PEDIATRIC GLOBAL DELAY DEFINE&DECIDE

BARCODE

PHYSICIAN INFORMATION

INSTITUTION/PRACTICE	ADDRESS (STREET NAME, NO., CITY, POSTAL CODE, COUNTRY)
FIRST NAME	TELEPHONE NUMBER (COUNTRY CODE & NUMBER)
LAST NAME	E-MAIL ADDRESS (FOR REPORT ACCESS)

PATIENT INFORMATION

FIRST NAME	ADDRESS (STREET NAME, NO., CITY, POSTCODE, COUNTRY)
LAST NAME	TELEPHONE NUMBER (COUNTRY CODE & NUMBER)
DATE OF BIRTH (DD/MM/YYYY)	GENDER (MALE/FEMALE/OTHER - SPECIFY KARYOTYPE)
PERSONAL IDENTIFICATION NO.	SAMPLE COLLECTION DATE (DD/MM/YYYY)
REASON FOR TEST (DIAGNOSIS, PREDICTIVE, CARRIER)	

DECLARATION OF CONSENT (ACCORDING TO GERMAN GENETIC DIAGNOSTICS ACT, GenDG)

Applicable only for the determination of genetic (hereditary) characteristics

The GenDG requires provision of detailed information and a written consent for all genetic investigations as well as genetic counselling prior to both predictive (applies to healthy individuals) and prenatal testing (with restrictions: prenatal testing is not performed for late manifesting disorders, including Hereditary Cancer Panels). The German Society of Human Genetics (GfH) and the Association of German Human Geneticists (BVDH) recommend clarifying the issues listed below during the information process. Please read the declaration of consent carefully and tick the boxes, in accordance with your consent.

By signing the form below I confirm that I:

- Have been fully informed by my physician about the significance and consequences of the genetic investigation, in compliance with GenDG.
- Have read/have been read the Information for Patients (page 5) which is attached to this form and which I fully understand.
- Have been given sufficient opportunity to discuss open questions.
- Authorize [insert legal entity here] to collect the necessary samples for investigation (blood, tissue, chorionic villus cells or amniotic fluid for prenatal diagnosis) and to send this form to MVZ Martinsried GmbH, Lochhamer Str. 29, 82152 Martinsried, Germany, in order to perform the tests requested through this form.
- Consent to the genetic test being carried out in order to clarify the disease/dysfunction/suspected diagnosis.

YES NO

- ☐ ☐ I agree that the investigation or parts of the investigation may be forwarded to collaborating medical laboratories, if necessary.
- ☐ ☐ I agree with the evaluation of additional genes in the same indication group as part of the research.
- ☐ ☐ I agree that the remaining specimens may be stored for further investigations after the examination is completed, yet not claiming storage.
- ☐ ☐ I agree that the specimens, and if applicable DNA sequence information, may be made available anonymously for quality management and scientific purposes.
- ☐ ☐ I agree that the results of the analysis may be stored for a longer period than the statutory period of 10 years, yet not claiming storage of results.
- ☐ ☐ I agree to the storage and use of my test results under the protection of anonymity in a statistical database used for scientific purposes and to help diagnose genetic diseases. I understand that
- ☐ ☐ I will remain under the protection of anonymity and I cannot be identified during the analysis of the data and that any personal information will be transformed into information of a non-personal nature.

By signing the form below I confirm that:

- I may stop the investigation at any time and ask for the results available until that time to be destroyed.
- I may withdraw any of my consents given through this form entirely or in part at any time without giving reasons.
- I will be charged for the costs incurred until the time of withdrawal of consent.
- I may choose not to be informed about the test results (right not to know).
- I know that the genetic investigation and evaluation is limited to the requested indication and no statements will be made about other diseases.
- All information I have provided is true and correct.

Communication of additional findings found during the course of the research

☐ YES, I wish to be informed about additional findings.
☐ NO, I do not wish to be informed about additional findings.

In addition,

- YES☐ NO☐ I agree that a copy of the results of the analysis may be sent to the following physician(s), in accordance with my express requests and according to [insert legal entity here] internal procedures.

DR(S) NAME

STREET

POSTCODE/CITY

COUNTRY

PLACE

DATE

SIGNATURE OF PATIENT OR PARENT/LEGAL GUARDIAN

PHYSICIAN'S SIGNATURE

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RELEVANT CLINICAL INFORMATION

Interpretation of the genetic results relies on an accurate and complete clinical picture of the patient, including clinical manifestations, family medical history and previous diagnoses.

