Patient data (please fill out clearly in **block letters**)

Family name

First name

Date of birth

Day Month Year

Id. No.

Age

☐ male
☐ female

160065170067

Request form

MOLECULAR GENETIC ANALYSES
(DNA Analyses)

Client data

Physician

BIOSCIENTIA
HUMAN GENETICS

Center for
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Ingelheim

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Sample type

☐ EDTA blood
☐ DNA
☐ Amniotic fluid
☐ Chorionic villi
☐ Umbilical cord blood
☐ others, please specify

Number of tubes sent

Sampling date

Indication - Clinical data

Please note that detailed pedigree and clinical information is important for interpretation of results. Please provide the following information:

Parental consanguinity ☐ yes ☐ no
Patient/proband is affected ☐ yes ☐ no
Family members affected ☐ yes ☐ no if yes, who

Clinical data (medical history, report copies welcome)

Ethnic origin

☐ Caucasian
☐ Middle East
☐ Asia
☐ Ashkenazi/Jewish
☐ French Canadian/Acadian
☐ Finnish
☐ Other

In our lab we offer different testing panels by Next-Generation Sequencing (NGS). The spectrum covered by these panels is being extended constantly.
For further information, please contact us (+49 6132-781-433).

Declaration of Informed Consent

With my signature, I declare that I was briefed on

_____._____._____

_____
(physician)

about the nature, importance and implications of the genetic test and that I give my consent to the following genetic analysis and to the collection of the blood or tissue samples needed for this purpose:

_____

I consent to the storage, in accordance with legal requirements, of the recorded data in paper and/or electronic form and to their use and/or publication in pseudoanonymized form for scientific purposes for quality assurance.

I agree that my test results will not be destroyed after 10 years (as is requested by law) to allow my family access to them in the event of my death.
I consent to the results of the tests being made available to the following persons in addition to the doctor who submitted them:

_____

I hereby agree to the transfer, in accordance with § 950 BGB, of any test material remaining at the end of the analysis to the laboratory that carried out the analysis and I consent to its use for scientific purposes in pseudoanonymized form.
I consent to the communication of my data to a medical billing clearing house for invoicing purposes.
I am aware that I may withdraw this consent at any time, verbally or in writing, without giving reasons and without this having any adverse consequences for me.

- Please delete as appropriate -

Place, date:

_____

Name of patient / legal representative:

_____

Signature of patient / legal representative:

_____

specimen material | specimen material | specimen material | specimen material | specimen material | specimen material

This label should be stuck onto the attached copy and kept for your records.

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☐ **Array-CGH** (specific form available)

NEXT-GENERATION SEQUENCING (NGS) PANELS

Eye (retinal) panels

- ☐ Leber congenital amaurosis (LCA)
- ☐ Retinitis pigmentosa, autosomal dominant
- ☐ Retinitis pigmentosa, autosomal recessive
- ☐ Cone-rod dystrophy
- ☐ Senior-Loken syndrome

Deafness panels

- ☐ Deafness, autosomal dominant
- ☐ Deafness, autosomal recessive
- ☐ Deafness, X-linked
- ☐ Usher syndrome
- ☐ Jervell- and Lange-Nielsen syndrome, SANDD syndrome

Kidney/Liver panels

- ☐ Polycystic kidney diseases (ADPKD/ARPKD)
- ☐ Nephronophthisis (NPHP)/ UMOD-related disorders
- ☐ Senior-Loken syndrome
- ☐ Renal hypo-/dysplasia/agenesis/CAKUT
- ☐ Renal tubular dysgenesis (RTD)
- ☐ Nephrotic syndrome (SRNS)
- ☐ Focal segmental glomerulosclerosis (FSGS)
- ☐ Alport syndrome
- ☐ Hemolytic uremic syndrome (aHUS)/ DDD/MPGN
- ☐ Polycystic liver diseases (PCLD)

Skeletal/Connective tissue panels

- ☐ Marfan syndrome and related diseases
- ☐ Ehlers-Danlos syndrome
- ☐ Osteogenesis imperfecta
- ☐ Osteopetrosis, autosomal recessive
- ☐ Filamin diseases

Other ciliopathy panels

- ☐ Bardet-Biedl syndrome/ Alstrom syndrome
- ☐ Joubert syndrome
- ☐ Meckel-Gruber syndrome
- ☐ Jeune syndrome (ATD)
- ☐ Ellis-van-Crefeld syndrome (EVC)
- ☐ Sensenbrenner syndrome (CED)
- ☐ Filamin diseases
- ☐ Primary ciliary dyskinesia (PCD)/ Kartagener syndrome

Other NGS-panels

- ☐ Neurofibromatosis (NF)
- ☐ Tuberous Sclerosis (TSC)
- ☐ Alport syndrome
- ☐ MODY diabetes

All genes analysed by NGS can still be examined by conventional Sanger-sequencing (see below).

MOLECULAR GENETIC ANALYSES (in alphabetical order)

A	*Gene(s)*
☐ Achondrogenesis, type 2	*COL2A1*
☐ Achondroplasia	*FGFR3*
☐ Acromesomelic dysplasia, type Grebe	*CDMP1*
☐ Acromesomelic dysplasia, type Maroteaux	*NPR2*
☐ Acute lymphoblastic leukemia* (ALL)	
☐ Acute myeloid leukemia* (AML)	
☐ ADMCKD (Autosomal dominant medullary cystic kidney disease)	*UMOD*
☐ ADPKD (see polycystic kidney disease)	*PKD1, PKD2*
☐ Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency	*CYP21A2*
☐ Adrenoleukodystrophy	*ABCD1*
☐ Afibrinogenemia, congenital*	*FGA, FGB, FGG*
☐ Agammaglobulinemia, X-linked*	*BTK*
☐ Alagille syndrome	*JAG1*
☐ Albright syndrome*	*GNAS*
☐ Alexander disease*	*GFAP*
☐ Alkaptonuria*	*HGD*
☐ Alpha-1 antitrypsin deficiency	*SERPINA1*
☐ Alpha galactosidase A deficiency (Fabry disease)	*GLA*
☐ Alpha thalassemia*	*HBA*
☐ Alport-like syndrome (Epstein/Fechtner)	*MYH9*
☐ Alport syndrome	
☐ X-linked	*COL4A5*
☐ autosomal recessive, autosomal dominant	*COL4A3, COL4A4*
☐ Alstrom syndrome	*ALMS1*
☐ Alzheimer disease*	
☐ familial, type 1	*APP*
☐ familial, type 3	*PSEN1*
☐ Andersen-Tawil syndrome	*KCNJ2*
☐ Androgen insensitivity syndrome*	*AR*
☐ Angelman syndrome	
☐ methylation analysis SNRPN	*SNRPN*
☐ microsatellite analysis UPD(15)pat	
☐ sequence analysis UBE3A	*UBE3A*
☐ Angiotensin I converting enzyme	*ACE*
☐ Aniridia	*PAX6*
☐ Anonychia	*RSPO4*
☐ Antley-Bixler syndrome*	
☐ Apert syndrome (see Craniosynostosis)	
☐ Apolipoprotein B	*APOB*
☐ Apolipoprotein E	*APOE*
☐ Apparent mineralcorticoid excess	*HSD11B2*
☐ Arginase deficiency*	*ARG1*
☐ Argininosuccinate lyase deficiency*	*ASL*
☐ Argininosuccinate synthetase deficiency*	*ASS1*
☐ Aristaless-gene-related disorders*	*ARX*
☐ Aromatic L-aminoacid decarboxylase deficiency*	*DDC*
☐ ARPKD (see polycystisc kidney disease)	*PKHD1*
☐ Arthrogryposis multiplex congenita*	*TPM2, TNNI2*
☐ Arylsulfatase A deficiency*	*ARSA*
☐ Ataxia, autosomal dominant[3]	*SCA1, SCA2, SCA3, SCA6, SCA7, SCA17*
☐ Ataxia with isolated Vitamin E deficiency*	*TTPA*
☐ Ataxia with oculomotor apraxia 1*	*APTX*
☐ Ataxia teleangiectasia*	*ATM*
☐ Atypical PKU* (Tetrahydropterin, BH4-Mangel)	
☐ Auditory neuropathy	
☐ Autoimmune lymphoproliferative syndrome type 1A*	*TNFRSF6*
☐ Autoimmune polyendocrine syndrome type 1*	*AIRE*
☐ Azoospermia*	*USP9Y*

