Diagenom GmbH
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Germany

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Patient

Surname, name	
Date of birth	
Street	
Town, postcode	

Physician

Name	
Clinic/institute	
Address	

Sex: ○ f ○ m
Invoice to: ○ clinic ○ patient ○ other (specify)

Tel.-/fax no. _____
Report by fax? ○ yes ○ no

Patient or family member(s) already known to Diagenom: ○ yes ○ no

Sample date: _____
Follow-up visit: _____

Samples:

Cytogenetics
- ○ Heparin blood (10 mL at RT)
- ○ Bone marrow aspirate in heparin (5 mL at RT)
- ○ Amniotic fluid (15-20 mL at RT)
- ○ CVS (10-20 mg in sterile medium at RT)
- ○ Other (please inquire)

Molecular genetics
- ○ EDTA blood (3-5 mL at RT)
- ○ DNA (1-5 µg at RT)
- ○ Cell culture e.g. fibroblasts (25 mL, confluent at RT)
- ○ CVS (10-20 mg in sterile medium at RT)
- ○ Amniotic fluid (15-20 mL at RT)

Sampling for cytogenetic analysis: Monday – Thursday; Friday and Saturday on arrangement. Please ensure delivery within 24h. Please contact us prior to sending prenatal samples for molecular genetic analysis
Address for sending samples: Diagenom GmbH, Robert-Koch-Str. 10, D-18059 Rostock, Germany

Clinical symptoms

_____
_____
_____
_____
_____
_____
_____
_____
_____

Family tree

Declaration

I affirm that the submitted patient material was obtained in accordance with relevant national legislation and the results likewise will only be used in accordance with the appropriate regulations.

Location/date: _____

Physician's signature: _____

Index

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Cytogenetics (Chromosome analysis/FISH)

- ○ Postnatal cytogenetic investigation
- ○ Prenatal cytogenetic investigation
- ○ **Suspected diagnosis:** _____

Tumor Cytogenetics

- ○ **First diagnosis** **Date :** | | | |
- ○ **Therapy control**
- ○ **Previous therapy** **Date :** | | | | **Therapy details :**_____
- ○ **Status after transplantation** **Gender change** ○ yes ○ **no**

Oncology

Material : EDTA-blood

- ○ **Alagille syndrome 1** (JAG1)
- ○ **Alagille syndrome 2** (NOTCH2)
- ○ **APC-associated polyposis, FAP** (APC)
- ○ **Beckwith-Wiedemann syndrome** (CDKN1C)
- ○ **Breast-/ovarian carcinoma, familial** (BRCA1, BRCA2)
- ○ **Breast-/ovarian carcinoma, familial** (RAD51C)
- ○ **Gastrointestinal stroma tumor, GIST** (KIT, PDGFRA)
- ○ **Hereditary diffuse gastric cancer** (CDH1)
- ○ **Hereditary non-polyposis colon cancer type 1, HNPCC1** (MSH2)
- ○ **Hereditary non-polyposis colon cancer type 2, HNPCC2** (MLH1)
- ○ **Hereditary non-polyposis colon cancer type 4, HNPCC4** (PMS2)
- ○ **Hereditary non-polyposis colon cancer type 5, HNPCC5** (MSH6)
- ○ **Lymphoproliferative syndrome, X-linked** (SH2D1A)
- ○ **Multiple endocrine neoplasia type 1** (MEN1)
- ○ **Multiple endocrine neoplasia type 2** (RET)
- ○ **MYH-associated polyposis** (MUTYH)
- ○ **Peutz-Jeghers syndrome** (STK11)
- ○ **Prostate cancer** (ELAC2, RNASEL, BRCA2, SRD5A2)
- ○ **Renal cell carcinoma, papillary** (MET)
- ○ **Sotos syndrom** (NSD1)
- ○ **Von Hippel-Lindau syndrome** (VHL)

Material : Tumor tissue (paraffin section)

