CENTER FOR HUMAN GENETICS AND LABORATORY DIAGNOSTICS
Dr. Klein, Dr. Rost and Colleagues

A1

Human Genetics	Laboratory Medicine	Transfusion Medicine	Pathology	Microbiology/Virology
Dr. med. Imma Rost Dr. med. H.-G. Klein	Dr. med. Hanns-Georg Klein Dr. med. Kaimo Hirv (komm.)	Dr. med. Kaimo Hirv Dr. med. H.-G. Klein (komm.)	Prof. Dr. med. Barbara Dockhorn-Dworniczak PD Dr. med. Marcus Kremer	Dr. med. Hartmut Campe Dr. med. Hanns-Georg Klein

Human Genetics/Transfusion Medicine/Pathology

Surname, first name(s) (patient): _____

Date of birth: _____ Sex: ☐ M ☐ F

Street address: _____

Postcode, City: _____

Telephone: _____

Payment Details

We require payment by bank transfer **prior** to the investigation.

We will send cost information for your agreement upon receipt of this completed form. Following agreement, we will send bank details for payment.

☐ I have read and understood the payment details.

Order/Indication – Diagnosis/Suspected Diagnosis

Note: unsigned material will be rejected

Responsible Medical Person (stamp) - Date of Order - Signature

✗

Informed consent and field for pedigree information below (not applicable to pathology)

Investigative Material

☐ EDTA blood [1]	☐ Amniotic fluid [5]	☐ FFPE material [8]	☐ Cerebrospinal fluid [11]
☐ Heparin blood [2]	☐ Chorionic villi [6]	☐ Native tissue [9]	☐ Urine [12]
☐ Streck blood [3]	☐ Abortion material [7]	☐ Formalin-fixed [10]	☐ Heparin bm [13]
☐ Serum [4]	☐ DNA from	☐ RNA from	☐ EDTA bm [14]

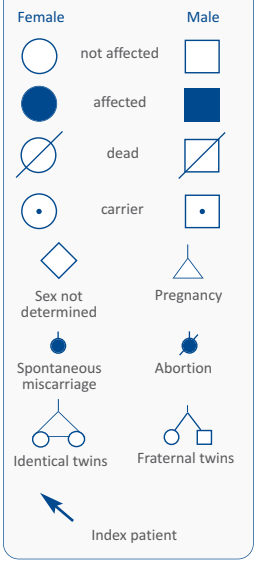
Sampling date: Time:

Additional Recipients of Results (if applicable, disclosure according to GenDG required)

Clinical Information

Previous Preliminary Diagnostics

Symbols



Pedigree Information, if applicable (please use the symbols on the left)

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DAkkS Deutsche Akkreditierungsstelle D-ML-13317-01-00

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Genetic Counseling
Dr. med. Imma Rost (Head.) Prof. Dr. med. J.-U. Walther
Dr. (Univ. Verona) Monika Cohen Dr. med. Dagmar Wahl

Informed Consent According to German Diagnostic Genetic Act (GenDG) – *applicable only to investigations to determine genetic (hereditary) characteristics*

The GenDG requires provision of detailed **information** and a **written consent** for all genetic investigations, as well as **genetic counseling** prior to both predictive (applies to healthy individuals) and prenatal analyzes. The German Society of Human Genetics (GfH) and the Association of German Human Geneticists (BVDH) recommend clarifying the issues listed below during the consent process. **Please read the declaration of consent carefully** and cross out the statements that you do not agree with.

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 been given sufficient opportunity to discuss open questions,
- agree to the sampling of specimens needed for investigation (blood, tissue, chorionic villus cells or amniotic fluid for prenatal diagnosis),
- consent to a **genetic test** being carried out in order to clarify the **disease/diagnosis/suspected diagnosis** given here

_____

- agree with the evaluation of additional genes in the same indication group as part of the research
- agree that the remaining sample material may be stored for further investigations after the examination is completed, yet not claiming storage,
- agree that the **sample material**, and if applicable DNA sequence information, may be made available anonymously for quality management and scientific purposes
- agree that the results of the analysis may be used anonymously for scientific publications,
- 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,
- agree that the investigation or parts of the investigation may be **forwarded** to collaborating medical laboratories if necessary,
- agree that a copy of the results of the investigation may be sent to the following physician

Dr(s). med. _____
Name Street Postcode/City Country

In addition, I have been informed that,

- I may stop the investigation at any time and ask for the results available until that time to be destroyed,
- I may withdraw my consent entirely or in part at any time without giving reasons,
- I will be liable for fees incurred until the time of withdrawal of consent,
- I may choose not to be informed about the test results (**right not to know**),
- the genetic investigation and evaluation is limited to the requested indication and no statements will be made about other diseases,
- in the case of **duo/trio/quattro analyzes** (large panels, exomes, genomes) the analysis results from those unaffected are only for the validation of variants from the index patient,
- in rare cases, clinically relevant **additional findings** that are not related to the investigation requested, but which have a treatment consequence for me and/or my family members, may occur. I have no right to claim completeness or future updates of the additional findings.

Important: Communication of additional findings with therapeutic or prophylactic relevance found by chance or during the research

☐ Yes, I wish to be informed about additional findings ☐ No, I do not wish to be informed about additional findings (no selection will be classified as "no")

_____ ✗ _____ ✗ _____
Place, date Signature of patient or parent/legal representative Signature of responsible physician

Mandatory field: Credit card information - to be completed by the patient

Type of card

☐ Mastercard
☐ Visa
☐ American Express

Owner of the card _____

Credit card number _____ Security code _____ Expiration date _____

Place _____ Date _____

Signature (owner of the card) _____

Human Genetics

Panel diagnostics: Basic diagnostics (blue), advanced diagnostics (green, subject to authorization by statutory health insurances)
Individual indications: Diagnostic defined individual indications (black)
Coding: OMIM-P-Number (6 digits), Disease (*AFFECTED GENE*),"*core genes*": ***in bold***, [ICD-10-Code], Material coding: [1-14]

Eye disease (Panel Diagnostics) [1]

- ☐ Retinitis pigmentosa *(EYS, PRPF31, PRPH2, RHO, RP1, RP2, RPGR)*
- ☐ Retinitis pigmentosa, complete *(99 genes)*
- ☐ Usher syndrome type 1 and 2 ***(USH2A, MYO7A)***
- ☐ Usher syndrome type 1 ***(MYO7A, CDH23, PCDH15**, USH1G)*
- ☐ Usher syndrome *(12 genes)*
- ☐ Stickler syndrome (ocular participation) *(COL11A1, **COL2A1**, COL9A1, COL9A2)*
- ☐ Senior-Løken syndrome *(7 genes)*
- ☐ Senior-Løken syndrome *(WDR19)*
- ☐ Bardet-Biedl syndrome *(16 genes)*
- ☐ Bardet-Biedl syndrome *(8 genes)*
- ☐ Macular degeneration (*CFH, HTRA1*)
- ☐ Wagner syndrome *(see skeletal dysplasia)*

Aortic and Connective Tissue Diseases (Panel Diagnostics) [1]

Marfan syndrome/Thoracic aortic aneurysm/Bicuspid aortic valve disease/Collagen type 4 associated intracerebral hemorrhage/Cutis laxa ----------

- ☐ Marfan syndrome and type 1 fibrillinopathies ***(FBN1, TGFBR1, TGFBR2)***
- ☐ Marfan-like diseases *(ADAMTSL4, FBN2, SKI, MEDF12, UPF3B, ZDHHC9)*
- ☐ Thoracic aortic aneurysm with risk of aortic dissection *(34 genes)*
- ☐ Bicuspid aortic valve disease with risk of aortic stenosis/-dilatation *(3 genes)*
- ☐ Collagen type 4 associated intracerebral hemorrhage *(COL4A1, COL4A2)*
- ☐ Cutis laxa *(ALDH18A1, ATP6V0A2, EFEMP2, ELN, FBLN5, LTBP4, PYCR1)*

