

Patient's Name
Date of Birth
Address
Country



LABOR LADEMANNBOGEN

MEDIZINISCHE EXPERTISE

Human Genetics

Labor Lademannbogen MVZ GmbH
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- Doctor's practice stamp -

Human Genetics (A-D)

Sampling date _____

- | | | | |
|---|----------------------|---|---------|
| <input type="checkbox"/> 1p36 deletion syndrome | Hep | <input type="checkbox"/> CADASIL (NOTCH3) | E |
| <input type="checkbox"/> 3-beta-HSD deficiency (HSD3B2) | E | <input type="checkbox"/> Cardiomyopathy, dilatative | E |
| <input type="checkbox"/> 5-fluorouracil toxicity (DPD deficiency) | E | <input type="checkbox"/> Cardiomyopathy, hypertrophic | E |
| <input type="checkbox"/> Aarskog syndrome (Faciogenital dysplasia) | E | <input type="checkbox"/> Cardiomyopathy, long-QT | E |
| <input type="checkbox"/> Aceruloplasminemia | E | <input type="checkbox"/> Cardiomyopathy, others | E |
| <input type="checkbox"/> Achondroplasia | E | <input type="checkbox"/> Carnitine palmitoyltransferase II deficiency | E |
| <input type="checkbox"/> Adiposity, Leptin | E | <input type="checkbox"/> Cataract (EPHA2, GALK) | E |
| <input type="checkbox"/> Adiposity, Leptin Receptor | E | <input type="checkbox"/> CDG syndrome, CDG 1a (PMM2) | E |
| <input type="checkbox"/> Adiposity, Melanocortin 4 Receptor | E | <input type="checkbox"/> CDG syndrome, CDG 1b (MPI) | E |
| <input type="checkbox"/> Adiposity, Proopiomelanocortin | E | <input type="checkbox"/> CDG syndrome, CDG 1c (ALG6) | E |
| <input type="checkbox"/> Adiposity, Proproteinconvertase | E | <input type="checkbox"/> CDG syndrome, CDG 2c (SLC35C1) | E |
| <input type="checkbox"/> Agammaglobulinemia, X-linked (BTK) | E | <input type="checkbox"/> Cholestasis, intrahepatic, benign recurrent (BRIC) | E |
| <input type="checkbox"/> Aicardi-Goutières syndrome | E | <input type="checkbox"/> Cholestasis, intrahepatic, of pregnancy (ICP) | E |
| <input type="checkbox"/> AIRE (APECED) | E | <input type="checkbox"/> Cholestasis, intrahepatic, progressive familial (PFIC) | E |
| <input type="checkbox"/> Alagille syndrome (JAG1) | E | <input type="checkbox"/> Chromosomal diagnosis of leukemia and lymphoma | BM etc. |
| <input type="checkbox"/> Albright osteodystrophy (GNAS1) | E | <input type="checkbox"/> Chromosomal diagnosis of spontaneous abortions | CVS |
| <input type="checkbox"/> Alpers syndrome (POLG1) | E | <input type="checkbox"/> Chromosomal diagnosis, postnatal | Hep |
| <input type="checkbox"/> Alpha 1 antitrypsin genotyping | E | <input type="checkbox"/> Chromosomal diagnosis, prenatal | AF etc. |
| <input type="checkbox"/> Alpha 1 fetoprotein (pregnancy) | S | <input type="checkbox"/> Chronic eosinophilic leukemia (CEL) | BM |
| <input type="checkbox"/> Alport syndrome | E | <input type="checkbox"/> Chronic lymphatic leukemia (B-CLL) | Hep+E |
| <input type="checkbox"/> Alzheimer disease (PSEN1/2, APP) | E | <input type="checkbox"/> Chronic myeloproliferative disorders | BM etc. |
| <input type="checkbox"/> Aminoglycoside ototoxicity (mt1555) | E | <input type="checkbox"/> CINCA syndrome (CIAS1) | E |
| <input type="checkbox"/> Amyloidosis, hereditary (APOA1) | E | <input type="checkbox"/> Cold urticaria, familial (CIAS1) | E |
| <input type="checkbox"/> Amyloidosis, hereditary (FGA) | E | <input type="checkbox"/> Collagen receptor (C807T) | E |