CHECK ALL BOXES THAT APPLY TO YOUR PATIENT:

☐ Patient has a confirmed or suspected diagnosis of an inherited global developmental delay disorder.

(Suspected) Diagnosis: _____

Select all features that apply to your patient. Please note that symptoms vary in type and severity between patients and that not all symptoms related to global developmental delay and intellectual disability are listed below.

DEVELOPMENTAL

- ☐ Behavioral disorders/problems (aggressiveness, anxiety, attention deficit, autistic, extrapyramidal movement disorders, hyperactivity, repetitive or stereotypical hand movements)
- ☐ Cognitive disorder/impairment
- ☐ Intellectual disability (mild, moderate, severe)
- ☐ Motor delay
- ☐ Speech/language development delay

MUSCULOSKELETAL

- ☐ Brachydactyly V
- ☐ Brachydactyly-clinodactyly V
- ☐ Fetal fingertip pads
- ☐ Gait ataxia
- ☐ Growth retardation/delay
- ☐ Hyperextensible joints
- ☐ Hypoplasia/aplasia of the end phalanx of the 5th finger or fingernail
- ☐ Hypoplastic distal phalanges of fingers and toes
- ☐ Long, slender fingers
- ☐ Muscle atrophy
- ☐ Muscle hypotonia
- ☐ Overgrowth/excessive growth
- ☐ Short height/stature
- ☐ Skeletal anomalies (e.g., scoliosis, kyphoscoliosis)
- ☐ Tall stature

GENITOURINARY

- ☐ Hypoplastic genitals
- ☐ Hypospadias
- ☐ Malformation of kidneys and the urinary tract
- ☐ Postpubertal macroorchidism

FACIAL DYSMORPHISM

- ☐ Chin (e.g., accentuated, pointed, prominent)
- ☐ Dental anomalies (e.g., decayed teeth, decreased root to crown ratio, decreased tooth size)
- ☐ Ear abnormalities (e.g., dysplastic, low-set, prominent, small)
- ☐ Eye involvement (e.g., coloboma, strabismus)
- ☐ Eyebrows (e.g., arched, broad, sparse, thick)
- ☐ Eyelids (e.g., laterally elongated, ptosis, slope downwards)
- ☐ Eyes (e.g., deep-set, widely-spaced)
- ☐ Forehead (e.g., deep hairline, high and broad, narrow receding, prominent)
- ☐ Hearing loss/deafness
- ☐ Jaw (e.g., micrognathia, retrognathia)
- ☐ Lips (e.g., cleft lip, everted lower lip, full lips, M-shaped upper lip, wide mouth)
- ☐ Macrocephaly
- ☐ Microcephaly
- ☐ Nose, flared nostrils
- ☐ Nose, nasal bridge (e.g., broad, curved)
- ☐ Nose, nasal root (e.g., broad, prominent)
- ☐ Nose, nasal tip (e.g., broad, flattened, rounded)
- ☐ Nose, short
- ☐ Palate (e.g., cleft palate, pointed and high)
- ☐ Short philtrum
- ☐ Visual impairment

OTHERS

- ☐ Brain malformations
- ☐ Chronic constipation
- ☐ Epilepsy
- ☐ Episodes of apnea or hyperpnea
- ☐ Episodes of hyperventilation
- ☐ Failure to thrive
- ☐ Heart defects
- ☐ Seizures

Please provide any additional clinical information and all relevant medical reports.

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FAMILY HISTORY

☐ Patient has a family member (first or second degree relative) diagnosed with an inherited global developmental delay disorder.

Diagnosis: _____

☐ Patient has a family member diagnosed with an inherited global developmental delay disorder who had genetic testing that identified a specific variant.

Diagnosis: _____ Variant: _____

☐ Patient has a family member with a similar clinical history.

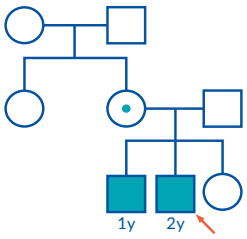
Please provide any additional clinical information and all relevant medical reports.