- ○ **BCR-ABL fusion gene** (BCR, ABL1)
- ○ **BRAF-associated tumor conditions [V600E]** (BRAF)
- ○ **EGFR- associated tumor conditions** (EGFR)
- ○ **Gastrointestinal stroma tumor, GIST** (KIT, PDGFRA)
- ○ **KRAS- associated tumor conditions** (KRAS)
- ○ **Microsatellite instability with HNPCC** (MMR-Gene)

Hemostaseology/Hematology

Hemostaseology

- ○ **Antithrombin deficiency** (AT3)
- ○ **Protein C deficiency** (PROC)
- ○ **Protein S deficiency** (PROS1)
- ○ **Fibrinogen; A-, dys-, hypofibrinogenemia** (FGA, FGB, FGG)
- ○ **Factor II deficiency** (F2)
- ○ **Factor V deficiency** (F5)
- ○ **Factor VII deficiency** (F7)
- ○ **Factor VIII deficiency (hemophilia A)** (F8)
- ○ **Factor IX deficiency (hemophilia B)** (F9)
- ○ **Factor X deficiency** (F10)
- ○ **Factor XI deficiency** (F11)
- ○ **Factor XII deficiency** (F12)
- ○ **Factor XIII deficiency** (F13A1, F13B)
- ○ **Von Willebrand syndrome** (VWF)
- ○ **Bernard-Soulier syndrome** (GP1BA, GP1BB, GP9)
- ○ **Glanzmann thrombasthenia** (ITGA2B, ITGB3)
- ○ **MYH9-related thrombocytopenia** (MYH9)

Hemoglobinopathy

- ○ **α-Thalassemia** (HBA1, HBA2)
- ○ **β-Hemoglobinopathy** (HBB)
  - ○ **β-Thalassemia**
  - ○ **Sickle cell anemia**
  - ○ **Other β-hemoglobinopathy (HbC, HbD etc.)**
- ○ **δβ-Thalassemia incl. Hb Lepore and HPFH** (HBB, HBD)
- ○ **Glucose-6-phosphate dehydrogenase deficiency** (G6PD)
- ○ **Pyruvat kinase deficiency** (PKLR)

Hematology

- ○ **Polycythaemia vera/ET** (JAK2)
- ○ **Mastocytosis [D816V]** (KIT)
- ○ **Acute myeloid leukemia [AML]** (FLT3)
- ○ **CML: BCR-ABL fusion gene, qualitative** (BCR, ABL1)
- ○ **CML: BCR-ABL fusion gene, quantitative** (BCR, ABL1)

Pharmacogenetics

- ○ **Azathioprin toxicity** (TPMT)
- ○ **Fluorpyrimidin (5-FU) toxicity** (DPYD IVS14+1 G>A)
- ○ **Irinotecan toxicity** (UGT1A1)
- ○ **Malignant hyperthermia** (RYR1)
- ○ **Phenprocoumon sensitivity** (VKORC1, CYP2C9)
- ○ **Postanesthetic apnea risk** (BCHE)

Endokrinology

- ○ **Congenital adrenal hyperplasia, 21-hydroxylase deficiency** (CYP21A2)
- ○ **Congenital adrenal hyperplasia 11-β- hydroxylase deficiency** (CYP11B1)
- ○ **Congenital adrenal hyperplasia, 3-β-hydroxysteroid-dehydrogenase def.** (HSD3B2)
- ○ **Pituitary hormone deficiency** (PROP1)
- ○ **Kallmann syndrome 1** (KAL1)
- ○ **MODY-Diabetes type 2 and 3**
- ○ **MODY-Diabetes type:**
  ○ **1** (HNF4A) ○ 2 (GCK) ○ **3** (HNF1A) ○ **4** (IPF1) ○ **5** (HNF1B) ○ **6** (NEUROD1)