| <input type="checkbox"/> Amyloidosis, hereditary (LYZ) | E | <input type="checkbox"/> Colon cancer with polyposis (APC) | E |
| <input type="checkbox"/> Amyloidosis, hereditary (TTR) | E | <input type="checkbox"/> Colon cancer without polyposis (HNPCC) | E |
| <input type="checkbox"/> Androgen receptor (AR) | E | <input type="checkbox"/> COMT1 (V158M) | E |
| <input type="checkbox"/> Angelman syndrome (UBE3A) | E | <input type="checkbox"/> Congenital adrenal hyperplasia (CAH), (CYP21A1) | E |
| <input type="checkbox"/> Angioedema, hereditary | E | <input type="checkbox"/> Congenital bilateral aplasia of the vas deferens | E |
| <input type="checkbox"/> Antithrombin | E | <input type="checkbox"/> Connexin 26 (GJB2) | E |
| <input type="checkbox"/> APC resistance (Factor 5 -Leiden mutation) | E | <input type="checkbox"/> Connexin 32 (GJB1/HMSN/CMT) | E |
| <input type="checkbox"/> Apolipoprotein A1 deficiency | E | <input type="checkbox"/> Cowden syndrome (PTEN) | E |
| <input type="checkbox"/> Apolipoprotein A5 deficiency | E | <input type="checkbox"/> Cri-du-chat syndrome (5p- syndrome) | Hep |
| <input type="checkbox"/> Apolipoprotein A-V deficiency (Hyperlipidemia type I) | E | <input type="checkbox"/> Crigler-Najjar syndrome | E |
| <input type="checkbox"/> Apolipoprotein B deficiency | E | <input type="checkbox"/> Crohn disease (NOD2/CARD15) | E |
| <input type="checkbox"/> Apolipoprotein C-II deficiency (Hyperlipidemia type I) | E | <input type="checkbox"/> Cutaneous T-cell lymphomas (NAV3) | E |
| <input type="checkbox"/> Apolipoprotein E subtyping (HLP type III) | E | <input type="checkbox"/> CVID (Immunodeficiency, common variable) (TACI) | E |
| <input type="checkbox"/> Array-CGH* postnatal/prenatal ¹ | E/AF ¹ +E | <input type="checkbox"/> Cystic fibrosis (CF) | E |
| <input type="checkbox"/> Arterial tortuosity syndrome (GLUT10) | E | <input type="checkbox"/> Cystinuria | E |
| <input type="checkbox"/> Autoimmune polyendocrinopathy type I | E | <input type="checkbox"/> Cytochrome P450 (CYP2C19) | E |
| <input type="checkbox"/> Azoospermia factor | E | <input type="checkbox"/> Cytochrome P450 (CYP2C9) | E |
| <input type="checkbox"/> Bannayan-Zonana syndrome (PTEN) | E | <input type="checkbox"/> Cytochrome P450 (CYP2D6) | E |
| <input type="checkbox"/> Benign recurrent intrahepatic cholestasis | E | <input type="checkbox"/> Darier-Disease (Dyskeratosis follicularis) | E |
| <input type="checkbox"/> Birt-Hogg-Dube syndrome (FLCN) | E | <input type="checkbox"/> Deafness (Connexin 26, DFNB1) | E |
| <input type="checkbox"/> Bladder cancer (FISH) | Urine | <input type="checkbox"/> Diabetes insipidus centralis | E |
| <input type="checkbox"/> Branchiootorenal syndrome (BOR syndrome) | E | <input type="checkbox"/> Diabetes insipidus renalis | E |
| <input type="checkbox"/> Brugada syndrome (BrS), SCN5A | E | <input type="checkbox"/> Diabetes renalis (SGLT2) | E |
| <input type="checkbox"/> Butyrylcholinesterase (BCHE) | E | <input type="checkbox"/> Diabetes, MODY 1 (HNF4A) | E |
| <input type="checkbox"/> Butyrylcholinesterase deficiency | E | <input type="checkbox"/> Diabetes, MODY 10 (INS) | E |
| <input type="checkbox"/> Byler disease | E | <input type="checkbox"/> Diabetes, MODY 11 (BLK) | E |

