

Request for diagnostic genetic testing^a

Please send 5 – 10 ccm EDTA blood

Craniofacial and Skeletal Disorders	Neurodegenerative Disorders
<input type="checkbox"/> Antley-Bixler syndrome (<i>POR</i> , <i>FGFR2</i>) <input type="checkbox"/> Basal cell nevus syndrome (<i>PTCH1</i> ¹ , <i>PTCH2</i> ¹) <input type="checkbox"/> Branchiootorenal syndrome (<i>EYA1</i> ¹ , <i>SIX1</i> , <i>SIX5</i>) <input type="checkbox"/> Branchiooculofacial syndrome (<i>TFAP2A</i>) <input type="checkbox"/> Craniofrontonasal syndrome (<i>EFNB1</i> ¹) <input type="checkbox"/> EEC3 syndrome/split-hand/foot malformation 4 (<i>p63</i> partial) <input type="checkbox"/> Ellis-van Creveld syndrome ² (<i>EVC</i> , <i>EVC2</i>) <input type="checkbox"/> <i>FGFR</i> associated Craniosynostosis, syndromal: Pfeiffer, Crouzon, Apert, Jackson-Weiss, Muenke syndrome (<i>FGFR1</i> , 2 and 3 partial) <input type="checkbox"/> <i>FGFR</i> associated skeletal disorders: Hypo- and Achondroplasia, Thanatophoric dysplasia (<i>FGFR3</i> partial); Osteoglyphonic Dysplasia (<i>FGFR1</i>) <input type="checkbox"/> <i>FLNA</i> associated OPD spectrum skeletal disorders: FMD, OPD1+2, MNS, TOD (<i>FLNA</i> partial) <input type="checkbox"/> <i>FLNB</i> associated skeletal disorders: BD, AOI, AOIII, Larsen syndrome, SCT <input type="checkbox"/> <i>Gli3</i> associated disorders ¹ : Pallister-Hall syndrome, Greig syndrome <input type="checkbox"/> Lacrimoauriculodentodigital syndrome (<i>FGFR2</i> , <i>FGFR3</i> , <i>FGF10</i>) <input type="checkbox"/> Mandibuloacral dysplasia with type B lipodystrophy/lethal restrictive dermopathy (<i>ZMPSTE24</i>) <input type="checkbox"/> Otofaciocervical syndrome (<i>EYA1</i> ¹) <input type="checkbox"/> Popliteal Pterygium syndrome (<i>IRF6</i> ¹) <input type="checkbox"/> Saethre-Chotzen syndrome (<i>FGFR3</i> : Pro250Arg, <i>TWIST1</i>) <input type="checkbox"/> Simpson-Golabi-Behmel syndrome (<i>GPC3</i> ¹) <input type="checkbox"/> <i>SLC26A2</i> associated skeletal dysplasias: ACG1B, AOII; DTD, EDM4 <input type="checkbox"/> Treacher Collins-Franceschetti Syndrome (<i>TCOF1</i> ¹ , <i>POLR1D</i> , <i>POLR1C</i>) <input type="checkbox"/> Van der Woude Syndrome (<i>IRF6</i> ¹)	<input type="checkbox"/> Andermann syndrome/Agenesis of the corpus callosum with peripheral neuropathy (<i>KCC3</i> = <i>SLC12A6</i> ²) <input type="checkbox"/> CADASIL syndrome (<i>NOTCH3</i> ¹) <input type="checkbox"/> Cerebral cavernous malformations (<i>CCM1</i> ¹ , <i>CCM2</i> ¹ , <i>CCM3</i> ¹) <input type="checkbox"/> Frontotemporal Dementia +/- Parkinsonism (<i>MAPT</i> ¹) <input type="checkbox"/> Metachromatic leukodystrophy (<i>ARSA</i>) <input type="checkbox"/> Spastic Paraplegia 1, X-linked/MASA syndrome (<i>L1CAM</i> ¹) <input type="checkbox"/> Spastic Paraplegia 3a, autosomal dominant (<i>Atlastin</i> ¹) <input type="checkbox"/> Spastic Paraplegia 4, autosomal dominant (<i>Spastin</i> ¹) <input type="checkbox"/> Spastic Paraplegia 31, autosomal dominant (<i>REEP1</i> ¹) <input type="checkbox"/> Spastic Paraplegia 5a, autosomal recessive (<i>CYP7B1</i> ²) <input type="checkbox"/> Spastic Paraplegia 7, autosomal recessive (<i>Paraplegin</i> ²) <input type="checkbox"/> Spastic Paraplegia 20/Troyer Syndrome, autosomal recessive (<i>Spartin</i> ²) <input type="checkbox"/> Spastic Paraplegia, autosomal-recessive ² Linkage analysis for SPG5a, 7, 11, 14, 15, 20, 21, 26