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Metabolic disease

Hemochromatosis

- ○ **Hemochromatosis, classic [C282Y/H63D/S65C]** (HFE)
- ○ **Hemochromatosis, classic [sequencing]** (HFE)
- ○ **Hemochromatosis type 2, juvenile** (HJV, HAMP)
- ○ **Hemochromatosis type 3** (TFR2)
- ○ **Hemochromatosis type 4** (SLC40A1)

Porphyria

- ○ **Acute intermittent porphyria** (HMBS)
- ○ **Chronic hepatic porphyria** (UROD)
- ○ **Erythrohepatic protoporphyria** (FECH)
- ○ **Hereditary coproporphyria** (CPOX)
- ○ **Porphyria variegata** (PPOX)

Lysosomal storage disease

- ○ **Alpha-Mannosidosis** (α-Mannosidase - MAN2B1)
- ○ **Beta-Mannosidosis** (β-Mannosidase - MANBA)
- ○ **Fabry disease** (α-Galactosidase - GLA)
- ○ **Farber disease** (ASAH1)
- ○ **Fucosidosis** (α-L-Fucosidase - FUCA1)
- ○ **Gaucher disease** (Glucocerebrosidase - GBA)
- ○ **GM1-Gangliosidosis** (β-Galactosidase - GLB1)
- ○ **Krabbe disease** (Galactocerebrosidase – GALC)
- ○ **McArdle disease, Glycogen storage disease type V** (PYGM)
- ○ **Metachromatic leukodystrophy** (Arylsulphatase A - ARSA)
- ○ **Mucopolysaccharidosis type 1, Hurler/Scheie disease** (IDUA)
- ○ **Mucopolysaccharidosis type 2, M. Hunter** (IDS)
- ○ **Mucopolysaccharidosis type 3a, Sanfilippo syndrome A** (SGSH)
- ○ **Mucopolysaccharidosis type 3b, Sanfilippo syndrome B** (NAGLU)
- ○ **Mucopolysaccharidosis type 3c, Sanfilippo syndrome C** (HGSNAT)
- ○ **Mucopolysaccharidosis type 3d, Sanfilippo syndrome D** (GNS)
- ○ **Mucopolysaccharidosis type 4a, Morquio A** (GALNS)
- ○ **Mucopolysaccharidosis type 4b, Morquio B** (GLB1)
- ○ **Mucopolysaccharidosis type 6, Maroteaux-Lamy synd.** (ARSB)
- ○ **Mucopolysaccharidosis type 7, Sly syndrome** (GUSB)
- ○ **Mucopolysaccharidosis type 9** (HYAL1)
- ○ **Mucolipidosis II, I-Cell disease** (GNPTAB)
- ○ **Multiple sulphatase deficiency** (SUMF1)
- ○ **Niemann-Pick disease type A/B** (SMPD1)
- ○ **Niemann-Pick disease type C1** (NPC1)
- ○ **Niemann-Pick disease type C2** (NPC2)
- ○ **Pompe disease, Glycogen storage dis. type II** (α-Glucosidase - GAA)
- ○ **Prosaposin deficiency** (PSAP)
- ○ **Sandhoff disease, GM2-gangliosidosis 2** (Hexosaminidase B - HEXB)
- ○ **Schindler disease** (α -N-Acetylgalactosaminidase - NAGA)
- ○ **Tay-Sachs disease, GM2-gangliosidosis** (Hexosaminidase A - HEXA)