Material: E = anticoagulated blood (EDTA, molecular genetic investigations), Hep = anticoagulated blood (Heparin, cytogenetic and FISH investigations), BM = Bone marrow, OM = Oral mucosa cells, AF = Amniotic fluid, CVS = Chorionic villus, S = Serum, *external laboratory performance, ¹we additionally need anticoagulated blood (EDTA, 2 ml) of both parents (in case of prenatal diagnosis it is mandatory)

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Human Genetics (D-K)

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<input type="checkbox"/> Diabetes, MODY 2 (Glucokinase)	E	<input type="checkbox"/> Hemolytic uremic syndrome (CFH)	E
<input type="checkbox"/> Diabetes, MODY 3 (HNF1A)	E	<input type="checkbox"/> Hemolytic uremic syndrome (CFI)	E
<input type="checkbox"/> Diabetes, MODY 4 (Insulin promotor factor-1)	E	<input type="checkbox"/> Hemolytic uremic syndrome (HUS)	E
<input type="checkbox"/> Diabetes, MODY 5 (HNF1B)	E	<input type="checkbox"/> Hemolytic uremic syndrome (MCP)	E
<input type="checkbox"/> Diabetes, MODY 6 (NEUROD1/BETA2)	E	<input type="checkbox"/> Hemolytic uremic syndrome (THBD)	E
<input type="checkbox"/> Diabetes, MODY 9 (PAX4)	E	<input type="checkbox"/> Hereditary non-polyposis colon cancer (HNPCC)	E
<input type="checkbox"/> Diabetes, permanent neonatal	E	<input type="checkbox"/> Hibernian fever (TRAPS)	E
<input type="checkbox"/> Diabetes, phosphate (PHEX)	E	<input type="checkbox"/> Hirschsprung disease	E
<input type="checkbox"/> DiGeorge syndrome (22q11.2)	Hep	<input type="checkbox"/> Huntington's disease (HD)	E
<input type="checkbox"/> Exostosis, multiple cartilaginous type 1	E	<input type="checkbox"/> Hyperbetalipoproteinemia (PCSK9)	E
<input type="checkbox"/> Exostosis, multiple cartilaginous type 2	E	<input type="checkbox"/> Hypercalcemia, hypocalciuric, familial (CASR)	E
<input type="checkbox"/> Fabry disease	E	<input type="checkbox"/> Hypercholesterinemia, autosomal dominant (PCSK9)	E
<input type="checkbox"/> Factor II	E	<input type="checkbox"/> Hypercholesterinemia, autosomal recessive	E
<input type="checkbox"/> Factor IX	E	<input type="checkbox"/> Hypercholesterinemia, familial (LDLR)	E
<input type="checkbox"/> Factor V HR2	E	<input type="checkbox"/> Hypereosinophilic syndrome (HES)	BM
<input type="checkbox"/> Factor V Leiden	E	<input type="checkbox"/> Hyperhomocysteinemia (MTHFR, C677T)	E
<input type="checkbox"/> Factor V mutation screening	E	<input type="checkbox"/> Hyper-IgD syndrome (HIDS)	E
<input type="checkbox"/> Factor VII	E	<input type="checkbox"/> Hyper-IgE syndrome, autosomal dominant (STAT3)	E
<input type="checkbox"/> Factor X	E	<input type="checkbox"/> Hyper-IgE syndrome, autosomal rezessiv (TYK2)	E
<input type="checkbox"/> Factor XI	E	<input type="checkbox"/> Hyper-IgM syndrome	E
<input type="checkbox"/> Factor XII	E	<input type="checkbox"/> Hyperinsulinism, congenital	E
<input type="checkbox"/> Factor XIII	E	<input type="checkbox"/> Hyperinsulinism-hyperammonemia syndrome	E
<input type="checkbox"/> Fanconi-Bickel syndrome (GLUT2)	E	<input type="checkbox"/> Hypermetaphase FISH (HM-FISH)	BM
<input type="checkbox"/> Favism	E	<input type="checkbox"/> Hyperoxaluria type 1	E