Ehlers-Danlos syndrome ----------

- ☐ Ehlers-Danlos syndrome, classic type (cEDS) ***(COL5A1, COL5A2, COL1A1)***
- ☐ Ehlers-Danlos syndrome, vascular type (vEDS) ***(COL3A1)***
- ☐ Ehlers-Danlos syndrome, arthrochalasis type (aEDS) ***(COL1A1, COL1A2)***
- ☐ Ehlers-Danlos syndrome, autosomal dominant subtypes *(5 genes)*
- ☐ Ehlers-Danlos syndrome, autosomal recessive subtypes *(9 genes)*

Connective Tissue Disease/Skeletal Dysplasia (Panel Diagnostics) [1]

- ☐ Osteogenesis imperfecta, autosomal dominant ***(COL1A1, COL1A2**, IFITM5, WNT1)*
- ☐ Osteogenesis imperfecta, autosomal recessive *(11 genes)*
- ☐ Stickler syndrome *(COL11A1, COL11A2, **COL2A1**, COL9A1, COL9A2)*
- ☐ Jeune-/Short-rib thoracic dysplasia ***(DYNC2H1, IFT80, NEK1, TTC21B, WDR34)***
- ☐ Jeune-/Short-rib thoracic dysplasia *(12 genes)*
- ☐ Craniosynostosis ***(FGFR1, FGFR2, FGFR3, TCF12, TWIST1)***
- ☐ Craniosynostosis *(32 genes)*

Connective Tissue Diseases (Individual Indications) [1]

- ☐ 610168 Aortic aneurysm, familial thoracic 3-8, (*TGFBR2, MYH11, TGFBR1, ACTA2, MYLK, PRKG1*) [I71.1, I71.2]
- ☐ 208050 Arterial tortuosity syndrome *(SLC2A10)* [Q25.4]
- ☐ 109730 Aortic valve disease 1, AOVD1 *(NOTCH1)* [Q23.1]
- ☐ 121050 Contractural arachnodactyly, congenital, CCA *(FBN2)* [Q87.5]
- ☐ 614437 Cutis laxa TYP 1B, ARCL1B *(EFEMP2)* [Q82.8]
- ☐ 130000 EDS, classic type, cEDS (*COL5A1, COL5A2, COL1A1*) [Q79.6]
- ☐ 130050 EDS, vascular type, vEDS (*COL3A1*) [Q79.6]
- ☐ 130060 EDS, arthrochalasis type, aEDS (*COL1A1, COL1A2*) [Q79.6]
- ☐ 225410 EDS, dermatosparaxis type, dEDS (*ADAMTS2*) [Q79.6]
- ☐ 225400 EDS, kyphoscoliotic type, kEDS (*PLOD1*) [Q79.6]
- ☐ 614557 EDS, kyphoscoliotic type, kEDS (*FKBP14*) [Q79.6]
- ☐ 601776 EDS, musculocontractural type 1, mcEDS (*CHST14*) [Q79.6]
- ☐ 615349 EDS, spondylodysplastic form, spEDS-B4GALT6 (*B3GALT6*) [Q79.6]
- ☐ 130070 EDS, spondylodysplastic form, spEDS-B4GALT7 (*B4GALT7*)) [Q79.6]
- ☐ 225320 EDS, cardiac valvular form, cvEDS (*COL1A2*) [Q79.6]
- ☐ 300049 EDS, periventricular heterotopia, PVNH4 (*FLNA*) [Q79.6]
- ☐ 606408 EDS, due to tenascin-X deficiency, classical like, clEDS (*TNXB*) [Q79.6]
- ☐ 609192 Loeys-Dietz syndrome type 1 *(TGFBR1)* [Q87.4]
- ☐ 610168 Loeys-Dietz syndrome type 2 *(TGFBR2)* [Q87.4]
- ☐ 613795 Loeys-Dietz syndrome type 3 *(SMAD3)* [Q87.4]
- ☐ 614816 Loeys-Dietz syndrome type 4 *(TGFB2)* [Q87.4]
- ☐ 615582 Loeys-Dietz syndrome type 5 *(TGFB3)* [Q87.4]
- ☐ 154700 Marfan syndrome *(FBN1)* [Q87.8]
- ☐ 264800 Pseudoxanthoma elasticum (ABCC6) [Q82.8]

Skelettdysplasien (Individual Indications) [1]

- ☐ 200610 Achondrogenesis type 2, Langer-Saldino *(COL2A1)* [Q77.0]
- ☐ 100800 Achondroplasia *(FGFR3)* [Q77.4]
- ☐ 200610 Hypochondrogenesis *(COL2A1)* [Q77.0]
- ☐ 146000 Hypochondroplasia *(FGFR3)* [Q77.4]
- ☐ 156550 Kniest dysplasia, type II collagenopathy *(COL2A1)* [Q78.9]
- ☐ 249700 Langer mesomelic dysplasia *(SHOX)* [Q77.8, Q87.1]
- ☐ 127300 Léri-Weill dyschondrosteosis *(SHOX)* [Q77.8, Q87.1]
- ☐ 154780 Marshall syndrome *(COL11A1)* [M35.9]
- ☐ 156500 Metaphyseal chondrodysplasia, Schmid type *(COL10A1)* [Q78.5]
- ☐ 166200 Osteogenesis imperfecta *(COL1A1, COL1A2)* [Q78.0]
- ☐ 610682 Osteogenesis imperfecta, recessive *(CRTAP, LEPRE1, PPIB)* [Q78.0]
- ☐ 166710 Osteoporosis, postmenopausal *(COL1A1-S/s)* [M81.0]
- ☐ 215150 Otospondylomegaepiphyseal dysplasia OSMED *(COL11A2)* [Q78.9]
- ☐ 183900 Spondyloepiphyseal dysplasia *(COL2A1)* [Q78.9]
- ☐ 108300 Stickler syndrome type I *(COL2A1)* [Q87.8]
- ☐ 604841 Stickler syndrome type II *(COL11A1)* [Q87.8]
- ☐ 184840 Stickler syndrome type III *(COL11A2)* [Q87.8]
- ☐ 187600 Thanatophoric dysplasia type I *(FGFR3)* [Q77.1]
- ☐ 187601 Thanatophoric dysplasia type II *(FGFR3)* [Q77.1]
- ☐ 143200 Wagner syndrome *(CSPG2)* [H33.5]

Fever Syndrome (Panel Diagnostics) [1]

- ☐ Hereditary periodic fever syndromes *(ELANE, IL1RN, IL36RN, LPIN2, MEFV, MVK, NLRC4, NLRP12, NLRP3, NOD2, PSMB8, PSTPIP1, TMEM173, TNFRSF1A)*

Fever Syndrome (Individual Indications) [1]

- ☐ 120100 Cryopyrin-associated periodic syndrome, CAPS *(NLRP3)*
- ☐ 249100 Familial mediterranean fever, FMF *(MEFV)*
- ☐ 610377 Mevalonic aciduria *(MVK)*
- ☐ 142680 TNF receptor 1-associated periodic syndrome, TRAPS *(TNFRSF1A)*

Bleeding Disorders (Panel Diagnostics) [1]

- ☐ Bleeding tendency *(F7, F8, F9, F13A1, F13B, VWF)*
- ☐ Tendency towards thrombosis/thrombophilia *(F5, F2, PROC, PROCS1)*

Bleeding Disorders (Individual Indications) [1]