Various

- ○ **Alpha-1-Antitrypsin deficiency** (SERPINA 1)
- ○ **Angioedema, hereditary, type I/II; C1-INH deficiency** (SERPING1)
- ○ **Angioedema, hereditary, type III** (F12)
- ○ **Angiotensin I-Converting Enzyme IVS16 polymorphism** (ACE)
- ○ **Apolipoprotein C2 deficiency** (APOC2)
- ○ **Apolipoprotein E** (APOE)
- ○ **Berardinelli-Seip congenital lipodystrophy type 1** (AGPAT2)
- ○ **Berardinelli-Seip congenital lipodystrophy type 2** (BSCL2)
- ○ **Cystathionine β-synthase deficiency** (CBS)
- ○ **Cystinosis** (CTNS)
- ○ **Cystic fibrosis (CF, mucoviscidosis)** (CFTR)
- ○ **Dubin-Johnson syndrome** (ABCC2)
- ○ **Fructose intolerance, hereditary** (ALDOB)
- ○ **Galactosemia** (GALT)
- ○ **Galactokinase deficiency** (GALK1)
- ○ **Glucose transport defect; GLUT1 deficiency** (SLC2A1)
- ○ **Glycine encephalopathy** (GLDC)
- ○ **Homocystinuria, B12-responsive, cbl G** (MTR)
- ○ **Hypercholesterinemia, familial, LDL-receptor deficiency** (LDLR)
- ○ **Hypercholesterinemia type B** (APOB)
- ○ **Lipoprotein lipase deficiency** (LPL)
- ○ **Pancreatitis, hereditary** (SPINK1, PRSS1, CFTR)
- ○ **Phenylketonuria/hyperphenylalaninemia** (PAH)
- ○ **Hyperphenylalaninemia, BH4-deficient, type A** (PTS)
- ○ **Hyperphenylalaninemia, BH4-deficient, type B** (GCH1)
- ○ **Hyperphenylalaninemia, BH4-deficient, type C** (QDPR)
- ○ **Hyperphenylalaninemia, BH4-deficient, type D** (PCBD1)
- ○ **Shwachman-Diamond syndrome** (SBDS)

Fever syndromes, hereditary

- ○ **Familial Mediterranean fever** (MEFV)
- ○ **Hyper-IgD syndrome** (MVK)
- ○ **Periodic fever** (TNFRSF1A)
- ○ **Cryopyrinopathy** (NALP3)

Nephrology

- ○ **Alport syndrome, AR** (COL4A3, COL4A4)
- ○ **Alport syndrome, X-linked** (COL4A5)
- ○ **Bartter syndrome type 3** (CLCNKB)
- ○ **Cystinuria** (SLC3A1, SLC7A9)
- ○ **Epstein syndrome** (MYH9)
- ○ **Gitelman syndrome** (SLC12A3)
- ○ **Lowe syndrome** (OCRL)
- ○ **Meckel syndrome** (MKS1)
- ○ **Nephrotic syndrome, Finnish type** (NPHS1)
- ○ **Nephrotic syndrome, steroid resistent** (NPHS2)
- ○ **Polycystic kidney disease, AD** (PKD1, PKD2)
- ○ **Polycystic disease, AR** (PKHD1)

Cardiomyopathy, hereditary

- ○ **Dilatative cardiomyopathy screen** (LMNA>MYH7>TNNT2>SCN5A)
- ○ **Hypertrophic cardiomyopathy screen** (MYH7>MYBPC3>TNNT2>TNNI3>TPM1>MYL3)

Bone / Connective tissue diseases

- ○ **Achondroplasia** (FGFR3)
- ○ **Apert/Crouzon/Pfeiffer syndrome** (FGFR2)
- ○ **Arthrogryposis type 1** (TPM2)
- ○ **Arthrogryposis type 2A, Freeman-Sheldon syndrome** (MYH3)
- ○ **Arthrogryposis type 2B, Freeman-Hall syndrome** (TNNT3, TNNI2, MYH3)
- ○ **DTDST-associated dysplasia (ACG1B, AO2, DTD)** (SLC26A2)
- ○ **Hypocalciuric hypercalcemia** (CASR)
- ○ **Hypophosphatasia** (ALPL)
- ○ **Hypophosphatemic rickets, autosomal dominant** (FGF23)
- ○ **Hypophosphatemic rickets, X-linked** (PHEX)
- ○ **Marfan syndrome** (FBN1)
- ○ **Noonan syndrome, type:**
  ○ **1** (PTPN11), ○ **3** (KRAS), ○ 4 (SOS1), ○ 5 (RAF1)
- ○ **Osteogenesis imperfecta** (COL1A1, COL1A2)
- ○ **Smith-Lemli-Opitz syndrome** (DHCR7)
- ○ **Stickler syndrome type 1** (COL2A1)
- ○ **Stickler syndrome type 2** (COL11A1)