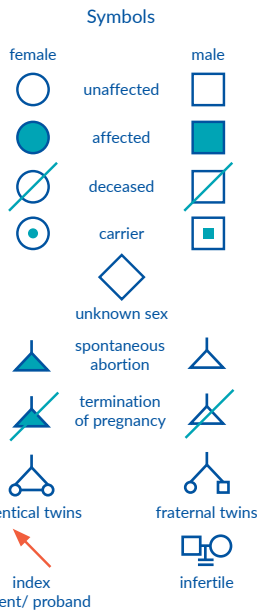
Testing the index patient will improve data interpretation. If this is not the index patient, is he/she available for genetic testing?

☐ Yes ☐ No ☐ N/A

PEDIGREE

Example of a pedigree:
Coffin-Lowry syndrome (included in Intellectual Disability Panel)





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OUR TESTS

Please select the most appropriate test for your patient from the following test and panel options:

☐ **FRAGILE X** ☐ **KARYOTYPING** ☐ **MICROARRAY CGH**

OUR PANELS

Please check our website for the most up-to-date gene list www.medicover-genetics.com

1 **AUTISM SPECTRUM DISORDERS PANEL**
ALDH5A1, AP1S2, ARX, ATRX, AUTS2, BRAF, CACNA1C, CASK, CDKL5, CHD7, CHD8, CNOT3, CNTNAP2, DHCR7, DPP6, EHMT1, FGD1, FOXG1, FOXP1, FOXP2, GNAI1, GRIN2B, HPRT1, KDM5C, L1CAM, MBD5, MECP2, MED12, MEF2C, MID1, NHS, NIPBL, NLGN3, NLGN4X, NRXN1, NSD1, OPHN1, PCDH19, PHF6, PNKP, PQBP1, PTCHD1, PTEN, PTPN11, RAB39B, RAI1, RPL10, SCN1A, SHANK2, SHANK3, SLC9A6, SMARCB1, SMC1A, SMC3, TBR1, TCF4, TMLHE, TSC1, TSC2, UBE2A, UBE3A, VPS13B, ZEB2

2 **BRAIN MALFORMATIONS, COMPREHENSIVE PANEL**
AHI1, ARFGEF2, ARX, CASK, CC2D2A, CEP290, CEP41, DCX, EOMES, FKRP, FKTN, FLNA, GPR56, KIF7, LAMC3, LARGE, MKS1, NDE1, NPHP1, OCLN, OPHN1, PAFAH1B1, POMGNT1, POMT1, POMT2, PQBP1, RARS2, RELN, RPGRIP1L, SRPX2, TMEM138, TMEM216, TMEM237, TMEM67, TSEN2, TSEN34, TSEN54, TUBA1A, TUBA8, TUBB2B, TUBB3, VLDLR, VRK1

3 **BRAIN MALFORMATIONS, LISSENCEPHALY PANEL**
ARX, DCX, KATNB1, MACF1, NDE1, PAFAH1B1, RELN, TMTC3, TUBA1A

4 **BRAIN MALFORMATIONS, PONTOCEREBELLAR HYPOPLASIA PANEL**
AMPD2, CHMP1A, CLP1, COASY, EXOSC3, EXOSC8, EXOSC9, PCLO, RARS2, SEPSECS, SLC25A46, TBC1D23, TOE1, TSEN15, TSEN2, TSEN34, TSEN54, VPS53, VRK1

5 **BRAIN MALFORMATIONS, TUBULINOPATHIES PANEL**
TBCD, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1

6 **COFFIN-SIRIS SYNDROME PANEL**
ARID1A, ARID1B, ARID2, DPF2, SMARCA4, SMARCB1, SMARCC2, SMARCE1, SOX11, SOX4

7 **CONGENITAL DISORDERS OF GLYCOSYLATION PANEL**
ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, B4GALT1, CAD, CCDC115, COG1, COG4, COG5, COG6, COG7, COG8, DDOST, DOLK, DPAGT1, DPM1, DPM2, DPM3, MGAT2, MOGS, MPDU1, MPI, NGLY1, PGM1, PMM2, RFT1, SLC35A1, SLC35A2, SLC35C1, SLC39A8, SRD5A3, SSR4, STT3A, STT3B, TMEM165, TMEM199, TUSC3

8 **CORNELIA DE LANGE SYNDROME PANEL**
ANKRD11, BRD4, HDAC8, NIPBL, RAD21, SMC1A, SMC3

9 **GPI ANCHOR DEFICIENCY PANEL**
GPAA1, PGAP1, PGAP2, PGAP3, PIGA, PIGB, PIGC, PIGG, PIGH, PIGL, PIGM, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGT, PIGU, PIGV, PIGW, PIGY