- ☐ 227500 Factor VII deficiency *(F7)* [D68.23]
- ☐ 613225 Factor XIII deficiency *(F13A1)* [D68.26]
- ☐ 306700 Hemophilia A *(F8)* [D66]
- ☐ 306900 Hemophilia B *(F9)* [D67]
- ☐ 176860 Protein C deficiency *(PROC)* [D68.5]
- ☐ 176880 Protein S deficiency *(PROS1)* [D68.5]
- ☐ 188050 Thrombophilia *(FV-R506Q, FII-G20210A)* [D68.5, I82.9]
- ☐ 193400 von-Willebrand syndrome *(VWF)* [D.68.0]

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ilac-MRA DAkkS Deutsche Akkreditierungsstelle D-ML-13317-01-00

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Hemoglobinopathies and Erythrocyte Membrane Disorders (Individual Indications) [1]

- ☐ 604131 Alpha-thalassemia *(HBA1, HBA2)* [D56.0]
- ☐ 613985 Beta-thalassemia *(HBB)* [D56.1]
- ☐ 305900 G6PD deficiency *(G6PD)* [D55.0]
- ☐ 604131 Hemoglobin anomalies *(HBB, HBA1, HBA2)* [D58.2]
- ☐ 141749 Hereditary persistence of fetal hemoglobin *(HBB, HBG1, HBG2)* [56.4]
- ☐ 603903 Sickle cell anemia *(HBB)* [D57]
- ☐ 182900 Spherocytosis *(ANK1, SPTA1, SPTB, SLC4A1, EPB42)* [D58.0]

Heart Muscle Diseases and Arrhythmogenic Diseases (Panel Diagnostics) [1]

Arhythmogenic diseases

- ☐ Arrhythmogenic right ventricular dysplasia ARVD *(7 genes)*
- ☐ Brugada syndrome, BS *(7 genes)*
- ☐ Brugada syndrome, BS *(12 genes)*
- ☐ Catecholaminergic polymorphic ventricular tachycardia, CPVT *(4 genes)*
- ☐ Dilated cardiomyopathy, DCM *(11 genes)*
- ☐ Dilated cardiomyopathy, DCM *(34 genes)*
- ☐ Dilated cardiomyopathy, DCM *(TTN gene)*
- ☐ Hypertrophic cardiomyopathy, HCM *(12 genes)*
- ☐ Hypertrophic cardiomyopathy, HCM *(16 genes)*
- ☐ Long QT syndrome, LQTS *(9 genes)*
- ☐ Long QT syndrome, LQTS *(6 genes)*
- ☐ Non-compaction cardiomyopathy, NCCM *(10 genes)*

Congenital heart defects

- ☐ Isolated heart defects *(12 genes)*
- ☐ Isolated heart defects *(13 genes)*
- ☐ Syndromic heart defects *(7 genes)*
- ☐ Syndromic heart defects *(29 genes)*
- ☐ Heart defects, heterotaxy associated *(10 genes)*
- ☐ Heart defects, heterotaxy associated *(2 genes)*
- ☐ RASopathies with heart defects *(14 genes)*
- ☐ RASopathies with heart defects *(4 genes)*

Primary Immunodeficiency (Panel Diagnostics) [1]

Combined T- and B-Cell Immunodeficiencies

- ☐ Combined T- and B-cell immunodeficiencies *(18 genes)*
- ☐ Combined T- and B-cell immunodeficiencies *(25 genes)*
- ☐ Severe combined immunodeficiencies, T-B- *(8 genes)*
- ☐ Severe combined immunodeficiencies, T-B+ *(9 genes)*
- ☐ Omenn syndrome *(10 genes)*

Hereditary Agammaglobulinemia

- ☐ Hereditary agammaglobulinemia *(8 genes)*

Congenital neutropenia

- ☐ Congenital neutropenia *(19 genes)*
- ☐ Congenital neutropenia *(LYST, VPS45)*

Primary Immunodeficiency (Individual Indications) [1]

- ☐ 300755 Agammaglobulinemia Bruton type, XLA (*BTK*) [D80.0]
- ☐ 240300 Autoimmune polyendocrinopathy syndrome, APECED (*AIRE*) [D84.8]
- ☐ 301000 Wiskott-Aldrich syndrome, WAS (*WAS*) [D82.0]
- ☐ 308240 X-linked lymphoproliferative syndrome, XLP1 (*SH2D1A*) [D82.3]
- ☐ 300400 X-linked severe combined immunodeficiency, X-SCID (*IL2RG*) [D81.9]

Mental Retardation/Developmental Disorders and Dysmorphia Syndromes (Panel Diagnostics) [1] - *for further information see www.medizinische-genetik.de*

- ☐ Autism *(8 genes)*
- ☐ Autism *(57 genes)*
- ☐ CDG syndrome *(31 genes)*
- ☐ Coffin-Siris syndrome *(6 genes)*
- ☐ Cornelia-de-Lange syndrome *(5 genes)*
- ☐ Developmental disorders *(genes up to 25 kb to choose from)*
- ☐ Developmental disorders *(341 genes)*
- ☐ GPI anchor deficiency, hyperphosphatasia with mental retardation syndrome *(12 genes)*
- ☐ Gigantism syndrome *(9 genes)*
- ☐ Kabuki syndrome *(2 genes)*
- ☐ Macrocephalies *(6 genes)*
- ☐ Macrocephalies *(70 genes)*
- ☐ Microcephaly, primary (a-r) *(5 genes)*
- ☐ Microcephalic osteodysplastic primordial dwarfism *(1 gene)*
- ☐ Microcephaly, primary (a-r) *(16 genes)*
- ☐ Rett syndrome/Rett-like syndrome *(10 genes)*
- ☐ Rett syndrome/Rett-like syndrome *(21 genes)*
- ☐ Robinow syndrome *(4 genes)*
- ☐ Rubinstein-Taybi syndrome *(2 genes)*
- ☐ Tubulinopathies/brain deformities *(7 genes)*

- ☐ "Clinical Exome Trio" analysis *(>4,800 genes) - only after consultation*
- ☐ "Whole Exome Trio" (*approx. 20,000 genes*) *- only after consultation*

Mental Retardation/Pediatric Genetic Syndromes (Individual Indications) [1]

- ☐ 105830 Angelman syndrome, methylation test *(UBE3A)* [Q93.5]
- ☐ 105830 Angelman syndrome, AS *(UBE3A)* [Q93.5]
- ☐ 214800 CHARGE syndrome *(CHD7)* [Q87.8]
- ☐ 300624 Fragile X syndrome, FRAXA *(FMR1)* [Q99.2]
- ☐ 142900 Holt-Oram syndrome *(TBX5)* [Q87.2]
- ☐ 147920 Kabuki syndrome *(MLL)* [Q87.8]
- ☐ 300260 MECP2 duplication syndrome *(MECP2)* [F84.9]
- ☐ 309580 Mental retardation-hypotonic facies syndrome, X-linked *(ATRX)* [F79.-]
- ☐ 608716 Microcephaly 5, primary, autosomal recessive *(ASPM)* [Q02]
- ☐ 235730 Mowat-Wilson syndrome *(ZEB2)* [F89.0]
- ☐ ..... Noonan syndrome *(PTPN11, SOS1, RIT1, RAF1, KRAS, BRAF, MAP2K1, MAP2K2, CBL, PPP1CB, SHOC2, NRAS, RASA2, RRAS, SOS2)* [Q87.1] - mark desired genes
- ☐ 607323 Okihiro syndrome *(SALL4)* [Q87.2]
- ☐ 610954 Pitt-Hopkins syndrome *(TCF4)* [F89.0]
- ☐ 176270 Prader-Willi syndrome (methylation test) [Q87.1]
- ☐ 312750 Rett syndrome *(MECP2)* [F84.2]
- ☐ 300672 Rett syndrome, atypical *(CDKL5)* [F84.9]
- ☐ 613454 Rett syndrome, congenital *(FOXG1)* [F84.2]
- ☐ 180849 Rubinstein-Taybi syndrome *(CREBBP)* [Q87.2]
- ☐ 270400 Smith-Lemli-Opitz syndrome *(DHCR7)* [Q87.1]
- ☐ 117550 Sotos syndrome *(NSD1)* [Q87.3]
- ☐ 615879 Tatton-Brown-Rahman syndrome *(DNMT3A)* [Q87.3]
- ☐ 277590 Weaver syndrome *(EZH2)* [T89.0]
- ☐ 300419 X-linked mental retardation *(ARX)* [F79]