B	*Gene(s)*
☐ Bannayan-Riley-Ruvalcaba syndrome*	*PTEN*
☐ Bardet-Biedl syndrome[1,2,3]	
☐ BCR/ABL (chronic myeloid leukemia)	*BCR/ABL*
☐ Beckwith-Wiedemann syndrome	
☐ methylation analysis KvDMR1 and H19-DMR	
☐ microsatellite analysis UPD(11)pat	
☐ sequence analysis CDKN1C	*CDKN1C*
☐ Beta thalassemia*	*HBB*
☐ Bloch-Sulzberger syndrome* (Incontinentia pigmenti, IP2)	*IKBKG*
☐ Bloom syndrome* (Sister Chromatid Exchange (SCE))	*RECQL3*
☐ BOR syndrome[1,3]	
☐ Brachydactyly type C	*CDMP1*
☐ Breast/Ovarian cancer*	*BRCA1, BRCA2*
☐ Brugada syndrome	*SCN5A*
☐ Burkitt lymphoma*	*MYC*
☐ Butyrylcholinesterase deficiency*	*BCHE*
☐ Byler disease	*ATP8B1*

C	*Gene(s)*
☐ CADASIL	*NOTCH3*
☐ Campomelic dysplasia*	*SOX9*
☐ Canavan syndrome*	*ASPA*
☐ Carnitine deficiency*, systemic primary	*SLC22A5*
☐ Carnitine palmitoyl transferase I deficiency*	*CPT1*
☐ Carnitine palmitoyl transferase II deficiency*	*CPT2*
☐ Catechol-O methyltransferase activity	*COMT*
☐ Ceroid lipofuscinosis, neural (all types)	
☐ CHARGE syndrome	*CHD7*
☐ Chloride diarrhea, familial*	*SLC26A3*
☐ Chondrodysplasia type Grebe	*CDMP1*
☐ Chorea Huntington*	*HTT*
☐ Chronic lymphatic leukemia (CLL)	*IgVH*
☐ Ciliary dyskinesia, primary	
☐	*DNAH5*
☐	*DNAI1*
☐ CINCA syndrome	*NLRP3*
☐ Cleidocranial dysplasia	*RUNX2*
☐ Clubbing of digits	*HPGD*
☐ COACH syndrome (see Joubert syndrome)	
☐ Coats disease*	*NDP*
☐ Cockayne syndrome*	*ERCC8, ERCC6*
☐ Colon carcinoma	
☐ non polyposis associated, familial (HNPCC)*	*MLH1, MSH2*
☐ poliposis coli, familial adenomatous	*APC, MUTYH*
☐ Congenital disorder of glycosylation type 1a*	*PMM2*
☐ Corneal dystrophy	*DCN*
☐ Cornelia-De-Lange syndrome*	*NIPBL*
☐ CORS syndrome (see Joubert syndrome)	
☐ Costello syndrome*	*HRAS*
☐ Coumarin resistance	*VKORC1*
☐ Cowden syndrome*	*PTEN*
☐ Cranioectodermal dysplasia	
☐ Craniosynostosis, FGFR associated	*FGFR1, FGFR2, FGFR3*
☐ Crigler-Najjar syndrome	*UGT1A1*
☐ Crouzon syndrome (see Craniosynostosis)	
☐ Cystinosis*	*CTNS*
☐ Cystinuria*	
☐ Cystic fibrosis[3]	*CFTR*
☐ most common mutations with OLA	
☐ Middle-East-Panel	
☐ Cystic kidney disease (see polycystic kidney disease)	
☐ Cytochrom P450 defects	

D	*Gene(s)*
☐ Deafness, autosomal recessive[1,2,3]	
☐ DNFB1	*GJB2/Cx26*
☐ DNFB1	*GJB6/Cx30*
☐ Pendred syndrome	*SLC26A4, FOXI1*
☐ Deafness, autosomal dominant[1,2,3]	
☐ Dentato-Rubro-Pallido-Luysiane atrophy*	*ATN1*
☐ Denys-Drash syndrome	*WT1*
☐ Desbuquois syndrome	*CANT1*

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000000000001

- Diabetes, (MODY)[3] Maturity Onset Diabetes of the Young
- Diabetes insipidus
- Diabetes mellitus, neonatal with congenital hypothyroidism — *GLIS3*
- Diamond-Blackfan anemia* — *RPS19*
- Dihydropteridine reductase deficiency* — *QDPR*
- DPD deficiency (Dihydropyrimidine dehydrogenase deficiency) — *DPYD*
- Dravet syndrome — *SCN1A*
- Dysautonomia, familial (Riley-Day syndrome) — *IKBKAP*
- Dyserythropoietic anemia with thrombocytopenia* — *GATA1*
- Dysfibrinogenemia, congenital[3] — *FGA, FGB, FGG*
- Dystonia (DOPA responsive) — *GCH1, TH*
- Dystonia (torsion 1, autosomal dominant) — *DYT1*

E — Gene(s)

- Ehlers-Danlos syndrome[1]
- Ectodermal dysplasia type 1* — *EDA*
- Ektrodactyly (EEC syndrome)* — *TP63*
- Ellis-van-Creveld syndrome — *EVC1, EVC2*
- Epilepsies
- Epidermolysis bullosa
- Ewing sarcoma, t(11;22) translocation* — *EWS*

F — Gene(s)

- Fabry disease — *GLA*
- Factor II deficiency — *FII*
- Factor V Leiden mutation — *FV*
- Factor V- and Factor VIII deficiency, combined — *LMAN1*
- Factor X deficiency — *FX*
- Familial mediterranean fever (FMF) — *MEFV*
- Fanconi anemia*
- Fatty acid oxidation disorder (acylcarnitine profile)*
- Feingold syndrome* — *MYCN*
- Fragile X syndrome — *FMR1*
- Fraser syndrome* — *FRAS1, FREM2*
- Frasier syndrome — *WT1*
- Friedreich ataxia* — *FXN*
- Fructose intolerance, hereditary — *ALDOB*
- Fructose-1,6-bisphosphatase-deficiency* — *FBP1*
- FSGS[1,3] (fokal segmental glomerulosclerosis)

G — Gene(s)

- Galactosemia* — *GALT*
- Gaucher disease* — *GBA*
- Glaucoma*
- Glomerulosclerosis, fokal segmental (FSGS)[1,3]
- Glucocorticoid deficiency — *MC2R, MRAP*
- Glucose-6-phosphatdehydrogenase-deficiency — *G6PD*
- Glucose transport defect* — *SLC2A1*
- Glutaric acidemia type 1* — *GCDH*
- Glutathione S-transferase defects*
- Glycerol kinase deficiency* — *GK*
- Glycogen storage diseases*[1]
- Granulomatosis, chronic*
- Greig syndrome — *GLI3*
- Gusher syndrome — *POU3F4*

