLC1, MTOR, NDUFA1, NFIB, NFIX, NONO, NRAS, NSD1, OFD1, PHF6, PIGA, PIGN, PIGT, PIGV, PIK3CA, PIK3R2, PPP1CB, PPP2R5D, PTCH1, PTCH2, PTEN, RAB39B, RAF1, RHEB, RIN2, RIT1, RNF135, ROR2, SETD2, SHANK3, SHOC2, SNX14, SOS1, STRADA, SUFU, SYN1, TBC1D7, TMCO1, UPF3B, WASHC5, WNT5A, ZDHHC9

* A detailed gene list can be provided on request.

Patient name: _____ **DOB:** _____

☐ **Meckel-Gruber syndrome**
AHI1, B9D1, CC2D2A, CEP120, CEP290, CEP55, CSPP1, KIAA0586, KIAA0753, MKS1, NPHP3, RPGRIP1L, TCTN1, TCTN2, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TTC21B, TXNDC15, WDPCP

☐ **Microcephaly und pontocerebellar hypoplasia**
AMPD2, ANKLE2, ASNS, ASPM, ATR, CASK, CDK5RAP2, CDK6, CENPE, CENPJ, CEP135, CEP152, CEP63, CHMP1A, CIT, CLP1, COASY, DYRK1A, EXOSC3, EXOSC8, EXOSC9, FOXG1, IER3IP1, KAT6A, KIF11, KIF14, KNL1, MBD5, MCPH1, MECP2, MED17, MFSD2A, NCAPD2, NIN, NSMCE2, NUP37, PCLO, PCNT, PHC1, PLK4, PNKP, PPP1R15B, RAB18, RARS2, RBBP8, SASS6, SEPSECS, SLC25A19, SLC15A46, SLC9A6, STAMBP, STIL, TBC1D23, TOE1, TOP3A, TRAIP, TRMT10A, TSEN15, TSEN2, TSEN54, TUBGCP4, TUBGCP6, VPS53, VRK1, WDR62, ZNF335

☐ **Senior-Loken-syndrome**
CEP164, CEP290, INVS, IQCB1, NPHP1, NPHP3, NPHP4, SCLT1, SDCCAG8, TMEM67, TRAF3I91, WDR19, ZNF423

☐ **Skeletal malformation**
(Gene panel with 319 genes)*

☐ **Zellweger-syndrome**
ABCD1, ACOX1, AMACR, HSD17B4, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX3, PEX5, PEX6, PEX7, SCP2

* A detailed gene list can be provided on request.