Craniosynostosis (Panel Diagnostics) [1]

- ☐ Craniosynostosis ***(FGFR1, FGFR2, FGFR3, TCF12, TWIST1)***
- ☐ Craniosynostosis *(32 genes)*

Craniosynostosis (Individual Indications) [1]

- ☐ 101200 Apert syndrome *(FGFR2)* [Q87.0]
- ☐ 123500 Crouzon syndrome *(FGFR2)* [Q75.1]
- ☐ 602849 Muenke syndrome *(FGFR3)* [Q75.0]
- ☐ 101600 Pfeiffer syndrome *(FGFR1, FGFR2)* [Q75.0]
- ☐ 101400 Saethre-Chotzen syndrome *(TWIST1, FGFR3)* [Q75.0]

Pulmonary Diseases (Panel Diagnostics) [1]

- ☐ Pulmonary arterial hypertension, PAH *(9 genes)*
- ☐ Interstitial-/diffuse parenchymal lung disease, ILD/DPLD *(8 genes)*

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Pulmonary Diseases (Individual Indications) [1]

- ☐ 613490 Alpha-1-antitrypsin deficiency *(SERPINA1)* [E88.0]
- ☐ 219700 Cystic fibrosis/mucoviscidosis *(CFTR)* [E84.9]
- ☐ 610978 Brain-lung-thyroid syndrome *(NKX2-1)* [J84.-]
- ☐ - FLNA-associated pulmonary disease *(FLNA)* [J84.-]
- ☐ 265380 Congenital alveolar capillary dysplasia *(FOXF1)* [J84.-]
- ☐ 265120 Surfactant metabolism dysfunction type 1 *(SFTPB)* [J84.0]
- ☐ 610913 Surfactant metabolism dysfunction type 2 *(SFTPC)* [J84.0]
- ☐ 610921 Surfactant metabolism dysfunction type 3 *(ABCA3)* [J84.0]
- ☐ 300770 Surfactant metabolism dysfunction type 4 *(CSF2RA)* [J84.0]
- ☐ 614370 Surfactant metabolism dysfunction type 5 *(CSF2RB)* [J84.0]
- ☐ 178600 Pulmonary arterial hypertension *(BMPR2)* [I27.0]

Mitochondrial Disease (Panel Diagnostics) [1]

- ☐ Mitochondrial disease *(examination of the entire mitochondrial genome in cases of suspected Leber's hereditary optic neuropathy (LHON), MELAS and MERFF (indicate heteroplasmy grade) and in cases of suspected mitochondrial deafness)*

Muscle Diseases (Panel Diagnostics) [1]

Spinal muscular atrophy

- ☐ Spinal muscular atrophy (neonatal/early onset) *(9 genes)*
- ☐ Spinal muscular atrophy (late onset) (9 genes) *(9 genes)*
- ☐ Spinal muscular atrophy *(21 genes)*

Congenital myopathies/muscular dystrophies/myofibrillar myopathies

- ☐ Congenital myopathies *(**ACTA1**, MYH7, **RYR1, TPM3**)*
- ☐ Muscular dystrophies (collagen-associated and other) *(7 genes)*
- ☐ Muscular dystrophies (dystroglycanopathy types A and B) *(14 genes)*
- ☐ Congenital myopathies *(22 genes)*
- ☐ Muscular dystrophies (other) *(21 genes)*

Myofibrillar myopathies (MFM)

- ☐ Myofibrillar myopathies *(DES, CRYAB, MYOT, LDB3, FLNC, BAG3)*

Progressive muscular dystrophies

- ☐ Progressive muscular dystrophies (Duchenne/Becker) (***DMD***)
- ☐ Progressive muscular dystrophies (limb-girdle, AD + AR) *(14 genes)*
- ☐ Emery-Dreifuss muscular dystrophy *(EMD, FHL1, **LMNA**, SYNE2)*
- ☐ Progressive muscular dystrophies *(36 genes)*

Non-dystrophic myotonia and periodic paralysis

- ☐ Non-dystrophic myotonia and periodic paralysis *(5 genes)*
- ☐ Non-dystrophic myotonia and periodic paralysis *(HSPG2)*

Metabolic myopathies

- ☐ Metabolic myopathies (glycogenosis with muscular symptoms and carnitine metabolic disorders) *(10 genes)*
- ☐ Metabolic myopathies (defects in mitochondrial β-oxidation and mitochondrial DNA depletion syndrome (MTDPS) with myopathy) *(15 genes)*
- ☐ Metabolic myopathies *(31 genes)*

Neurogenetic Disorders (Panel Diagnostics) [1]

Ataxias

- ☐ Autosomal dominant spinocerebellar ataxia *(genes up to 25 kb for selection)*
- ☐ Autosomal recessive spinocerebellar ataxia *(genes up to 25 kb for selection)*
- ☐ Autosomal dominant spinocerebellar ataxia *(18 genes)*
- ☐ Autosomal recessive spinocerebellar ataxia *(12 genes)*
- ☐ Autosomal dominant and autosomal recessive spinocerebellar ataxia *(30 genes)*
- ☐ Ataxias, syndromic forms *(genes up to 25 kb for selection)*
- ☐ Ataxias, syndromic forms including Joubert syndrome (ataxia) (56 genes)
- ☐ Ataxia with oculomotor apraxia (AR) *(APTX, PIK3R5, PNKP, SETX)*
- ☐ Episodic ataxia *(CACNA1A, CACNB4, KCNA1, SCN2A, SLC1A3)*
- ☐ Leukoencephalopathy with vanishing white matter *(5 genes)*
- ☐ Spastic ataxia *(7 genes)*
- ☐ Total ataxia panel *(99 genes)*

Hereditary epilepsies

- ☐ Absence epilepsy *(8 genes)*
- ☐ Benign familial epilepsy (neonatal, infantil) *(6 genes)*
- ☐ Dravet syndrome *(7 genes)*
- ☐ Epilepsy with increased therapy resistance *(7 genes)*
- ☐ Familial hemiplegic migraine *(3 genes)*
- ☐ Early infantile epileptic encephalopathy *(ARX, CDKL5, KCNQ2, PCDH19, SCN1A, SCN2A, STXBP1)*
- ☐ Early infantile epileptic encephalopathy *(DNM1, GABRA1, GABRB3, GNAO1, KCNA2, KCNT1, SCN8A, SLC12A5, SLC13A5, UBA5, WWOX)*
- ☐ Early infantile epileptic encephalopathy *(GRIN2B, HCN1, KCNB1, PIGA, PLCB1, PNKP, SLC35A2, SPTAN1)*
- ☐ Early infantile epileptic encephalopathy *(60 genes)*
- ☐ Focal epilepsy *(9 genes)*
- ☐ Generalized epilepsy with seizures plus *(7 genes)*
- ☐ Generalized epilepsy with seizures plus *(8 genes)*
- ☐ Juvenile myoclonic epilepsy *(9 genes)*
- ☐ Progressive myoclonic epilepsy *(12 genes)*
- ☐ Temporal lobe epilepsy and frontal lobe epilepsy *(8 genes)*

Other

- ☐ Alzheimer's disease, familial *(APP, PSEN1, PSEN2)*
- ☐ Charcot-Marie-Tooth neuropathy type 1 and type 2 *(15 genes)*
- ☐ Hereditary neuropathies *(55 genes)*
- ☐ Choreatic movement disorders *(12 genes)* - triplet analyses *ATXN1-7, TBP, ATN1*
- ☐ Choreatic movement disorders *(15 genes)*
- ☐ Hyperekplexia *(GLRA1, SLC6A5, GLRB)*