H — Gene(s)

- Haemophilia A* — *FVIII*
- Haemophilia B* — *FX*
- Hallervorden-Spatz disease — *PANK2*
- Hemochromatosis — *HFE*
- Hemolytic uremic syndrome (HUS)[1,2,3]
- HIV susceptibility* — *CCR5, CCR2, CXCL12*
- HNF1-β disease (see RCAD) — *TCF/HNF1β*
- Holocarboxylase synthetase deficiency — *HLCS*
- Holoprosencephaly*
- Homocystinuria* — *CBS*
- HPA-1 genotyping* — *HPA1*
- Hydroxymethylbilane synthase* — *HMBS*
- Hyperaldosteronism, familial type 1* — *CYP11B1, CYP11B2*
- Hypercholesterinemia
- Hypereosinophilic syndrome* — *FIP1L1, PDGFRA*
- Hyperekplexia (KOK disease)* — *GLRA1*
- Hyper-IgD syndrome — *MVK*
- Hyper-IgM syndrome* — *CD40LG*
- Hyperinsulinism, congenital* — *ABCC8, KCNJ11, GLUD1*
- Hyperkalemic periodic paralysis — *SCN4A*
- Hyperoxaluria, primary
- Hyperthyroidism, familial, non autoimmune — *TSHR*
- Hypochondroplasia — *FGFR3*
- Hypokalemic periodic paralysis — *CACNA1A, SCN4A*
- Hypophosphatasia, infantile* — *ALPL*
- Hypophosphatemia*
  - autosomal dominant — *FGF23*
  - X-linked — *PHEX*
- Hypothyroidism — *TSHR*
- Hypoventilation syndrome — *PHOX2B*

I — Gene(s)

- Idiopathic short stature — *SHOX*
- Insulin-like growth factor 1 deficiency (IGF deficiency) — *IGF1*
- Immunodeficiency, severe, combined, autosomal recessive, T-cell neg., B-cell pos., NK-cells neg. — *JAK3*
- Ivemark syndrome[2] — *NPHP3*

J — Gene(s)

- Jervell- and Lange-Nielsen syndrome
- Jeune syndrome[1,2,3]
- Joubert syndrome[1,2,3]

K — Gene(s)

- Kabuki syndrome — *MLL2*
- Kallmann syndrome[1,3]
- Kartagener syndrome (see ciliary dyskinesia, primary)
- Kearns-Sayre syndrome* — *MTTL2*
- Kell incompatibility* — *KEL*
- Kniest dysplasia — *COL2A1*

L — Gene(s)

- Lafora syndrome — *EPM2A, NHLRC1*
- Lactose intolerance, congenital — *LCT*
- Langer, mesomelic dysplasia — *SHOX*
- Laron syndrome — *GHR*
- Larsen syndrome
  - — *FLNB*
  - — *CHST3*
- Lathosterolosis* — *SC5DL*
- Leber congenital amaurosis (LCA)[1,2,3]
- Leber hereditary opticus neuropathy (LHON) — *mtDNA*
- LEOPARD syndrome — *PTPN11*
- Leri-Weil syndrome — *SHOX*
- Lesh-Nyhan syndrome — *HPRT1*
- Leukocyte adhesion deficiency
  - type 1 — *ITGB2*
  - type 2 — *SLC35C1*
- Li-Fraumeni syndrome* — *p53*
- Liddle syndrome* — *SCNN1B, SCNN1G*
- Lissencephaly*
- Loeys-Dietz syndrome — *TGFBR1, TGFBR2*
- Long QT syndrome
  - Romano-Ward syndrome[3]
  - Jervell- and Lange-Nielsen syndrome
  - Andersen-Tawil syndrome
  - Timothy syndrome
- Lowe syndrome* — *OCRL1*
- Lymphohistiocytosis*, hemophagocytic, familial
- Lymphoma* — *BCL2*
- Lymphedema type 1* — *FLT4*

M — Gene(s)

- Mantle cell lymphoma* — *BCL1*
- Marfan syndrome
  - type 1 — *FBN1*
  - type 2 — *TGFBR1, TGFBR2*
- MASA syndrome* — *L1CAM*
- Meckel-Gruber syndrome[1,2,3]
- Mediterranean fever, familial (FMF) — *MEFV*
- Melanoma* — *CMM2, CDKN2A*
- MELAS syndrome*
- Menkes syndrome* — *ATP7A*
- MERRF syndrome*
- Metaphyseal chondrodysplasia (Schmid type) — *COL10A1*
  - hot spot analysis
  - complete gene analysis incl. hot spots
- Methylentetrahydrofulate reductase — *MTHFR*
- Methylmalonic aziduria* — *MUT*
- Meulengracht syndrome — *UGT1A1*
- Mikrosatellite analysis (maternal contamination of fetal specimen)
- Mitochondrial complex II deficiency* — *SDHA*
- Mitochondrial DNA depletion syndrome*
- Molybdenum cofactor deficiency*
- Morbus Crohn (CARD15 polymorphism) — *CARD15*
- Morbus Fabry — *GLA*
- Morbus Gaucher* — *GBA*
- Morbus Gilbert (UGT1A1 promotor, TA insertion) — *UGT1A1*
- Morbus Krabbe* — *GALC*
- Morbus Meulengracht — *UGT1A1*
- Morbus Osler*
- Morbus Pompe* — *GAA*
- Morbus Wilson — *ATP7B*
- MORM syndrome — *INPP5E*
- Muckle-Wells syndrome — *NLRP3*
- Muenke syndrome (see Craniosynostosis)
- Mucolipidosis* — *GNPTAB*
- Mucopolysaccharidosis*
- Multi drug resistance*
- Multiple exostosis* — *EXT1, EXT2*
- Muscular dystrophies
- Myasthenia syndrome*, congenital combined with AChR deficiency — *CHRNE, CHAT*
- Mycobacteriosis*, familial atypical — *IFNGR1, IFNGR2*
- Myeloproliferative disorders — *JAK2*
- Myoclonic epilepsy
  - severe of infancy — *SCN1A*
  - Unverricht-Lundborg — *CSTB*
- Myotonia congenita* — *CLCN1*
- Myotonic dystrophy
  - type 1 — *DMPK*
  - type 2 — *PROMM*
- Myotubular myopathy,* X-linked — *MTM1*

N — Gene(s)

- N-acetyl glutamate synthetase deficiency* — *NAGS*
- N-acetyltransferase 2 deficiency — *NAT2*
- Nail-Patella syndrome — *LXM1B*
- NARP syndrome* — *MTATP6*
- NDH syndrome — *GLIS3*
- Neoplasia, multiple endocrine
  - type 1* — *MEN1*
  - type 2A — *RET*
  - type 2B — *RET*
- Nephropathy, familial, juvenile, hyperuricemic — *UMOD*
- Nephronophthisis[1,2,3]
- Nephrotic syndrome[1,2,3]
- Netherton syndrome — *SPINK5*
- Neurofibromatosis
  - type 1 — *NF1*
  - type 2 — *NF2*
- Neuropathies[1,2,3]
- Neutropenia*, congenital, autosomal dominant — *ELA2*
- Nevoid basal cell carcinoma[3]*
- Nieman-Pick disease*
- Non small cell lung cancer* — *EGFR*
- Noonan syndrome — *PTPN11, SOS1*
- Norrie syndrome* — *NDP*