10 **INTELLECTUAL DISABILITY PANEL**
ABCD1, ACSL4, AFF2, AGTR2, AIFM1, ALG13, AMER1, AP1S2, AP4B1, AP4E1, AP4M1, AP4S1, ARHGEF6, ARHGEF9, ARX, ATP6AP2, ATP7A, ATRX, BCAP31, BCOR, BRWD3, CA8, CASK, CC2D1A, CCDC22, CDH15, CDKL5, CLCN4, CLIC2, CNKSR2, CNTNAP2, CRBN, CREBBP, CUL4B, DCX, DDX3X, DKC1, DLG3, DMD, EBP, EIF2S3, EP300, ERLIN2, FAAH2, FANCB, FGD1, FLNA, FMR1, FOXG1, FOXP1, FRMPD4, FTSJ1, GDI1, GJB1, GK, GPC3, GPKOW, GRIA3, GRIK2, GRIN2B, GSPT2, HCCS, HCFC1, HDAC6, HDAC8, HMGB3, HNRNPH2, HPRT1, HSD17B10, HUWE1, IDS, IGBP1, IKBKG, IL1RAPL1, IQSEC2, KDM5C, KDM6A, KIAA2022, KIF4A, KIRREL3, KLF8, KLHL15, L1CAM, LAMP2, LAS1L, MAGT1, MAN1B1, MAOA, MBTPS2, MECP2, MED12, MEF2C, MID1, MID2, MSL3, MTM1, NAA10, NDP, NDUFA1, NEXMIF, NHS, NLGN3, NLGN4X, NONO, NRXN1, NSDHL, NXF5, OCRL, OFD1, OGT, OPHN1, OTC, PAK3, PCDH19, PDHA1, PGK1, PHF6, PHF8, PIGA, PLP1, PORCN, PQBP1, PRPS1, PRSS12, PTCHD1, RAB39B, RAB40AL, RAI1, RBM10, RBMX, RLIM, RNF113A, RPL10, RPS6KA3, SHROOM4, SLC16A2, SLC25A5, SLC6A8, SLC9A6, SMC1A, SMS, SOBP, SOX3, SRPX2, SSR4, ST3GAL3, STAG2, STXBP1, SYN1, SYNGAP1, SYP, TAF1, TCF4, THOC2, TIMM8A, TMLHE, TRAPPC9, TSPAN7, TUSC3, UBE2A, UBE3A, UPF3B, USP27X, USP9X, VLDLR, WDR13, WDR45, ZC3H14, ZC4H2, ZCCHC12, ZDHHC15, ZDHHC9, ZEB2, ZMYM3, ZNF41, ZNF526, ZNF674, ZNF711, ZNF81

11 **MACROCEPHALY PANEL**
ABCC9, AKT3, AMER1, ASPA, BRWD3, CCDC22, CCND2, CDKN1C, CHD8, CUL4B, DIS3L2, DNMT3A, DVL1, DVL3, EZH2, FOXP1, GCDH, GFAP, GLI3, GPC3, GRIA3, HEPACAM, HERC1, HRAS, HUWE1, KIF7, KPTN, KRAS, LZTR1, MED12, MLC1, MTOR, NDUFA1, NFIB, NFIX, NONO, NRAS, NSD1, NXN, OFD1, PIGA, PIGN, PIGT, PIGV, PIK3R2, PPP1CB, PPP2R5D, PTCH1, PTCH2, PTEN, RAB39B, RAF1, RHEB, RIT1, RNF135, ROR2, SETD2, SHOC2, SOS1, SUFU, TBC1D7, TMCO1, UPF3B, WASHC5, WNT5A, ZDHHC9

12 **MICROCEPHALY PANEL**
ANKLE2, ASPM, CDK5RAP2, CDK6, CENPE, CENPF, CENPJ, CEP135, CEP152, CIT, COPB2, DONSON, KDM6A, KIF14, KMT2D, KNL1, MCPH1, MFSD2A, NCAPD2, NCAPD3, NCAPH, NUP37, PCNT, PHC1, SASS6, STIL, WDFY3, WDR62, ZEB1, ZNF335