Neurological/Neurodegenerative/Neuromuscular Disorders (Individual Indications) [1]

- ☐ 104300 Alzheimer's disease, familial, early-onset *(PSEN1, APP)* [F00.0]
- ☐ 104310 Alzheimer's disease, late-onset *(APOE)* [F00.1]
- ☐ 105210 Amyloidosis, familial *(TTR, APOA1)* [E85.2]
- ☐ 229300 Friedreich ataxia *(FXN)* [G11.1]
- ☐ 108500 Episodic ataxia, type 2 *(CACNA1A)* [G43.1]
- ☐ 164400 Spinocerebellar ataxia *(ATXN1,2,3,7, CACNA1A,TBP)* [G11.9]
- ☐ 125310 CADASIL *(NOTCH3)* [I67.8]
- ☐ 117000 Central core disease of muscle *(RYR1)* [M62.5]
- ☐ 118220 Charcot-Marie-Tooth disease type 1A *(PMP22)* [G60.0]
- ☐ 118200 Charcot-Marie-Tooth disease type 1B *(MPZ)* [G60.0]
- ☐ 302800 Charcot-Marie-Tooth neuropathy type X1 *(GJB1)* [G60.0]
- ☐ 118300 Charcot-Marie-Tooth neuropathy type 1E *(PMP22)* [G60.0]
- ☐ 143100 Huntington disease *(HTT)* [G10]
- ☐ 123400 Creutzfeldt-Jakob disease, familial *(PRNP)* [A81.0]
- ☐ 123400 Creutzfeldt-Jakob disease, variant *(PRNP-M129V)* [A81.0]
- ☐ 125370 Dentatorubro-pallidoluysian atrophy *(ATN1)* [G11.9]
- ☐ 607208 Severe myoclonic epilepsy of infancy (Dravet syndrome) *(SCN1A, SCN1B, GABRG2, SCN2A)* [G40.3]
- ☐ 604233 Epilepsy, generalized, with febrile seizures plus, GEFS+ *(SCN1A, SCN1B, GABRG2, SCN2A)* [G40.3]
- ☐ 300088 Epileptic encephalopathy, early infantile, X-linked with mental retardation *(PCDH19)* [G40.3]
- ☐ 308350 Epileptic encephalopathy, early infantile, 1, EIEE1 *(ARX)* [G40.8]

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Neurological/Neurodegenerative/Neuromuscular Disorders (Individual Indications) (Individual Indications) [1] - *continued*

- ☐ 612164 Epileptic encephalopathy, early infantile, 4, EIEE4 *(STXBP1)* [G40.3]
- ☐ 611942 Epilepsy, childhood absence, susceptibility to, 6 *(CACNA1H)* [G40.3]
- ☐ 141500 Migraine, familial hemiplegic, 1, FHM1 *(CACNA1A)* [G43.1]
- ☐ 602481 Migraine, familial hemiplegic, 2, FHM2 *(ATP1A2)* [G43.1]
- ☐ 609634 Migraine, familial hemiplegic, 3, FHM3 *(SCN1A)* [G43.1]
- ☐ 266100 Pyridoxine-dependent epilepsy *(ALDH7A1)* [G40.8]
- ☐ 300623 Fragile X tremor/ataxia syndrome, FXTAS *(FMR1)* [G11.2]
- ☐ 137440 Gerstmann-Sträussler-Scheinker disease *(PRNP)* [A81.9]
- ☐ 606777 GLUT1 deficiency syndrome 1 *(SLC2A1)* [G40.3]
- ☐ 162500 Hereditary neuropathy, with liability to pressure palsies *(PMP22)* [G60.0]
- ☐ 170400 Hypokalemic periodic paralysis, type 1 *(CACNA1S, KCNJ18, KCNE3)* [G72.3]
- ☐ 117000 Neuromuscular disease, congenital, with uniform type 1 fiber *(RYR1)* [G60]
- ☐ 255320 Minicore myopathy *(RYR1)* [M62.5]
- ☐ 253300 Spinal muscular atrophy *(SMN1)* [G12.9]
- ☐ 313200 Spinal and bulbar muscular atrophy, Kennedy disease *(AR)* [M62.59]
- ☐ 300376 Muscular dystrophy, Becker type *(DMD)* [G71.0]
- ☐ 310200 Muscular dystrophy, Duchenne type *(DMD)* [G71.0]
- ☐ 160900 Myotonic dystrophy 1, Curschmann-Steinert disease *(DMPK)* [G71.1]

Kidney Diseases (Panel Diagnostics) [1]

Congenital anomalies of the kidney and urinary tract (CAKUT) ----------

- ☐ CAKUT *(15 genes)*
- ☐ CAKUT *(35 genes)*
- ☐ Deformities of the urinary tract *(12 genes)*
- ☐ Deformities of the urinary tract *(11 genes)*
- ☐ Renal agenesis/hypoplasia *(12 genes)*
- ☐ Renal agenesis/hypoplasia *(6 genes)*
- ☐ Renal tubular dysgenesis *(ACE, AGT, AGTR1, REN)*
- ☐ Polycystic kidney disease - autosomal dominant *(BMP4, HNF1B, PAX2, PKD1, PKD2)*
- ☐ Polycystic kidney disease - autosomal recessive *(FRAS1, PKHD1)*
- ☐ Polycystic kidney disease *(9 or 12 genes)*

Others ----------

- ☐ Alport syndrome *(OL4A3, COL4A4, COL4A5, MYH9)*
- ☐ Nephronophthisis *(CEP290, GLIS2, INVS, IQCB1, NPHP1, NPHP3, NPHP4)*
- ☐ Nephronophthisis *(19 genes)*
- ☐ Hyperoxaluria *(AGXT, GRHPR, HOGA1)*
- ☐ Nephrotic syndrome/FSGS *(ACTN4, CD2AP, INF2, NPHS1, NPHS2, PLCE1, TRPC6, WT1)*
- ☐ Nephrotic syndrome/FSGS *(31 genes)*

Kidney Diseases (Individual Indications) [1]

- ☐ 301050 Alport syndrome *(COL4A3, COL4A4, COL4A5, MYH9)* [Q87.8]
- ☐ 113650 Branchiootorenal syndrome *(EYA1, SIX1, SIX5)* [Q87.8]
- ☐ 137920 CAKUT, congenital anomalies of the kidney and urinary tract *(HNF1B)* [Q63.9]
- ☐ 614650 Coenzyme Q10 deficiency *(COQ6)* [N04.9]
- ☐ 304800 Diabetes insipidus, nephrogenic *(AVPR2)* [N25.1]
- ☐ 603278 Focal segmental glomerulosclerosis 1 *(ACTN4)* [N04.9]
- ☐ 603965 Focal segmental glomerulosclerosis 2 *(TRPC6)* [N04.9]
- ☐ 607832 Focal segmental glomerulosclerosis 3 *(CD2AP)* [N04.9]
- ☐ 613237 Focal segmental glomerulosclerosis 5 *(INF2)* [N04.9]
- ☐ 263800 Gitelman syndrome *(SLC12A3)* [N25.1]
- ☐ 259900 Hyperoxaluria, primary *(AGXT)* [E74.8]
- ☐ 256100 Nephronophthisis 1 *(NPHP1)* [Q61.9]
- ☐ 141200 Thin-basement-membrane nephropathy *(COL4A3, COL4A4, COL4A5)* [N02.9]
- ☐ 162000 Hyperuricemic nephropathy, familial juvenile type 1 *(UMOD)*
- ☐ 256300 Nephrotic syndrome type 1 *(NPHS1)* [N04.9]
- ☐ 600995 Nephrotic syndrome type 2 *(NPHS2)* [N04.9]
- ☐ 610725 Nephrotic syndrome type 3 *(PLCE1)* [N04.9]
- ☐ 256370 Nephrotic syndrome type 4 *(WT1)* [N04.9]
- ☐ 614199 Nephrotic syndrome type 5 *(LAMB2)* [N04.9]
- ☐ 603860 Kidney disease, cysts/hyperuricaemia/isosthenuria *(UMOD)* [Q61.5]
- ☐ 173900 Polycystic kidney disease *(PKD1, PKD2)* [2] [Q61.2]
- ☐ 100100 Prune belly syndrome *(CHRM3)* [2] [Q79.4]
- ☐ 236730 Urofacial syndrome type 1 *(HPSE2)* [Q63.9]
- ☐ 615112 Urofacial syndrome type 2 *(LRIG2)* [Q63.9]