<input type="checkbox"/> Fibrinogen (FGA, FGB, FGG)	E	<input type="checkbox"/> Hyperparathyroidism, primary (HRPT2)	E
<input type="checkbox"/> First trimester screening	S	<input type="checkbox"/> Hyperparathyroidism, primary, severe neonatal	E
<input type="checkbox"/> Fish-eye disease	E	<input type="checkbox"/> Hypoalphalipoproteinemia APOA1, ABCA1, LCAT	E
<input type="checkbox"/> Fragile X syndrome (fra (X))	E	<input type="checkbox"/> Hypocalcemia, autosomal dominant (CASR)	E
<input type="checkbox"/> Fructose 1,6-bisphosphatase deficiency	E	<input type="checkbox"/> Hypocholinesterasemia	E
<input type="checkbox"/> Fructose intolerance, hereditary	E	<input type="checkbox"/> Hypochondroplasia	E
<input type="checkbox"/> Galactokinase deficiency	E	<input type="checkbox"/> Hypogammaglobulinaemia (CD19 deficiency)	E
<input type="checkbox"/> Galactosamine (GALT)	E	<input type="checkbox"/> Hypoparathyroidism, familial isolated (CASR)	E
<input type="checkbox"/> Galactosemia (GALK)	E	<input type="checkbox"/> Hypophosphatasia (ALPL)	E
<input type="checkbox"/> Gaucher disease	E	<input type="checkbox"/> Hypophosphatemia X-chromosomal (PHEX)	E
<input type="checkbox"/> Gilbert syndrome	E	<input type="checkbox"/> Hypophosphatemia, autosomal dominant (FGF23)	E
<input type="checkbox"/> Gilbert syndrome (Hyperbilirubinemia)	E	<input type="checkbox"/> Hypophosphatemia, autosomal rezessiv (DMP1)	E
<input type="checkbox"/> Gitelman syndrome (SLC12A3)	E	<input type="checkbox"/> Hypophosphatemic rickets, X-linked, dominant	E
<input type="checkbox"/> Glucose galactose malabsorption (SLC5A1, SGLT1)	E	<input type="checkbox"/> Ichthyoses X/steroid sulfatase (STS) deficiency	Hep
<input type="checkbox"/> Glucose transporter protein 1 syndrome (GLUT1)	E	<input type="checkbox"/> Ichthyosis vulgaris and atopic dermatitis	E
<input type="checkbox"/> Glukose-6-phosphate dehydrogenase (G6PDH)	E	<input type="checkbox"/> Idiopathic short stature*	E*/Hep
<input type="checkbox"/> Glycogen storage disease (Glycogenosis type 1a)	E	<input type="checkbox"/> IL28B genotyping	E
<input type="checkbox"/> Glycogen storage disease (Glycogenosis type 1b)	E	<input type="checkbox"/> Integrated screening with NT*	S
<input type="checkbox"/> Glycosuria, renal (SLC5A2, SGLT2)	E	<input type="checkbox"/> Integrated screening without NT*	S
<input type="checkbox"/> Gunther disease	E	<input type="checkbox"/> IPEX / XLAAD syndrome (FOXP3)	E
<input type="checkbox"/> Hailey-Hailey disease (ATP2C1-Gen)	E	<input type="checkbox"/> IRIDA (Iron-refractory iron deficiency anemia)	E
<input type="checkbox"/> HDL deficiency, familial	E	<input type="checkbox"/> Irinotecan Toxicity (UGT1A)	E
<input type="checkbox"/> Hemochromatosis type 1 (hereditary)	E	<input type="checkbox"/> Iron-refractory iron deficiency anemia (IRIDA)	E
<input type="checkbox"/> Hemochromatosis type 2A/2B (HAMP, HEPCIDIN)	E	<input type="checkbox"/> ITPA genotyping	E
<input type="checkbox"/> Hemochromatosis type 3 (TFR2)	E	<input type="checkbox"/> JAK2 (V617F)	E
<input type="checkbox"/> Hemochromatosis type 4 (SCL40A1)	E	<input type="checkbox"/> Kallmann syndrome (Mutation analysis)*	E