O — Gene(s)

- Obesity — *MC4R*
- Oculocutaneous albinism*
  - type 1A, type 1B — *TYR*
  - type 2 — *OCA2*
- Onedine syndrome — *PHOX2B*
- Ophthalmoplegia*
- Optic athropy — *OPA1*
- Ornithine transcarbamylase deficiency* — *OTC*
- Orofaciodigital syndrome 1 — *OFD1*
- Osteogenesis imperfecta
- Osteogenesis imperfecta, autosomal recessive

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- ☐ Osteopetrosis,* autosomal recessive
- ☐ Osteoporosis — *COL1A1, VDR*
- ☐ Otopalatodigital syndrome — *FLNA*
  - ☐ type 1 (OPD1)
  - ☐ type 2 (OPD2)
  - ☐ Melnick-Needles syndrome (MNS)
  - ☐ frontometaphyseal dysplasia (FMD)

P	*Gene(s)*
☐ PAI gene analysis	*PAI1*
☐ Pancreatitis, hereditary	*PRSS1, SPINK1, CFTR*
☐ Paramyotonia congenita, of von Eulenberg	*SCN4A*
☐ Parkinson disease*	
☐ Pelizaeus-Merzbacher disease*	*PLP*
☐ Peutz-Jeghers syndrome*	*STK11*
☐ Pfeiffer syndrome (see Craniosynostosis)	
☐ Phenylalanine hydroxylase deficiency*	*PAH*
☐ Pierson syndrome	*LAMB2*
☐ *PKD1/TSC 2* microdeletion syndrome	
☐ Polyposis coli, familial adenomatous (FAP)	*APC, MUTYH*
☐ Polysyndactyly*	*HOXD13*
☐ Polycystic kidney disease [1,2,3]	
☐ autosomal dominant (type 1)	*PKD1*
☐ autosomal dominant (type 2)	*PKD2*
☐ autosomal recessive	*PKHD1*
☐ Polycystic kidney disease, severe infantile, with tuberous sclerosis	*PKD1/TSC2*
☐ Pontocellebar hypoplasia type 1	*VRK1*
☐ Prader-Willi syndrome	*SNRPN*
☐ methylation analysis SNRPN gene	
☐ microsatellit analysis UPD(15)mat	
☐ Primary ciliary dyskinesia	
☐	*DNAH5*
☐	*DNAI1*
☐ Primary hypertrophic Osteoarthropathy (Pachydermoperiostose/Tourraine-Solente-Gole syndrome)	*HPGD*
☐ Progressive familial intrahepatic cholestasis	
☐ Propionic acidemia*	*PCCA, PCCB*
☐ Protein-C deficiency*	*PROC*
☐ Proteus syndrome*	*PTEN*
☐ Protoporphyria, erythropoietic*	*FECH*
☐ Pseudoachondroplasia*	*COMP*
☐ Pyruvate carboxylase deficiency*	*PC*
☐ Pyruvate dehydrogenase deficiency*	*PDHA1*

R	*Gene(s)*
☐ RCAD (Renal Cyst and Diabetes syndrome)	*TCF/HNF1β*
☐ Renal agenesis/dysplasia	*TCF/HNF1β, RET*
☐ Renal cell carcinoma	*MET*
☐ Renal-Coloboma syndrome	*PAX2*
☐ Renal cysts (see polycystic kidney disease)	
☐ Renal-tubular dysgenesia [1,2,3]	
☐ Retinitis pigmentosa [1,2,3]	
☐ autosomal dominant	
☐ autosomal recessive	
☐ Retinoblastoma	*RB1*
☐ Retinoschisis	*RS1*
☐ Rett syndrome	*MECP2*
☐ Rhesus-(Rh) compatibility*	
☐ RHCE	*RHCE*
☐ RHD	*RHD*
☐ Romano-Ward syndrome [3]	
☐ Rubinstein-Taybi syndrome*	*CREBBP*

S	*Gene(s)*
☐ Sandhoff disease	*HEXB*
☐ Schinzel-Giedeon syndrome	*SETBP1*
☐ Senior-Loken syndrome [1,2,3]	
☐ Sensenbrenner syndrome	
☐ Septooptic dysplasia*	*HESX1*
☐ SESAME syndrome	*KCNJ10*
☐ Shwachman-Diamond syndrome*	*SBDS*
☐ Sialuria*	*SLC17A5*
☐ Sickle cell anemia	*HBB*
☐ Silver-Rusell syndrome	
☐ methylation analysis KvDMR1 and H19DMR	
☐ microsatellite analysis UDP(7)mat	
☐ Sjögren-Larsson syndrome*	*ALDH3A2*
☐ Smith-Lemli-Opitz syndrome*	*DHCR7*
☐ Sotos syndrome	*NSD1*
☐ Spastic paraplegia [3]	
☐ Spinal bulbar muscular atrophy*, type Kennedy	*AR*
☐ Spinal muscular atrophy (SMA)	*SMN1*
☐ Spinocerebellar ataxia [3]	
☐ Spondylocostal dysostosis*	*DLL3*
☐ Spondyloepiphyseal dysplasia	*COL2A1*
☐ Spondyloepiphyseal dysplasia, X-linked	*SEDL*
☐ Sterility (Congenital aplasia of vas deferens)	*CFTR*
☐ Stickler syndrome type 1	*COL2A1*
☐ Succinate dehydrogenase deficiency*	*SDHB, SDHD*
☐ Surfacant dysfunction	
☐ pulmonary 1	*SFTPB*
☐ pulmonary 3*	*ABCA3*
☐ pulmonary 4*	*CSF2RB*

T	*Gene(s)*
☐ Tay-Sachs disease	
☐ Jewish sreening	*HEXA*
☐ complete gene	*HEXA*
☐ *HEXA and HEXB* genes	*HEXA, HEXB*
☐ T-cell receptor-γ rearrangement*	
☐ Thanatophoric dysplasia	*FGFR3*
☐ Thiopurine methyltransferase deficiency	*TPMT*
☐ Thrombophilia	*MTHFR*
☐ Thrombotic thrombocytopenic purpura (TTP)	*ADAMTS13*
☐ Thyroid hormone resistance	*THR-beta*
☐ Timothy syndrome	*CACNA1C*
☐ TNF receptor associated periodic fever	*TNFRSF1A*
☐ Treacher-Collins-Franceschetti syndrome*	*TCOF1*
☐ Trimethylaminuria*	*FMO3*
☐ *TSC2/PKD1* microdeletion syndrome	
☐ Tuberous sclerosis	
☐ type 1	*TSC1*
☐ type 2	*TSC2*
☐ Tyrosinemia*	*CD40LG*

U	*Gene(s)*
☐ UMOD disease (see ADMCKD)	*UMOD*
☐ Uniparental disomy of chromosome: [3] 2, 4, 6, 7, 11, 13, 14, 15, 16	
☐ Usher syndrome [1,2,3]	
☐ type 1	
☐ type 2	
☐ USH1: FC-/Acadian Panel	
☐ USH2: FC-/Acadian Panel	

V	*Gene(s)*
☐ Very long chain acyl-CoA dehydrogenase deficiency*	*ACADVL*
☐ Vitamin D resistant rachitis*	
☐ Vitreoretinopathy, familial exsudative*	*TSPAN12, NDP*
☐ Von Hippel-Lindau syndrome	*VHL*
☐ von Willebrand disease*	*VWF*