Pancreatitis (Panel Diagnostics) [1] - *alle gene auch als Einzelindikation anforderbar*

- ☐ Hereditary pancreatitis *(CASR, **CFTR, CPA1, CTRC, PRSS1, SPINK1**)* [K86.9]

RASopathies (Panel Diagnostics) [1]

- ☐ Noonan syndrome level I ***(PTPN11)***
- ☐ Noonan syndrome level II *(futher 15 genes)*
- ☐ Cardiofaciocutaneous syndrome ***(BRAF, KRAS, MAP2K1, MAP2K2)***
- ☐ LEOPARD syndrome ***(BRAF, PTPN11, RAF1)***

RASopathies (Individual Indications) [1]

- ☐ 115150 Cardiofaciocutaneous syndrome *(BRAF, KRAS, MEK1, MEK2)* [Q87.8]
- ☐ 218040 Costello syndrome *(HRAS)* [Q87.0]
- ☐ 611431 Legius syndrome *(SPRED1)* [Q85.8]
- ☐ 162200 Neurofibromatosis type 1 *(NF1)* [Q85.0]
- ☐ 601321 Neurofibromatosis-Noonan syndrome *(PTPN11,NF1)* [Q87.8]
- ☐ 163950 Noonan syndrome *(14 genes)* [Q87.1]

Reproduction Genetics (Panel Diagnostics) [1]

- ☐ Hypogonadotropic hypogonadism (Kallmann syndrome) level I: *(**ANOS1**, CHD7, FGF8, **FGFR1**, PROK2, PROKR2)* [E23]
- ☐ Hypogonadotropic hypogonadism (Kallmann syndrome) level II: *(FSHB, GNRH1, GNRHR, KISS1, KISS1R, LHB, SEMA3A, SOX10, TAC3, TACR3)* [E23]
- ☐ Hypogonadotropic hypogonadism (Kallmann syndrome) *(25 genes)* [E23]
- ☐ Premature ovarian failure, POF *(BMP15, DIAPH2, ESR1, FIGLA, FOXL2, FSHR, GDF9, INHA, LHCGR, NOBOX, NR5A1, SOHLH1, SOHLH2, STAG3)* [E28]

Reproduction Genetics (Individual Indications) [1]

- ☐ 400003 Azoospermia deletion analysis [N46]
- ☐ 277180 Congenital bilateral aplasia of vas deferens *(CFTR)* [Q55.4]
- ☐ 311360 Premature ovarian failure (POF) *(FMR1)* [N97.0]
- ☐ 201910 Adrenal hyperplasia, AGS *(CYP21A2; CYP11B1 on request)* [E25.09]
- ☐ 300068 Androgen insensitivity, AIS (*AR*) [N46]

- ☐ Prenatalis® NIPT sampling set (aneuploidy 13,18,21,X,Y)
  *MVZ Martinsried is **accredited** for the provision of NIPTs*
- ☐ Polar Body Diagnostics (PKD)/Pre-implantation Diagnostics (PID) - only after consultation
  *MVZ Martinsried is an approved **PID center***

Impaired Hearing/Deafness (Panel Diagnostics) [1]

- ☐ Deafness, autosomal dominant *(COCH, COL11A2, DFNA5, DIAPH1, KCNQ4, MYH14, WFS1)*
- ☐ Deafness, autosomal recessive 1 ***(CDH23, MYO7A, PCDH15, SLC26A4)***
- ☐ Deafness, autosomal recessive 2 *(ILDR1, MYO15A, OTOA, STRC, TMC1, TMPRSS3)*
- ☐ Syndromic deafness *(genes up to 25 kb for selection – after consultation)*
- ☐ Usher syndrome type 1 and 2 ***(MYO7A, USH2A)***
- ☐ Usher syndrome type 1 ***(CDH23, MYO7A, PCDH15**, USH1G)*
- ☐ Usher syndrome *(12 genes)*
- ☐ Deafness, all *(112 genes)*

Impaired Hearing/Deafness (Individual Indications) [1] - *bitte beachten Sie auch unser detailliertes Untersuchungsangebot Molekulargenetik*

- ☐ - Deafness, autosomal recessive, type 1A *(GJB2)*/type 1B *(GJB6)* [H91.9]
- ☐ 274600 Pendred syndrome *(SLC26A4)* [E07.1]
- ☐ Impaired hearing, autosomal recessive *(CDH23, MYO7A, PCDH15, STRC)* [H91.9]
- ☐ - Deafness, autosomal dominant *(COL11A2, MYH9)* [H91.9]
- ☐ 276901 Usher syndrome type 2A *(USH2A)* [H54.9]
- ☐ Mitochondrial deafness, "drug-induced" *(MTRNR1, MTCO1, MTTS1)* [T88.7]

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Humangenetik

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Metabolic Disorders/Endocrinology (Panel Diagnostics) [1]

Lipid Metabolism Disorders

- ☐ Hypercholesterolemia, familial *(APOB, **LDLR**, PCSK9,LDLRAP1)*
- ☐ Primary hypertriglyceridemia *(APOA5, **APOC2**, GPIHBP1, **LPL**)*
- ☐ Hypoalphalipoproteinemia *(ABCA1, **APOA1, LCAT**)*
- ☐ Hypobetalipoproteinemia *(ANGPTL3, **APOB, MTTP**, PCSK9)*
- ☐ Mixed hyperlipoproteinaemia *(APOA1, APOE, LIPC)*

Others

- ☐ Hyperoxaluria *(**AGXT, GRHPR, HOGA1**)*
- ☐ MODY diabetes *(14 genes)*
- ☐ Porphyria *(ALAD, CPOX, HMBS, PPOX, ALAS2, FECH, UROD, UROS)*
- ☐ Malignant hyperthermia *(CACNA1S, RYR1)*
- ☐ Mucopolysaccharidosis *(10 genes)*
- ☐ Congenital defects of glycosylation *(38 genes)*

Metabolic Disorders/Endocrinology (Individual Indications) [1]