13 **OVERGROWTH SYNDROMES PANEL**
CDKN1C, DIS3L2, DNMT3A, EED, EZH2, GPC3, HERC1, HIST1H1E, NFIX, NSD1, OFD1

14 **PEDIATRIC NEUROTRANSMITTER DISORDERS PANEL**

15 **RASOPATHIES, COMPREHENSIVE PANEL**
BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MRAS, NF1, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, RRAS, SHOC2, SOS1, SOS2, SPRED1

16 **RETT SYNDROME PANEL**
CDKL5, FOXG1, MECP2

17 **RETT SYNDROME AND RELATED DISORDERS PANEL**
ALDH5A1, ARX, BDNF, CDKL5, CNTNAP2, FOXG1, FOXP2, IQSEC2, KCNA2, KCNQ2, KIF1A, MECP2, MEF2C, NRXN1, NTNG1, PLP1, SCN2A, SCN8A, STXBP1, TCF4, UBE3A, ZEB2

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INFORMATION FOR PATIENTS

BARCODE

PATIENT INFORMATION

FIRST NAME	GENDER (MALE/FEMALE/OTHER - SPECIFY KARYOTYPE)
LAST NAME	TELEPHONE NUMBER (COUNTRY CODE & NUMBER)
DATE OF BIRTH (DD/MM/YYYY)	E-MAIL ADDRESS
CLINICAL DIAGNOSIS	

Genetic counselling or counselling by the ordering physician is necessary before ordering a test in order to inform the patient of all of the possible outcomes and the limitations of the genetic test.

I understand that I will be tested for:
(to be filled in by physician)

I understand that the biological sample will be used to determine if I, or members of my family, are carriers of a genetic variant causing the disease, or are carriers of the disease, or have an increased risk of developing a disease.

The role of genetic testing. In many cases, a genetic test can directly detect a genetic alteration. Molecular tests can identify structural changes in the DNA (variants). Cytogenetic tests identify the chromosomal changes (structural or numerical). The sensitivity and specificity of each test varies. The tests offered are complex analyses and are performed using high-end equipment. The methods are externally validated, but there is a minimal possibility of errors.

The significance of the results. If the result is identified as being directly causative of the clinical manifestations, it is considered to be conclusive. If the test does not identify the causative mutations of the clinical manifestations, it is considered to be inconclusive and this does not preclude other genetic changes (or non-genetic factors) responsible for the disease (a genetic disease or susceptibility to a genetic condition is not excluded). Therefore, an inconclusive result (no causative mutation identified) does not exclude the existence of other pathogenic genetic changes (variants) not tested through the current analysis.
Interpretation of the genetic results relies on a complete clinical picture of the patient, including clinical manifestations, family medical history and previous diagnoses. An error in diagnosis could occur due to a clinical picture that is different from that declared. In addition, the test can identify a possible nonpaternity. The test results will be forwarded to the patient by the geneticist or ordering physician and are confidential.

Incidental findings. Genetic testing can provide information unrelated to the purpose of the test, but that may have medical importance for the patient or family (information correlated with an increased risk for incurable disorders).

Use of the sample/result. The sample provided will be used solely for the purpose of the test and for which I have given my written consent.
Test results can also be used for research and to improve the diagnosis and treatment of genetic diseases.

The genetic material can be used for other purposes only with my prior express written consent.

Post-testing genetic counselling. A conclusive result may offer the patient information on the susceptibility, diagnosis, possible prognosis and/or heritability of the disease. An inconclusive result may lead to confusion and anxiety or may suggest the need for further genetic testing. Therefore, post-testing genetic counselling is advised for the clinical interpretation of the results.

By my signature, I hereby certify that:

1. I have been informed of the nature and purpose of the genetic test.
2. I have been informed of the benefits and limitations of the genetic test by _____ (name of physician).
3. I have been informed that the genetic test can provide information/results which have no connection with the purpose of testing. I understand that only I decide if I want those additional results to be provided.
4. I have received clear answers to my questions in relation to the genetic test.
5. I have received a copy of this form.
6. I agree to provide a sample for the above mentioned genetic test.

Completed by: ☐ Parent/Legal Guardian ☐ Patient

FIRST NAME

LAST NAME

DATE OF COMPLETION

SIGNATURE

I have explained the risks and benefits of the test as well as alternative test methods to the parent/legal guardian. I have answered all the questions from the parent/legal guardian.

Name of the ordering physician

FIRST NAME

LAST NAME

SIGNATURE OF THE ORDERING PHYSICIAN

DATE OF SIGNATURE

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