W	*Gene(s)*
☐ Waardenburg syndrome [2,3]	
☐ Warfarin or Coumarin sensitivity*	*VKORC1*
☐ Weaver syndrome	*NSD1*
☐ Werner syndrome	*RECQL2*
☐ Wilms tumor	*WT1*
☐ Wiskott-Aldrich syndrome*	*WAS*
☐ Wolfram syndrome type 2	*CISD2*

X	*Gene(s)*
☐ Xanthinuria	*XDH*
☐ X-linked hydrocephalus*	*L1CAM*
☐ X-linked lymphproliferative syndrome*	*SH2D1A*
☐ X-linked sideroblastic anemia*	*ALAS2*
☐ X-linked sideroblastic anemia and Ataxia*	*ABCB7*

Others

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MOLECULAR GENETIC ANALYSES (by disease groups)

Blood disorders — Gene(s)

- Afibrinogenemia, congenital* — FGA, FGB, FGG
- Alpha thalassemia* — HBA
- Beta thalassemia* — HBB
- Diamond-Blackfan anemia* — RPS19
- Dyserythropoietic anemia with thrombocytopenia* — GATA1
- Dysfibrinogenemia, congenital[3] — FGA, FGB,FGG
- Factor II deficiency — FII
- Factor V-Leiden mutation — FV
- Factor V- and Factor VIII deficiency, combined — LMAN1
- Factor X deficiency — FX
- Haemophilia A* — FVIII
- Haemophilia B* — FX
- HPA-1-genotyping* — HPA1
- Kell incompatibility* — KEL
- Neutropenia*, congenital, autosomal dominant — ELA2
- PAI gene analysis — PAI1
- Rhesus (Rh) incompatibility*
  - RHD — RHD
  - RHCE — RHCE
- Sickle cell anemia — HBB
- Thrombophilia — METHFR
- Thrombotic thrombocytopenic purpura (TTP) — ADAMTS13
- von Willebrand disease* — VWF
- Warfarin or Coumarin sensitivity* — VKORC1
- Wiskott-Aldrich syndrome* — WAS
- X-linked sideroblastic anemia* — ALAS2
- X-linked sideroblastic anemia and Ataxia* — ABCB7

Cancer/Leukemia/Tumor — Gene(s)

- Acute lymphoblastic leukemia* (ALL)
- Acute myeloid leukemia* (AML)
- Ataxia teleangiectasia* — ATM
- Bannayan-Riley-Ruvalcaba syndrome* — PTEN
- BCR/ABL (chronic myeloid leukemia) — BCR/ABL
- Bloom syndrome* (Sister Chromatid Exchange (SCE)) — RECQL3
- Breast/Ovarian cancer* — BRCA1, BRCA2
- Burkitt lymphoma* — MYC
- Chronic lymphatic leukemia (CLL)* — IgVH
- Colon carcinoma
  - non polyposis associated, familial (HNPCC)* — MLH1, MSH2
  - poliposis coli, familial adenomatous — APC, MUTYH
- Cowden syndrome* — PTEN
- Ewing sarcoma, t(11;22) translocation* — EWS
- Fanconi anemia*
- Li-Fraumeni syndrome* — p53
- Lymphoma* — BCL2
- Mantle cell lymphoma* — BCL1
- Melanoma* — CMM2, CDKN2A
- Myeloproliferative disorder — JAK2
- Neoplasia, multiple endocrine
  - type 1* — MEN1
  - type 2A — RET
  - type 2B — RET
- Neurofibromatosis
  - type 1 — NF1
  - type 2 — NF2
- Nevoid basal cell carcinoma[3]*
- Non small cell lung cancer* — EGFR
- Peutz-Jeghers syndrome* — STK11
- Proteus syndrome* — PTEN
- Renal cell carcinoma — MET
- Retinoblastoma — RB1
- Shwachman-Diamond syndrome* — SBDS
- Tuberous sclerosis
  - type 1 — TSC1
  - type 2 — TSC2
- Von Hippel-Lindau syndrome — VHL
- Wilms tumor — WT1
- X-linked lymphoproliferative syndrome* — SH2D1A

Cardiac disorders — Gene(s)

- Angiotensin I converting enzyme — ACE
- Brugada syndrome — SCN5A
- Long QT syndrome
  - Romano-Ward syndrome[3]
  - Jervell- and Lange-Nielsen syndrome
  - Andersen-Tawil syndrome
  - Timothy syndrome

Chromosome instability — Gene(s)

- Ataxia teleangiectasia* — ATM
- Bloom syndrome* (Sister Chromatid Exchange (SCE)) — RECQL3
- Fanconi anemia*

Connective tissue disorders — Gene(s)

- Alpha-1 antitrypsin deficiency — SERPINA1
- Ehlers-Danlos syndrome[1]
- Surfacant dysfunction
  - pulmonary 1 — SFTPB
  - pulmonary 3* — ABCA3
  - pulmonary 4* — CSF2RB

Deafness — Gene(s)

- Alstrom syndrome — ALMS1
- Auditory neuropathy
- BOR syndrome1,3
- Gusher syndrome — POU3F4
- Jervell- and Lange-Nielsen syndrome
- SANDD syndrome — CACNA1D
- SESAME syndrome — KCNJ10
- Deafness, autosomal recessive[1,2,3]
  - DNFB1 — GJB2/Cx26
  - DNFB1 — GJB6/Cx30
  - Pendred syndrome — SLC26A4, FOXI1
- Deafness, autosomal dominant[1,2,3]
- Usher syndrome[1,2,3]
- Waardenburg syndrome[2,3]

Dementia — Gene(s)

- Alzheimer disease*
  - familial, type 1 — APP
  - familial, type 3 — PSEN1
- Dentato-Rubro-Pallido-Luysiane atrophy* — ATN1

Endocrinology — Gene(s)

- Albright syndrome* — GNAS
- Androgen insensivity syndrome* — AR
- Apparent mineralcorticoid excess — HSD11B2
- Diabetes, (MODY)[3] Maturity Onset Diabetes of the Young
- Diabetes insipidus
- Diabetes mellitus, neonatal with congenital hypothyreodism — GLIS3
- Glucocorticoid deficiency — MC2R, MRAP
- Hyperinsulinism, congenital* — ABCC8, KCNJ11, GLUD1
- Hyperthyroidsm, familial, non autoimmune — TSHR
- Hypothyroidsm — TSHR
- Insulin like growth factor 1 deficiency (IGF deficiency) — IGF1
- Kallmann syndrome[1,3]
- Laron syndrome — GHR
- Lathosterolosis* — SC5DL
- Liddle syndrome — SCNN1B, SCNN1G
- NDH syndrome — GLIS3
- Shwachman-Diamond syndrome* — SBDS
- Thyroid hormone resistance — THR-beta
- Vitamin D resistent rachitis*
- Wolfram syndrome type 2 — CISD2

Epigenetic disorders — Gene(s)

- Angelman syndrome
  - methylation analysis *SNRPN* — SNRPN
  - microsatellite analysis UPD(15)pat
  - sequence analysis UBE3A — UBE3A
- Beckwith-Wiedemann syndrome
  - methylation analysis KvDMR1 and H19-DMR
  - microsatellite analysis UPD(11)pat
  - sequence analysis *CDKN1C* — CDKN1C
- Prader-Willi syndrome — SNRPN
  - methylation analysis *SNRPN gene*
  - microsatellit analysis UPD(15)mat
- Silver-Rusell syndrome
  - methylation analysis KvDMR1 and H19DMR
  - microsatellite analysis UDP(7)mat
- Uniparental disomy of chromosome:[3] 2, 4, 6, 7, 11, 13, 14, 15, 16