- ☐ 200100 Abetalipoproteinemia *(MTP)* [E78.6]
- ☐ 201910 Congenital adrenal hyperplasia *(CYP21A2, CYP11B1 on request)* [E25.09]
- ☐ 107680 Apolipoprotein A-I deficiency *(APOA1)* [E78.6]
- ☐ 606368 Apolipoprotein A-V deficiency *(APOA5)* [E78.6]
- ☐ 144010 Apolipoprotein B-100, familial ligand-defective *(APOB)* [E78.6]
- ☐ 207750 Apolipoprotein C-II deficiency *(APOC2)* [E78.6]
- ☐ 238600 Chylomicronemia, familial *(LPL, APOC2)* [E78.3]
- ☐ 218800 Crigler-Najjar syndrome *(UGT1A1-TA-Expansion)* [E80.5]
- ☐ 304800 Diabetes insipidus, nephrogenic *(AVPR2)* [N25.1]
- ☐ 107741 Dysbetalipoproteinemia (Hyperlipoproteinemia, type III) *(APOE2/3/4)*[E78.2]
- ☐ 136120 Fish-eye disease *(LCAT)* [E78.6]
- ☐ 305900 G6PD deficiency *(G6PD)* [D55.0]
- ☐ 604091 HDL deficiency *(APOA1, LCAT, ABCA1)* [E78.6]
- ☐ 235200 Hemochromatosis *(HFE-C282Y/H63D)* [E83.1]
- ☐ 614025 Hepatic lipase deficiency *(HTGL)* [E78.4]
- ☐ 143890 Hypercholesterolemia, familial *(APOB, LDLR, PCSK9,LDLRAP1)* [E78.0]
- ☐ 259900 Hyperoxaluria, primary *(AGXT)* [E74.8]
- ☐ 605019 Hypobetalipoproteinemia, familial *(APOB)* [E78.6]
- ☐ 236250 Homocystinuria *(MTHFR-C677T/A1298C)* [E72.1]
- ☐ 245900 Lecithin: cholesterol acyltransferase deficiency *(LCAT)* [E78.6]
- ☐ 238600 Lipoprotein lipase deficiency, familial *(LPL)* [E78.9]
- ☐ 236250 MTHFR deficiency *(MTHFR-C677T/A1298C)* [E72.1]
- ☐ 143500 Meulengracht (Gilbert) syndrome *(UGT1A1-TA-Expansion)* [E80.4]
- ☐ 201450 Medium chain acyl-CoA dehydrogenase deficiency *(MCAD)* [E85.0]
- ☐ 125850 MODY type 1 *(HNF4A)* [E11.9]
- ☐ 125851 MODY type 2 *(GCK)* [E11.9]
- ☐ 600496 MODY type 3 *(HNF1A)* [E11.9]
- ☐ 606392 MODY type 4 *(PDX1)* [E11.9]
- ☐ 137920 MODY type 5 *(HNF1B)* [E11.9]
- ☐ 301500 Fabry disease *(GLA)* [E75.2]
- ☐ 230800 Gaucher disease *(GBA)* [E75.2]
- ☐ 232300 Pompe disease *(GAA)* [E74.0]
- ☐ 176100 Porphyria cutanea tarda *(UROD)* [E80.2]
- ☐ 176000 Porphyria, acute intermittent *(HMBS)* [E80.2]
- ☐ 270400 Smith-Lemli-Opitz syndrome *(DHCR7)* [Q87.1]
- ☐ 277900 Wilson disease *(ATP7B)* [E83.0]

Tumor Disposition Syndrome (Panel Diagnostics) [1]

- ☐ Breast/ovarian cancer, step I *(**BRCA1, BRCA2**, CHEK2,PALB2, RAD51C)*
- ☐ Breast/ovarian cancer, step II *(ATM, CDH1, NBN, RAD51D, TP53)*
- ☐ HNPCC (Lynch syndrome) *(**MLH1/PMS2** or **MSH2/MSH6**)*
- ☐ Gastrointestinal polyposis syndrome *(**APC, MUTYH, SMAD4, STK11, BMPR1**)*
- ☐ Familial adenomatous polyposis coli (FAP, MAP) *(**APC, MUTYH**)*
- ☐ Multiple endocine neoplasia *(**MEN1, RET, CDKN1B**)*
- ☐ Paraganglioma/Pheochromocytoma *(MAX, SDHA, SDHAF2, SDHB, SDHC, SDHD, RET, VHL)*

Tumor Disposition Syndrome (Individual Indications) [1]

- ☐ 109400 Basal cell nevus syndrome (PTCH1, PTCH2, SUFU) [1,2] [Q87.8]
- ☐ 114480 Breast cancer, familial form *(**BRCA1, BRCA2**)* [C50.9]
- ☐ 158350 Cowden syndrome 1 *(PTEN)* [Q85.8]
- ☐ 615109 Cowden syndrome 6 *(AKT1)* [Q85.8]
- ☐ 137215 Gastic cancer, hereditary *(CDH1)* [C16.9]
- ☐ 174900 Juvenile polyposis *(SMAD4, BMPR1A)* [D12.6]
- ☐ 151623 Li-Fraumeni syndrome *(TP53)* [C97]
- ☐ 155601 Malignant melanoma, familial form type 2 *(CDKN2A)* [C43.9]
- ☐ 155240 Medullary thyroid carcinoma, familial *(RET)* [C73]
- ☐ 131100 Multiple endocrine neoplasia type 1 *(MEN1)* [D44.8]
- ☐ 171400 Multiple endocrine neoplasia type 2 *(RET)* [D44.8]
- ☐ 610755 Multiple endocrine neoplasia type 4 *(CDKN1B)* [D44.8]
- ☐ 162200 Neurofibromatosis type 1 (*NF1*) [Q85.0]
- ☐ 175200 Peutz-Jeghers syndrome *(STK11)* [Q85.8]
- ☐ 191100 Tuberous sclerosis complex *(TSC1, TSC2)* [Q85.1]

Ciliopathiens (Panel Diagnostics) [1]

- ☐ Joubert syndrome *(7 genes)*
- ☐ Meckel-Gruber syndrome *(12 genes)*
- ☐ Jeune syndrome/short-rib thoracic dysplasia *(**DYNC2H1, IFT80, NEK1, TTC21B, WDR34**)*
- ☐ Senior-Løken syndrome *(7 genes)*
- ☐ Bardet-Biedl syndrome *(16 genes)*
- ☐ Orofaciodigital syndrome *(7 genes)*
- ☐ Primary ciliary dyskinesia *(CCDC39, DNAH5, DNAI1)*
- ☐ Heterotaxy *(10 genes)*
- ☐ Nephronophthisis *(7 genes)*
- ☐ Polycystic kidney disease, autosomal dominant *(BMP4, **HNF1B**, PAX2, **PKD1, PKD2**)*
- ☐ Polycystic kidney disease, autosomal recessive *(FRAS1, **PKHD1**)*
- ☐ Joubert syndrome *(19 genes)*
- ☐ Meckel-Gruber syndrome *(6 genes)*
- ☐ Jeune syndrom short-rib thoracic dysplasia *(12 genes)*
- ☐ Senior-Løken syndrome *(1 gene)*
- ☐ Bardet-Biedl syndrome *(8 genes)*
- ☐ Primary ciliary dyskinesia *(30 genes)*
- ☐ Heterotaxy *(2 genes)*
- ☐ Nephronophthisis *(19 genes)*
- ☐ Polycystic kidney disease *(12 genes)*

Cytogenetics - *please refer to our detailed cytogenetic form*

- ☐ Classical chromosome analysis (karyogram) [2]
- ☐ Microdeletions-/duplications syndrome (FISH) [2]
- ☐ Detection of deletions and duplications (qPCR) [2]
- ☐ Subtelomer diagnostics [2]
- ☐ Array CGH [1]
- ☐ Uniparental disomy (UPD) [1] from child and parents
  - ☐ 180 k
  - ☐ high resolution
  - ☐ Chrom. 14
  - ☐ Chrom. 15

Pharmacogenetics (Individual Indications) - *please refer to our detailed pharmacogenetics form*

- ☐ Gaucher disease therapy: Eliglustat (*CYP2D6*) [1]

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Pharmaco-/Nutrigenetics (Individual Indications) [1] - *for the complete range of pharmacogenetics investigations see www.medical-genetics.de*

Therapy psychiatric-neurologic disorders

- ☐ Psychotropic medications (*CYP2D6*, PM)
- ☐ Psychotropic medications (*CYP2D6*, UM)
- ☐ Psychotropic medications (*CYP2C19*, PM)
- ☐ Psychotropic medications (*CYP2C19*, UM)
- ☐ Psychotropic medications (*CYP3A5*3*, UM)
- ☐ Psychotropic medications (*CYP1A2*1F*, UM)
- ☐ Psychotropic medications (*CYP3A4*22*, PM)
- ☐ Antiepileptics (*CYP2C9*)
- ☐ L-Dopa (*COMT*)
- ☐ Multi-drug resistance (*ABCB1=MDR1*)