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Human Genetics (K-P)

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<input type="checkbox"/> Kallmann syndrome I (del(X)(p22.3))	Hep	<input type="checkbox"/> Mucopolysaccharidosis type 3B (NAGLU))	E
<input type="checkbox"/> Kearns-Sayre syndrome	E	<input type="checkbox"/> Mucopolysaccharidosis type 3D (GNS)	E
<input type="checkbox"/> Kininogen (KNG)	E	<input type="checkbox"/> Multiple endocrine neoplasia type 1 (Men1)	E
<input type="checkbox"/> Lactose intolerance	E	<input type="checkbox"/> Multiple endocrine neoplasia type 2A/B FMTC	E
<input type="checkbox"/> Langer syndrom*	E	<input type="checkbox"/> Muscular atrophy, spinal (SMA)	E
<input type="checkbox"/> LCAT deficiency	E	<input type="checkbox"/> Muscular atrophy, spinal and bulbar (SBMA)	E
<input type="checkbox"/> Leber hereditary optic neuropathy (LHON)	E	<input type="checkbox"/> Myelodysplastic syndrome (MDS)	BM
<input type="checkbox"/> Lecithin cholesterol acyltransferase deficiency	E	<input type="checkbox"/> N-acetyltransferase 1/2 (NAT1/2)	E
<input type="checkbox"/> Leigh syndrome (mt8993)	E	<input type="checkbox"/> NARP syndrome (mt8993)	E
<input type="checkbox"/> LEOPARD syndrome	E	<input type="checkbox"/> Neurofibromatosis type 1	E
<input type="checkbox"/> Leri-Weill dyschondrosteosis*	E	<input type="checkbox"/> Neurofibromatosis type 2	E
<input type="checkbox"/> Leukemia, acute lymphoblastic (ALL)	BM	<input type="checkbox"/> Non-polyposis colon cancer (HNPCC)	E
<input type="checkbox"/> Leukemia, chronic lymphocytic (B-CLL)	Hep/BM	<input type="checkbox"/> Non-syndromic hearing loss, hereditary	E
<input type="checkbox"/> Leukemia, chronic myeloid (CML)	BM etc.	<input type="checkbox"/> Noonan syndrome	E
<input type="checkbox"/> Lipoprotein lipase deficiency (Hyperlipidemia type I)	E	<input type="checkbox"/> Obesity, leptin and leptin receptor	E
<input type="checkbox"/> Loeys-Dietz syndrome (TGFB1, TGFB2)	E	<input type="checkbox"/> Obesity, melanocortin 4 receptor	E
<input type="checkbox"/> Long-chain 3-hydroxyacyl CoA dehydrog. de. (HADHA)	E	<input type="checkbox"/> Obesity, proconvertase deficiency	E
<input type="checkbox"/> Long-QT syndrome	E	<input type="checkbox"/> Obesity, proopiomelanocortin deficiency	E
<input type="checkbox"/> Lymphoma B cell receptor rearrangement	E	<input type="checkbox"/> Occipitalhorn syndrome	E
<input type="checkbox"/> Lymphoma T cell receptor rearrangement	E	<input type="checkbox"/> Ophthalmoplegia, autos. dom. progressive externe	E
<input type="checkbox"/> Lymphoma, Non-Hodgkin (CBCC lymphoma)	E	<input type="checkbox"/> Ophthalmoplegia, autos. rec. progressive external	E
<input type="checkbox"/> Lymphome follicular	E	<input type="checkbox"/> Ophthalmoplegia, chronic progressive external	E
<input type="checkbox"/> Marfan syndrome (FBN1)	E	<input type="checkbox"/> Optic atrophy, autosomal dominant (OPA1)	E
<input type="checkbox"/> Marfan syndrome type 2 (TGFB2)	E	<input type="checkbox"/> Optic atrophy, Leber	E