Epilepsy — Gene(s)

- Aristaless-gene-related disorders* — ARX
- Epilepsies
- Lafora syndrome — EPM2A, NHLRC1
- Myoclonic epilepsy
  - severe, of infancy — SCN1A
  - Unverricht-Lundborg — CSTB

Eye (retinal) disorders — Gene(s)

- Alstrom syndrome — ALMS1
- Aniridia — PAX6
- Bardet-Biedl syndrome[1,2,3]
- Coats disease* — NDP
- Congenital stationary night blindness
- Corneal dystrophy — DCN
- Glaukoma*
- Joubert syndrome[1,2,3]
- Leber congenital amaurosis (LCA)[1,2,3]
- Leber hereditary optic neuropathy (LHON) — mtDNA
- Lowe syndrome* — OCRL1
- Norrie syndrome* — NDP
- Oculocutaneous albinism*
  - type 1A, type 1B — TYP
  - type 2 — OCA2
- Ophthalmoplegia*
- Retinitis pigmentosa[1,2,3]
  - autosomal dominant
  - autosomal recessive
- Retinoschisis — RS1
- Usher syndrome[1,2,3] (see Usher syndrome)
- Vitreoretinopathy, familial exsudative* — TSPAN12. NDP

Gastroenterologic disorders — Gene(s)

- Alagille syndrome — JAG1
- Byler disease — ATP8B1
- Chloride diarrhea, familial* — SLC26A3
- Crigler-Najjar syndrome — UGT1A1
- Cystic fibrosis[3] — CFTR
  - most common mutations with OLA
  - Middle-East-Panel
- Morbus Crohn (CARD15 polymorphism) — CARD15
- Morbus Gilbert — UGT1A1
- Pancreatitis, hereditary — PRSS1, SPINK1, CFTR
- Peutz-Jeghers syndrome* — STK11
- Polyposis coli, familial adenomatous (FAP) — APC, MUTYH
- Progressive familial intrahepativ Cholestasis

Immune system disorders — Gene(s)

- Agammaglobulinemia, X-linked* — BTK
- Autoimmune lymphoproliferative syndrome type 1* — TNFRSF6
- Autoimmune polyendocrine syndrome type 1A* — AIRE
- Bloom syndrome* (Sister Chromatid Exchange (SCE)) — RECQL3
- Granulomatosis, chronic*
- Hypereosinophilic syndrome* — FIP1L1, PDGFRA
- Hyper-IgD syndrome — MVK
- Hyper-IgM syndrome* — CD40LG
- Immunodeficiency, severe, combined; autosomal recessive, T-cell neg., B-cell pos., NK-cells neg. — JAK3
- Leukocyte adhesion deficiency
  - type 1 — ITGB2
  - type 2 — SLC35C1

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- Lymphohistiocytosis*, hemophagocytic, familial
- Lymphoma*
- Muckle-Wells syndrome — *NLRP3*
- Neutropenia*, congenital, autosomal dominant — *ELA2*
- T-cell receptor-γ rearrangement*

Infection	Gene(s)
HIV susceptibility*	*CCR5, CCR2, CXCL12*
Mycobacteriosis*, familial atypical	*IFNGR1, INFGR2*

Infertility	Gene(s)
Azoospermia*	*USP9Y*
Cystic fibrosis[3]	*CFTR*
most common mutations with OLA	
Middle-East-Panel	
Kallmann syndrome[1,3]	
Kartagener syndrome (see primary ciliary dyskinesia)	
Primary ciliary dyskinesia	
	DNAH5
	DNAI1
Sterility (congenital aplasia of vas deferens)	*CFTR*

Inflammation	Gene(s)
CINCA syndrome	*NLRP3*
Familial mediterranean fever (FMF)	*MEFV*
Hyper-IgD syndrome	*MVK*
TNF receptor associated periodic fever	*TNFRSF1A*

Kidney disorders	Gene(s)
ADMCKD (autosomal dominant medullary cystic kidney disease)	*UMOD*
ADPKD (see polycystic kidney disease)	*PKD1, PKD2*
Alport-like syndrome (Epstein/Fechtner)	*MYH9*
Alport syndrome	
X-linked	*COL4A5*
autosomal recessive, autosomal dominant	*COL4A3, COL4A4*
Alstrom syndrome	*ALMS1*
ARPKD (see polycystic kidney disease)	*PKHD1*
Bardet-Biedl syndrome[1,2,3]	
BOR syndrome[1,3]	
Cystinosis*	*CTNS*
Denys-Drash syndrome	*WT1*
Diabetes insipidus	
Frasier syndrome	*WT1*
Glomerulosclerosis, fokal segmental (FSGS)[1,3]	
Hemolytic uremic syndrome (HUS)[1,3]	
HNF1-β disease (see RCAD)	*TCF/HNF1β*
Hyperoxaluria, primary	
Ivemark syndrome[2]	*NPHP3*
Joubert syndrome[1,2,3]	
Meckel-Gruber syndrome[1,2,3]	
Morbus Fabry	*GLA*
Nail-Patella syndrome	*LMX1B*
Nephropathy, familial, juvenile hyperuricemic	*UMOD*
Nephronophthisis[1,2,3]	
Nephrotic syndrome[1,2,3]	
Orofaciodigital syndrome 1	*OFD1*
PKD1/TSC 2 microdeletion syndrome	
Polycystic kidney disease[1,2,3]	
autosomal dominant (type 1)	*PKD1*
autosomal dominant (type 2)	*PKD2*
autosomal recessive	*PKHD1*
Polycystic kidney disease, severe infantile, with tuberous sclerosis	*PKD1/TSC2*
RCAD (Renal Cysts and Diabetes syndrome)	*TCF/HNF1β*
Renal agenesis/dysplasia	*TCF/HNF1β, RET*
Renal-Coloboma syndrome	*PAX2*
Renal cysts (see polycystic kidney disease)	
Renal-tubular dysgenesis[1,2,3]	
Senior-Loken syndrome[1,2,3]	
Sialuria*	*SLC17A5*
Tuberous sclerosis	
type 1	*TSC1*
type 2	*TSC2*
TSC2/PKD1 microdeletion syndrome	
UMOD disease (see ADMCKD)	*UMOD*

Makrosomia/Giantism	Gene(s)
Beckwith-Wiedemann syndrome	
methylation analysis KvDMR1 and H19-DMR	
microsatellite analysis UPD(11)pat	
sequence analysis CDKN1C	*CDKN1C*
Sotos syndrome	*NSD1*
Weaver syndrome	*NSD1*

Malformation syndromes	Gene(s)
Alagille syndrome	*JAG1*
Alstrom syndrome	*ALMS1*
Antley-Bixler syndrome*	
Bardet-Biedl syndrome[1,2,3]	
BOR syndrome[1,3]	
CHARGE syndrome	*CHD7*
COACH syndrome (see Joubert syndrome)	
Cornelia-De-Lange syndrome*	*NIPBL*
CORS syndrome (see Joubert syndrome)	
Costello syndrome*	*HRAS*
Denys-Drash syndrome	*WT1*
Ellis-van-Creveld syndrome	*EVC1, EVC2*
Feingold syndrome*	*MYCN*
Greig syndrome	*GLI3*
Ivemark syndrome[2]	*NPHP3*
Jeune syndrome[1,2,3]	
Joubert syndrome[1,2,3]	
LEOPARD syndrome	*PTPN11*
Meckel-Gruber syndrome[1,2,3]	
MORM syndrome	*INPP5E*
Orofaciodigital syndrome 1	*OFD1*
Schinzel-Giedeon syndrome	*SETBP1*
Smith-Lemli-Opitz syndrome*	*DHCR7*