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Neurogenetics

Spastic paraplegia (HSP)

Pure/Complicated
- **SPG2 [X-linked]** (PLP1)
- **SPG3A [AD]** (Atlastin, SPG3A)
- **SPG4 [AD]** (Spastin)
- **SPG7 [AR]** (Paraplegin, SPG7)
- **SPG10 [AD]** (KIF5A)
- **SPG11 [AR]** (KIAA1840)
- **SPG15 [AR]** (ZFYVE26)

Pure
- **SPG5A [AR]** (CYP7B1)
- **SPG6 [AD]** (NIPA1)
- **SPG8 [AD]** KIAA0196)
- **SPG13 [AD]** (HSPD1)
- **SPG31 [AD]** (REEP1)

Complicated
- **SPG1 [X-linked]** (L1CAM)
- **SPG15 [AR]** (ZFYVE26)
- **SPG17 [AD]** (BSCL2)
- **SPG20 [AR]** (Spartin, SPG20)
- **SPG21 [AR]** (ACP33)

Motor and sensory neuropathy (HMSN/CMT)

- **CMT (most common forms**: PMP22, MPZ, GJB1 (~90% CMT1)

Demyelinating
- **CMT1A/HNPP [AD] Del./dup. [MLPA]** (PMP22)
- **CMT1A/HNPP [AD] sequencing** (PMP22)
- **CMT1B [AD]** (MPZ)
- **CMT1C [AD]** (LITAF)
- **CMT1D [AD]** (EGR2)
- **CMT1F [AD]** (NEFL)
- **CMT4A [AR]** (GDAP1)
- **CMT4B1 [AR]** (MTMR2)
- **CMT4B2 [AR]** (SBF2)
- **CMT4C [AR]** (SH3TC2)
- **CMT4D [AR]** (NDRG1)
- **CMT4F [AR]** (PRX)
- **CMTX1 [X-linked]** (GJB1)
- **Dejerine-Sottas syndrome** (MPZ, PMP22, PRX, EGR2)

Axonal
- **CMT2A1 [AD]** (KIF1B)
- **CMT2A2 [AD]** (MFN2)
- **CMT2B [AD]** (RAB7A)
- **CMT2B1 [AR]** (LMNA)
- **CMT2D [AD]** (GARS)
- **CMT2E ([AD]** (NEFL)
- **CMT2F [AD]** (HSPB1)
- **CMT2I/2J [AD]** (MPZ)
- **CMT2K [AR]** (GDAP1)
- **CMT2L [AD]** (HSPB8)
- **CMTX1 [X-linked]** (GJB1)
- **CMTX5 [X-linked]** (PRPS1)

Intermediate
- **DI-CMT B [AD]** (DNM2)
- **DI-CMT C [AD]** (YARS)
- **DI-CMT D [AD]** (MPZ)
- **CMTX1 [X-linked]** (GJB1)

Other neuropathies

Motor neuropathy
- **HMN2A** (HSPB8)
- **HMN2B** (HSPB1)
- **HMN5** (GARS)
- **HMN6 / DSMA1** (IGHMBP2)
- **HMN7B** (DCTN1)

Autonomic neuropathy
- **HSAN1** (SPTLC1)
- **HSAN2** (HSN2)
- **HSAN3** (IKBKAP)
- **HSAN4** (NTRK1)
- **HSAN5** (NGFB)

Other
- **Hereditary neuralgic amyotrophy** (SEPT9)