Therapy oncological disorders

- ☐ Azathioprine (*TPMT*)
- ☐ Irinotecan (*UGT1A1*)
- ☐ Paclitaxel (*CYP2C8*)
- ☐ 5-Fluorouracil (*DPD*)
- ☐ Tamoxifen (*CYP2D6*)

Therapy of coronary heart disease/metabolic syndromes/diabetes

- ☐ ß-blockers (*CYP2D6*, PM)
- ☐ Clopidogrel (*CYP2C19*, UM)
- ☐ Statins (*SLCO1B1*)
- ☐ Sartans (*CYP2C9*)
- ☐ Sulfonylureas (*CYP2C9*)

Therapy with muscle relaxants/local anesthesia

- ☐ Postoperative apnea (*BCHE*)

Therapy of acute and chronic pain

- ☐ NSAID (*CYP2C9*)
- ☐ Morphine (*CYP2D6*, PM)

Food intolerance

- ☐ Fructose intolerance (hereditary) aldose B deficiency *(ALDOB)*
- ☐ Fructose 1,6 bisphosphatase deficiency *(FBP1)*
- ☐ Hereditary lactose intolerance (adult onset form) *(LCT-C(-)13910T)*
- ☐ Congenital lactase deficiency *(LCT)*
- ☐ Alcohol intolerance *(ADH2, ALDH2)*

Transfusion Medicine

HLA Typing - *for the complete range of immunogenetic investigations see www.medical-genetics.de*

- ☐ High resolution HLA typing (*6 genes,* [Exon2, Exon 3]) [1]
- ☐ Ultra high resolution HLA typing (*6 genes*, [5'UTR - 3'UTR]) [1]
- ☐ KIR genotyping [1]
- ☐ Blood grouping [1]
- ☐ HLA antibody screening and specification [serum]
- ☐ HLA cross match [4] for recipient and CPDA1 blood for donor

HLA disease associations [1]	Characteristic	RR**
☐ Abacavir hypersensitivity	*B*57:01*	33 ***
☐ Allopurinol-induced Stevens-Johnson s.	*B*58:01*	580 ***
☐ Carbamazepine-ind. Stevens-Johnson s.	*B*15:02*	2504 ***
☐ Diabetes mellitus type I	*DR4/DQ3*	3.6
	DR3/DQ2	3.3
☐ Bechterew syndrome (HLA-Subtyp)	*B27*	69.1
☐ Behçet disease	*B5*	3.8

HLA disease associations [1]	Characteristic	RR**
☐ Narcolepsy (HLA subtype)	*DQB1*06:02*	129.8
☐ Psoriasis vulgaris	*Cw6*	33.0
☐ Rheumatoid arthritis, shared epitope	*DR4/DR1/DR10*	4.2
☐ Celiac disease (HLA subtype)	*DQ2/DQ8/DQA1*	11.0-52.0

* Molecular genetic nomenclature
** RR = relative risk, Tiwari JL, Terasaki PI, in HLA and Disease Association, Springer-Verlag (1985), Thorsby E, Human Immunology, 53:1 (1997)
*** Odds Ratio according to Becquemont L, Pharmacogenomics, 11:277 (2010)

Hemato-oncology

Hematological Neoplasms (Panel Diagnostics) [1,14] - *for the complete range of hemato-oncology investigations see www.medical-genetics.de*

- ☐ Acute myeloid leukemia (AML) *(17 genes)*
- ☐ Myelodysplastic syndrome (MDS) *(12 genes)*
- ☐ Myeloid panel (CMML, aCML, MPN, CNL) [MP] *(17 genes)*
- ☐ Lymphatic panel for CLL, ALL B- and T-cell lines [LP] *(13 genes)*
- ☐ Myelodysplastic syndrome (MDS) *(8 genes)*

Hematological Neoplasms (Selected Individual Indications)

- ☐ Classical chromosome analysis (karyotype) [2]
- ☐ AML fusion genes (CBFB-MYH11-, PML-RARa-, RUNX1-RUNX1T1-, KMT2A(MLL)- translocation, *DEK-NUP214* [DEK-CAN]) [1]
- ☐ AML mutation detection *(17 single genes)* [1,14]
- ☐ CLL mutation detection (IgVH mutation status, *7 single genes*) [1,14]
- ☐ Myelodysplastic syndrome (MDS) mutation detection *(9 single genes)* [14]
- ☐ FISH analysis (according to indication) [2]
- ☐ Chronic myeloid leukemia (CML) *(BCR-ABL1, ABL1)* [1,14]
- ☐ Myeloproliferative neoplasms (MPN) *(27 single genes)* [1,14]

Pathology/Neuropathology

"Targeted Cancer Panels"/Target Diagnostics [8,9] - *for the complete range of molecular pathology investigations see www.medical-genetics.de*

- ☐ Gastrointestinal stromal tumors, GIST (*KIT, PDGFRa*, Hotspots)
- ☐ Glioma basic panel (*ATRX, BRAF, CIC, EGFR, FUBP1, H3F3A, IDH1, IDH2, TERT, TP53*)*
- ☐ Glioma pediatric panel *(basic panel plus ACVR1, CTNNB1, DDX3X, HIST1H3B/C)**
  / incl. MGMT methylation status and LOH 1p/19q analysis
- ☐ Glioma master panel *(33 genes)**
- ☐ Bladder cancer (*FGFR3*, Hotspots)
- ☐ Lung cancer mutations detection (*EGFR, KRAS, NRAS, MET, ERBB2,* Hotspots)
- ☐ Lung cancer translocations *(ALK, ROS1, RET)*
- ☐ Colorectal cancer, CRC (*KRAS, NRAS, BRAF, PIK3CA*, Hotspots)
- ☐ Malignant melanoma (*BRAF, NRAS, KIT, GNAQ, GNAS*, Hotspots)
- ☐ Ovarian cancer (*BRCA1, BRCA2,* mutation search)
- ☐ Pancreatic cancer (*GNAS, KRAS,* Hotspots)
- ☐ Paraganglioma/pheochromocytoma (*MAX, SDHA, SDHAF2, SDHB, SDHC, SDHD, RET, VHL*)
- ☐ Thyroid cancer (*BRAF, HRAS, KRAS, RET, NRAS,* Hotspots)

"Companion Diagnostics" [8,9] - *for continually updated information see www.medical-genetics.de*

- ☐ *BRAF* (Bevacizumab, Binimetinib, Cetuximab, Combimetinib, Dafrabenib etc.)
- ☐ *BRCA1/BRCA2* (Olaparib) *consent according to GenDG for examination of the germline!*
- ☐ *EGFR* (Erlotinib, Gefitinib etc.)
- ☐ *KRAS* (Bevacizumab, Cetuximab, Erlotinib, Gefitinib etc.)
- ☐ *KIT* (Regorafenib, Sunitinib etc.)
- ☐ *RET* (Cabozantinib, Vandetanib etc.)

"Liquid Biopsy" [3]

- ☐ Gastrointestinal stromal tumors (GIST) (*KIT, PDGFRa*, Hotspots)
- ☐ Glioma (*IDH1, BRAF, H3F3A*, Hotspots) [11]
- ☐ Colorectal cancer (CRC) (*KRAS, NRAS, BRAF, PIK3CA, APC, P53*, Hotspots)
- ☐ Lung cancer (*EGFR, KRAS, NRAS, BRAF*, Hotspots)
- ☐ Malignant melanoma (*BRAF, NRAS, KIT*, Hotspots)

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