Mental retardation	Gene(s)
Alexander disease*	*GFAP*
Angelman syndrome	
methylation analysis *SNRPN*	*SNRPN*
microsatellite analysis UPD(15)pat	
sequence analysis UBE3A	*UBE3A*
Antley-Bixler syndrome*	
Aristaless-gene-related disorders*	*ARX*
Canavan syndrome*	*ASPA*
Fragile X syndrome	*FMR1*
Fraser syndrome*	*FRAS1, FREM2*
Holoprosencephaly*	
Kabuki syndrome	*MLL2*
Lissencephaly*	
Rett syndrome	*MECP2*

Metabolic disorders	Gene(s)
Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency	*CYP21A2*
Alkaptonuria*	*HGD*
Alpha galactosidase deficiency (Morbus Fabry)	*GLA*
Apolipoprotein B	*APOB*
Apolipoprotein E	*APOE*
Arginase deficiency*	*ARG1*
Argininosuccinate lyase deficiency*	*ASL*
Argininosuccinate synthetase deficiency*	*ASS1*
Aromatic L-aminoacid decarboxylase deficiency*	*DDC*
Atypical PKU* (Tetrahydropterin, BH4 deficiency)	
Butyrylcholinesterase deficiency*	*BCHE*
Carnitine deficiency*, systemic primary	*SLC22A5*
Carnitine palmitoyl transferase I deficiency*	*CPT1*
Carnitine palmitoyl transferase II deficiency*	*CPT2*
Catechol-O methyltransferase activity	*COMT*
Congenital disorder of glycosylation type 1a*	*PMM2*
Dihydropteridine reductase deficiency	*QDPR*
DPD deficiency (Dihydropyrimidindehydrogenase deficiency)	*DPYD*
Fatty acid oxidation disorder (Acylcarnitine profile)*	
Fructose intolerance, hereditary	*ALDOB*
Fructose-1,6-bisphosphatase-deficiency*	*FMP1*
Glucose-6 phosphat dehydrogenase deficiency	*G6PD*
Glucose transport defect*	*SLC2A1*
Glutaric acidemia type 1*	*GCDH*
Glutathione S-transferase defects*	
Glycerol kinase deficiency*	*GK*
Holocarboxylase synthetase deficiency*	*HLCS*
Homocystinuria*	*CBS*
Hydroxymethylbilane synthase*	*HMBS*
Hyperaldosteronism, familial type 1*	*CYP11B1, CYP11B2*
Hypercholesterinemia	
Lactose intolerance, congenital	*LCT*
Methylentetrahydrofulate reductase	*MTHFR*
Methylmalonic aciduria*	*MUT*
Meulengracht syndrome	*UGT1A1*
Molybdenum cofactor deficiency*	
Morbus Gaucher*	*GBA*
Morbus Gilbert (UGT1A1promotor, TA insertion)	*UGT1A1*
Morbus Krabbe*	*GALC*
Morbus Meulengracht	*UGT1A1*
Morbus Osler*	
N-acetyl glutamate synthetase deficiency*	*NAGS*
Obesity	*MC4R*
Ornithine transcarbamylase deficiency*	*OTC*
Phenylalanine hydroxylase deficiency*	*PAH*
Propionic acidemia*	*PCCA, PCCB*
Protoporphyria, erythropoietic*	*FECH*
Pyruvate carboxylase deficiency*	*PC*
Pyruvate dehydrogenase deficiency*	*PDHA1*
Sialuria*	*SLC17A5*
Smith-Lemli-Opitz syndrome*	*DHCR7*
Succinate dehydrogenase deficiency*	*SDHB, SDHD*
Thiopurine methyltransferase deficiency*	*TPMT*
Trimethylaminuria*	*FMO3*
Tyrosinemia*	*CD40LG*
Very long chain acyl-CoA dehydrogenase deficiency*	*ACADVL*
Xanthinuria	*XDH*

Mitochondriopathies	Gene(s)
Kearns-Sayre syndrome*	*MTTL2*
Leber hereditary opticus neuropathy (LHON)	*mtDNA*
MELAS syndrome*	
MERRF syndrome*	
Mitochondrial complex II deficiency*	*SDHA*
Mitochondrial DNA depletion syndrome*	

Neuromuscular disorders	Gene(s)
Muscular dystrophies	
Myotonia congenita*	*CLCN1*
Myotonic dystrophy	
type 1	*DMPK*
type 2	*PROMM*
Myotubular myopathy,* X-linked	*MTM1*
Paramyotonia congenita, of von Eulenberg	*SCN4A*

Neurology/Neuropathies	Gene(s)
Ataxia, autosomal dominant[3]	*SCA1, SCA2, SCA3, SCA6, SCA7, SCA17*
Ataxia with isolated Vitamin E deficiency*	*TPPA*
Ataxia with oculomotor apraxia 1*	*APTX*
Ataxia teleangiectasia*	*ATM*
Chorea Huntington*	*HTT*
Dentato-Rubro-Pallido-Luysiane atrophy*	*ATN1*

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- ☐ Dysautonomia, familial (Riley-Day syndrome) — *IKBKAP*
- ☐ Dystonia (DOPA responsive) — *GCH1, TH*
- ☐ Dystonia (torsion 1, autosomal dominant) — *DYT1*
- ☐ Friedreich ataxia* — *FXN*
- ☐ Hallervorden-Spatz disease — *PANK2*
- ☐ Hyperekplexia (KOK disease)* — *GLRA1*
- ☐ Hyperkalemic periodic paralysis — *SCN4A*
- ☐ Hypokalemic periodic paralysis — *CACNA1A, SCN4A*
- ☐ Hypoventilation syndrome — *PHOX2B*
- ☐ Lafora syndrome — *EPM2A, NHLRC1*
- ☐ Lesh-Nyhan syndrome — *HPRT1*
- ☐ MASA syndrome* — *L1CAM*
- ☐ Menkes syndrome* — *ATP7A*
- ☐ Myasthenic syndrome*, congenital combined with AChR deficiency — *CHRNE, CHAT*
- ☐ Myoklonic epilepsy
  - ☐ severe of infancy — *SCN11A*
  - ☐ Unverricht-Lundborg — *CSTB*
- ☐ NARP syndrome* — *MTATP6*
- ☐ Neuropathies[1,2,3]
- ☐ Onedine syndrome — *PHOX2B*
- ☐ Parkinson disease*
- ☐ Pelizaeus-Merzbacher disease* — *PLP*
- ☐ Pontocellebar hypoplasia type 1 — *VRK1*
- ☐ Rett syndrome — *MECP2*
- ☐ Smith-Lemli-Opitz syndrome* — *DHCR7*
- ☐ Spastic paraplegies[3]
- ☐ Spinal bulbar muscular atrophy*, type Kennedy — *AR*
- ☐ Spinal muscular atrophy (SMA) — *SMN1*
- ☐ Spinocerebellar ataxies[3]
- ☐ X-linked hydrocephalus* — *L1CAM*