<input type="checkbox"/> MASS syndrome (FBN1)	E	<input type="checkbox"/> Osler-Rendu-Weber disease (ALK1, ENG)	E
<input type="checkbox"/> MCAD deficiency (MCAD)	E	<input type="checkbox"/> Osteogenesis imperfecta (COL1A1)	E
<input type="checkbox"/> McCune-Albright syndrome (GNAS1)	E	<input type="checkbox"/> Osteogenesis imperfecta (COL1A2)	E
<input type="checkbox"/> Mediterranean fever, familial (MEFV)	E	<input type="checkbox"/> p53 mutation analysis (Li-Fraumeni syndrome)	E
<input type="checkbox"/> Melanoma, familial malignant	E	<input type="checkbox"/> PAI1 (4G/5G)	E
<input type="checkbox"/> MELAS syndrome	E	<input type="checkbox"/> Pancreatitis, hereditary (PRSS1)	E
<input type="checkbox"/> Menkes syndrome	E	<input type="checkbox"/> Pancreatitis, idiopathic, chronic (PSTI, CFTR, CTRC)	E
<input type="checkbox"/> Mental retardation (Array-CGH*)	E	<input type="checkbox"/> Paraganglioma (SDHB, SDHC, SDHD)	E
<input type="checkbox"/> Mental retardation (Chromosome analysis)	Hep	<input type="checkbox"/> Parathormone deficiency	E
<input type="checkbox"/> Mevalonate kinase deficiency (MVK)	E	<input type="checkbox"/> Pendred syndrome	E
<input type="checkbox"/> Mevalonic aciduria (MVK)	E	<input type="checkbox"/> Peutz-Jeghers syndrome (Polyposis intestinal II)	E
<input type="checkbox"/> Microdeletion/-duplication syndromes	Hep	<input type="checkbox"/> Phacomatoses	E
<input type="checkbox"/> Microduplication 22q11.2	Hep	<input type="checkbox"/> Pheochromocytoma	E
<input type="checkbox"/> Miller-Dieker lissencephaly syndrome (del(17)(p13.3))	Hep	<input type="checkbox"/> Phelan-McDermid syndrome	E
<input type="checkbox"/> MODY 1 (HNF4A)	E	<input type="checkbox"/> Phenylketonuria	E
<input type="checkbox"/> MODY 10 (INS)	E	<input type="checkbox"/> Plasmacytoma (Multiples myeloma)	BM
<input type="checkbox"/> MODY 11 (BLK)	E	<input type="checkbox"/> Plasminogen activator inhibitor 1, PAI1 (4G/5G)	E
<input type="checkbox"/> MODY 2 (Glucokinase)	E	<input type="checkbox"/> Platelet collagen receptor (GP6)	E
<input type="checkbox"/> MODY 3 (HNF1A)	E	<input type="checkbox"/> Polyposis coli	E
<input type="checkbox"/> MODY 4 (insulin promoter factor-1)	E	<input type="checkbox"/> Porphyria, acute intermittent	E
<input type="checkbox"/> MODY 5 (HNF1B)	E	<input type="checkbox"/> Porphyria, congenital erythropoietic	E
<input type="checkbox"/> MODY 6 (NEUROD1/BETA2)	E	<input type="checkbox"/> Porphyria, cutanea tarda	E
<input type="checkbox"/> MODY 9 (PAX4)	E	<input type="checkbox"/> Porphyria, erythropoietic protoporphyria	E
<input type="checkbox"/> Molecular cytogenetic diagnostics (FISH)	Hep etc.	<input type="checkbox"/> Porphyria, hereditary coproporphyria	E
<input type="checkbox"/> Muckle-Wells syndrome (CIAS1)	E	<input type="checkbox"/> Porphyria, variegata	E
<input type="checkbox"/> Mucopolysaccharidosis type 2 (Hunter syndrome)	E	<input type="checkbox"/> Prader-Willi syndrome (PWS)	E