Dystonia

- **Primary torsion dystonia, DYT1** (TOR1A)
- **DOPA-resp. dystonia, AD, DYT5A** (GCH1)
- **DOPA-resp. dystonia, AR, DYT5B** (TH)
- **Torsion dystonia 6, DYT6** (THAP1)
- **Paroxysmal nonkinesigenic dyskinesia, DYT8** (MR1)
- **Myoclonus dystonia, DYT11** (SGCE)
- **Dystonia 12, DYT12** (ATP1A3)
- **Dystonia 16, DYT16** (PRKRA)
- **Dystonia 18, DYT18** (SLC2A1)

Neurodegenerative disease

- **Alzheimer, familial, FAD1** (APP)
- **Alzheimer, familial, FAD 3** (PSEN1)
- **Alzheimer, familial, FAD 4** (PSEN2)
- **Amyotrophic lateral sclerosis 1, ALS1** (SOD1)
- **Amyotrophic lateral sclerosis 2,** (ALS2)
- **Ataxia-oculomotor apraxia 1, AOA1**, (APTX)
- **Ataxia-oculomotor apraxie 2, AOA2**, (SETX)
- **Ataxia telangiectasia** (ATM)
- **Duchenne/Becker muscular dystrophy** (DMD)
- **Friedreich ataxia, FRDA** (FXN1)
- **Fronto-temporal dementia** (MAPT, GRN)
- **Huntington disease** (HD)
- **Huntington disease-like 1** (PRNP)
- **Huntington disease-like 2** (JPH3)
- **Infantile neuroaxonale dystrophy** (PLA2G6)
- **Kennedy disease, SBMA** (AR)
- **Muscular dystrophy, congenital, 1C** (FKRP)
- **Myotonic dystrophy 1, DM1** (DMPK)
- **Panthotenat kinase-assoc. neurodegen.** (PANK2)
- **Parkinson, familial, type 1/4** (SNCA)
- **Parkinson, familial, type 2** (PARK2)
- **Parkinson, familial, type 6** (PINK1)
- **Parkinson, familial, type 7** (DJ1)
- **Parkinson, familial, type 8** (LRRK2)
- **Parkinson, familial, type 9** (ATP13A2)
- **Prion disease (Creutzfeldt-Jakob, Gerstmann-Straussler, Fatal familial insomnia)** (PRNP)
- **Rett syndrome** (MECP2)
- **Spinal muscular atrophy 1/2/3/4 [MLPA]** (SMN1)
- **Spinal muscular atrophy 1/2/3/4 seq.** (SMN1)
- **SCA – most common forms (SCA1, 2, 3, 6)**
- **SCA5** (SPTBN2)
- **SCA7** (ATXN7)
- **SCA8** (SCA8)
- **SCA10** (ATXN10)
- **SCA12** (PPP2R2B)
- **SCA13** (KCNC3)
- **SCA14** (PRKCG)
- **SCA17** (TBP)
- **SCA27** (FGF14)
- **SCA31** (PLEKHG4)

Leukodystrophy & vascular disease

- **Adrenoleukodystrophy** (ABCD1)
- **Aicardi-Goutieres syndrome** (TREX1)
- **Alexander disease** (GFAP)
- **CADASIL** (NOTCH3)
- **Canavan disease** (ASPA)
- **Cerebrotendinous xanthomatosis** (CYP27A1)
- **Leukoencephalopathy with brainstem and spinal cord involvement and elevated lactate** (DARS2)
- **Leukoenceph. w. vanishing white matter** (EIF2B1–5)
- **Megalencephalic leukoencephalopathy with subcortical cysts** (MLC1)
- **Pelizaeus-Merzbacher disease (PMD)** (PLP1)
- **PMD-like 1** (GJC2)
- **Peroxisomal acyl-CoA oxidase deficiency** (ACOX1)
- **Wolman disease** (LIPA)

Mitochondriopathy

- **Leber optic neuropathy, hereditary**
- **MELAS syndrome**
- **MERFF syndrome**

Please inquire about analyses not listed on this form – we are continually developing new tests to complement our analysis spectrum.