Pharmacogenetics — *Gene(s)*

- ☐ Cytochrome P450 defects
- ☐ DPD deficiency (Dihydropyrimidindehydrogenase deficiency) — *DPYD*
- ☐ Multi drug resistance*
- ☐ N-acetyltransferase 2 deficiency — *NAT2*

Short stature — *Gene(s)*

- ☐ Albright syndrome* — *GNAS*
- ☐ Cockayne syndrome* — *ERCC8, ERCC6*
- ☐ Costello syndrome* — *HRAS*
- ☐ Idiopathic short stature — *SHOX*
- ☐ Insulin like growth factor 1 deficiency (IGF deficiency) — *IGF1*
- ☐ Langer, mesomelic dysplasia — *SHOX*
- ☐ Laron syndrome — *GHR*
- ☐ Leri-Weill syndrome — *SHOX*
- ☐ Noonan syndrome — *PTPN11, SOS1*
- ☐ Silver-Rusell syndrome
  - ☐ methylation analysis KvDMR1 and H19DMR
  - ☐ microsatellite analysis UDP(7)mat

Skeletal disorders — *Gene(s)*

- ☐ Achondrogenesis type 2 — *COL2A1*
- ☐ Achondroplasia — *FGFR3*
- ☐ Acromesomelic dysplasia, type Grebe — *CDMP1*
- ☐ Acromesomelic dysplasia, type Maroteaux — *NPR2*
- ☐ Albright syndrome* — *GNAS*
- ☐ Apert syndrome (see Craniosynostosis)
- ☐ Arthrogryposis multiplex congenita* — *TPM2, TNNI2*
- ☐ Brachydactyly type C — *CDMP1*
- ☐ Campomelic dysplasia* — *SOX9*
- ☐ Chondrodysplasia, type Grebe — *CDMP1*
- ☐ Cleidocranial dysplasia — *RUNX2*
- ☐ Clubbing of digits — *HPGD*
- ☐ Cranioectodermal dysplasia
- ☐ Craniosynostosis, FGFR associated — *FGFR1, FGFR2, FGFR3*
- ☐ Crouzon syndrome (see Craniosynostosis)
- ☐ Cystinuria*
- ☐ Desbuquois syndrome — *CANT1*
- ☐ Ektrodactyly (EEC syndrome)* — *TP63*
- ☐ Hypochondroplasia — *FGFR3*
- ☐ Hypophosphatasia, infantile* — *ALPL*
- ☐ Hypophosphatemia*
  - ☐ autosomal dominant — *FGF23*
  - ☐ X-linked — *PHEX*
- ☐ Kniest dysplasia — *COL2A1*
- ☐ Langer, mesomelic dysplasia — *SHOX*
- ☐ Larsen syndrome
  - ☐ — *FLNB*
  - ☐ — *CHST3*
- ☐ Leri-Weill syndrome — *SHOX*
- ☐ Loeys-Dietz syndrome — *TGFBR1, TGFBR2*
- ☐ Marfan syndrome
  - ☐ type 1 — *FBN1*
  - ☐ type 2 — *TGFBR1, TGFBR2*
- ☐ Metaphyseal Chondrodysplasia (type Schmid) — *COL10A1*
  - ☐ hot spot analysis
  - ☐ complete gene analysis incl. hot spots
- ☐ Multiple exostosis* — *EXT1, EXT2*
- ☐ Nail-Patella syndrome — *LMX1B*
- ☐ Orofaciodigital syndrome 1 — *OFD1*
- ☐ Osteogenesis imperfecta
- ☐ Osteogenesis imperfecta, autosomal recessive
- ☐ Osteopetrose,* autosomal recessive
- ☐ Osteoporosis — *COL1A1, VDR*
- ☐ Otopalatodigital syndrome — *FLNA*
  - ☐ type 1 (OPD1)
  - ☐ type 2 (OPD2)
  - ☐ Melnick-Needles syndrome (MNS)
  - ☐ frontometaphyseal dysplasia (FMD)
- ☐ Pfeiffer syndrome (see Craniosynostosis)
- ☐ Polysyndactyly* — *HOXD13*
- ☐ Pseudoachondroplasia* — *COMP*
- ☐ Rubinstein-Taybi syndrome* — *CREBBP*
- ☐ Sensenbrenner syndrome
- ☐ Shwachman-Diamond syndrome* — *SBDS*
- ☐ Spondylocostal dysostosis* — *DLL3*
- ☐ Spondyloepiphyseal dysplasia — *COL2A2*
- ☐ Spondyloepiphyseal dysplasia, X-linked — *SEDL*
- ☐ Stickler-syndrome type 1 — *COL2A2*
- ☐ Thanatophoric dysplasia — *FGFR3*
- ☐ Treacher-Collins-Franceschetti syndrome* — *TCOF1*

Skin and nail disorders — *Gene(s)*

- ☐ Anonychia — *RSPO4*
- ☐ Bloch-Sulzberger syndrome* (Incontinentia pigmenti, IP2) — *IKBKG*
- ☐ Bloom syndrome* (Sister Chromatid Exchange (SCE)) — *RECQL3*
- ☐ Ectodermal dysplasia type 1* — *EDA*
- ☐ Epidermolysis bullosa
- ☐ Netherton syndrome — *SPINK5*
- ☐ Neurofibromatosis
  - ☐ type 1 — *NF1*
  - ☐ type 2 — *NF2*
- ☐ Primary hypertrophic Osteoarthropathy (Pachydermoperiostose/Tourraine-Solente-Gole syndrome) — *HPGD*
- ☐ Sjögren-Larsson syndrome* — *ALDH3A2*
- ☐ Werner syndrome — *RECQL2*

Storage disorders — *Gene(s)*

- ☐ Arylsulfatase A deficiency* — *ARSA*
- ☐ Ceroid lipofuscinosis, neural (all types)
- ☐ Fabry disease — *GLA*
- ☐ Gaucher disease* — *GBA*
- ☐ Glycogen storage diseases*[1]
- ☐ Hemochromatosis — *HFE*
- ☐ Hallervorden-Spatz disease — *PANK2*
- ☐ Morbus Krabbe*
- ☐ Morbus Pompe* — *GAA*
- ☐ Morbus Wilson — *ATP7B*
- ☐ Mucolipidosis* — *GNPTAB*
- ☐ Mucopolysaccharidosis*
- ☐ Nieman-Pick disease*
- ☐ Sandhoff disease — *HEXB*
- ☐ Tay-Sachs disease
  - ☐ Jewish screening — *HEXA*
  - ☐ complete gene — *HEXA*
  - ☐ *HEXA and HEXB* genes — *HEXA, HEXB*
- ☐ Very long chain acyl-CoA dehydrogenase deficiency* — *ACADVL*

Usher syndrome — *Gene(s)*

- ☐ Usher syndrome[1,2,3]
  - ☐ type 1
  - ☐ type 2
  - ☐ USH1: FC-/Acadian Panel
  - ☐ USH2: FC-/Acadian Panel

Vasculopathies — *Gene(s)*

- ☐ CADASIL — *NOTCH3*
- ☐ Lymphedema type 1* — *FLT4*

[1] We offer molecular testing for all genes known for this disease. In some cases, specific genotype - phenotype correlations exist that allow for targeted testing. Please contact us.

[2] We offer linkage analysis where applicable (e. g., samples available from several family members, parental consanguinity) for prioritization of genes to be tested.

[3] Stepwise analysis

*Analyses are carried out in collaborating certified labs.

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