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Human Genetics (P-W)

Sampling date _____

- | | | | |
|---|-------|---|-----|
| <input type="checkbox"/> Prekallikrein (KLKB1) | E | <input type="checkbox"/> Von Hippel-Lindau syndrome | E |
| <input type="checkbox"/> Progressive familial intrahepatic cholestasis (PFIC) | E | <input type="checkbox"/> Von Recklinghausen disease | E |
| <input type="checkbox"/> Protein C (PROC) | E | <input type="checkbox"/> von Willebrand factor VWF | E |
| <input type="checkbox"/> Protein S (PROS1) | E | <input type="checkbox"/> Warfarin (VKORC1, CYP2C9) | E |
| <input type="checkbox"/> Protein Z | E | <input type="checkbox"/> Weill-Marchesani syndrome | E |
| <input type="checkbox"/> Prothrombin mutation (G20210A) | E | <input type="checkbox"/> Williams-Beuren syndrome (del(7)(7q11.23)) | Hep |
| <input type="checkbox"/> Pseudohypoparathyroidism (GNAS1) | E | <input type="checkbox"/> Wilson disease | E |
| <input type="checkbox"/> Purinergic receptor P2Y (P2RY12) | E | <input type="checkbox"/> Wiskott-Aldrich syndrome | E |
| <input type="checkbox"/> Pyruvate kinase | E | <input type="checkbox"/> Wolf-Hirschhorn syndrome (4p-syndrome) | Hep |
| <input type="checkbox"/> Rapid PCR test (Aneuploidy screening) | AF/E | <input type="checkbox"/> | |
| <input type="checkbox"/> Recklinghausen disease | E | <input type="checkbox"/> | |
| <input type="checkbox"/> Retinoschisis, juvenile,X-linked | E | <input type="checkbox"/> | |
| <input type="checkbox"/> RETT syndrom | E | <input type="checkbox"/> | |
| <input type="checkbox"/> Roberts syndrome (ESCO2) | E | <input type="checkbox"/> | |
| <input type="checkbox"/> SANDO syndrome | E | <input type="checkbox"/> | |
| <input type="checkbox"/> SCID, Omenn syndrome | E | <input type="checkbox"/> | |
| <input type="checkbox"/> Secondtrimester-screening* | S | <input type="checkbox"/> | |
| <input type="checkbox"/> Sequential screening I (Secondtrimester)* | S | <input type="checkbox"/> | |
| <input type="checkbox"/> Sequential screening II (Secondtrimester)* | S | <input type="checkbox"/> | |
| <input type="checkbox"/> SHOX (Deletion Xp22) | Hep | <input type="checkbox"/> | |
| <input type="checkbox"/> SHOX (Mutation analysis)* | E | <input type="checkbox"/> | |
| <input type="checkbox"/> Skin diseases | E | <input type="checkbox"/> | |
| <input type="checkbox"/> Smith-Lemli-Opitz syndrome | E | <input type="checkbox"/> | |
| <input type="checkbox"/> Smith-Magenis syndrome (del(17)(p11.2)) | Hep | <input type="checkbox"/> | |
| <input type="checkbox"/> Sotos syndrome (NSD1) | E/Hep | <input type="checkbox"/> | |
| <input type="checkbox"/> Sperm FISH | Ejac. | <input type="checkbox"/> | |
| <input type="checkbox"/> SRY (del(Y)(p11.3)) | Hep/E | <input type="checkbox"/> | |
| <input type="checkbox"/> Steroid-11-beta-hydroxylase deficiency (CYP11B1) | E | <input type="checkbox"/> | |
| <input type="checkbox"/> Stickler syndrome | E | <input type="checkbox"/> | |
| <input type="checkbox"/> Subtelomere diagnostics (FISH) | Hep | <input type="checkbox"/> | |
| <input type="checkbox"/> Systemic lupus erythematosus (TREX1-Gen) | E | <input type="checkbox"/> | |
| <input type="checkbox"/> Tangier disease | E | <input type="checkbox"/> | |
| <input type="checkbox"/> Telangiectasia, hereditary hemorrhagic (ALK1, ENG) | E | <input type="checkbox"/> | |
| <input type="checkbox"/> Thalassemia (Beta-thalassemia) | E | <input type="checkbox"/> | |
| <input type="checkbox"/> Thanatophoric dysplasia type I/II | E | <input type="checkbox"/> | |
| <input type="checkbox"/> Thiopurin deficiency | E | <input type="checkbox"/> | |
| <input type="checkbox"/> Thiopurin-S-methyltransferase (TPMT) | E | <input type="checkbox"/> | |
| <input type="checkbox"/> Thromboxane A2 receptor (TBXA2R) | E | <input type="checkbox"/> | |
| <input type="checkbox"/> Thyroid hormone resistance | E | <input type="checkbox"/> | |
| <input type="checkbox"/> TNF-Receptor associated periodic fever (TRAPS) | E | <input type="checkbox"/> | |
| <input type="checkbox"/> Townes-Brock syndrome | E | <input type="checkbox"/> | |
| <input type="checkbox"/> Triple test* | S | <input type="checkbox"/> | |
| <input type="checkbox"/> TTP (ADAMTS13) | E | <input type="checkbox"/> | |
| <input type="checkbox"/> Tuberous sclerosis type 1 | E | <input type="checkbox"/> | |
| <input type="checkbox"/> Tuberous sclerosis type 2 | E | <input type="checkbox"/> | |
| <input type="checkbox"/> Tyrosinemia type 1 | E | <input type="checkbox"/> | |
| <input type="checkbox"/> Uniparentale disomy | E | <input type="checkbox"/> | |
| <input type="checkbox"/> Vitamin D-dependent rickets type 1 (CYP27B1) | E | <input type="checkbox"/> | |
| <input type="checkbox"/> Vitamin D-dependent rickets type 2 (VDR) | E | <input type="checkbox"/> | |
| <input type="checkbox"/> Von Gierke disease | E | <input type="checkbox"/> | |

Material: E = anticoagulated blood (EDTA, molecular genetic investigations), Hep = anticoagulated blood (Heparin, cytogenetic and FISH investigations), BM = Bone marrow, OM = Oral mucosa cells, AF = Amniotic fluid, CVS = Chorionic villus, S = Serum, *external laboratory performance, †we additionally need anticoagulated blood (EDTA, 2 ml) of both parents (in case of prenatal diagnosis it is mandatory)