	Ektodermal Dysplasias
	<input type="checkbox"/> EEC3 syndrome (<i>p63</i> partial) <input type="checkbox"/> Ectodermal an-/hypohidrotic dysplasia, autosomal dominant (<i>EDAR</i>) <input type="checkbox"/> Ectodermal an-/hypohidrotic dysplasia, autosomal recessive (<i>EDAR</i>) <input type="checkbox"/> Ectodermal an-/hypohidrotic dysplasia, X-linked (<i>EDA</i>) <input type="checkbox"/> Hay-Wells Syndrome/AEC Syndrome (<i>p63</i> partial)
	Metabolic Disorders
	<input type="checkbox"/> Benign recurrent intrahepatic cholestasis ² (<i>ATP8B1</i> , <i>ABCB11</i>) <input type="checkbox"/> Glucose-6-Phosphat Dehydrogenase-Deficiency (<i>G6PD</i>) <input type="checkbox"/> Immunodysregulation, Polyendocrinopathy, and Enteropathy, X-linked (<i>FOXP3</i>) <input type="checkbox"/> Intrahepatic cholestasis of pregnancy (<i>ABCB4</i> , <i>ATP8B1</i>) <input type="checkbox"/> Cystic Fibrosis (<i>CFTR</i> : 36 mutations, on request sequencing) <input type="checkbox"/> Progressive fam. intrahepatic cholestasis ² (<i>ATP8B1</i> , <i>ABCB11</i> , <i>ABCB4</i>) <input type="checkbox"/> Surfactant metabolism dysfunction (<i>SFTPB</i> , <i>ABCA3</i> , <i>SFTPC</i> , <i>CSF2RA</i>) <input type="checkbox"/> Trimethylaminuria (<i>FMO3</i>)
	Reproductive Genetics
	<input type="checkbox"/> Adrenal Hyperplasia, 11 β -hydroxylase deficiency (<i>CYP11B1</i>) <input type="checkbox"/> Adrenal Hyperplasia, 21-hydroxylase deficiency (<i>CYP21</i> ¹) <input type="checkbox"/> Adrenal Hyperplasia, 3 β -hydroxysteroid dehydrogenase deficiency (<i>HSD3B2</i>) <input type="checkbox"/> Adrenocortical insufficiency, <i>NR5A1</i> associated (<i>NR5A1</i>) <input type="checkbox"/> Androgen insensitivity syndrome (<i>AR</i>) <input type="checkbox"/> Kallmann syndrome (<i>KAL1</i> ¹ , <i>FGFR1</i> ¹ , <i>PROK2</i> ¹ , <i>PROKR2</i> ¹) <input type="checkbox"/> Leydig cell hypoplasia/LH resistance (<i>LHCGR</i>) <input type="checkbox"/> Ovarian hyperstimulation syndrome, spontaneous (<i>FSHR</i>)
	Other:
	<input type="checkbox"/> _____ <input type="checkbox"/> _____

^a Laboratory and Medical Practice of Human Genetics PD Dr. Hehr¹ in addition MLPA to detect exon deletions/duplications² optional prior linkage analysis for suitable families

Request for diagnostic genetic testing⁶	
Please send 5 – 10 ccm EDTA blood	
Retinal diseases	Retinal diseases
Single-gene diagnostics (accredited nach DIN EN ISO 15189)³	Module-based diagnostics of gene clusters (not accredited)⁴
<input type="checkbox"/> Achromatopsia (<i>CNGA3, CNGB3, GNAT2</i>) <input type="checkbox"/> Adult vitelliform macular dystrophy, autosomal dominant (<i>BEST1, PRPH2</i>) <input type="checkbox"/> Best vitelliform macular dystrophy, autosomal dominant (<i>BEST1</i>) <input type="checkbox"/> Bestrophinopathy, autosomal recessive (<i>BEST1</i>) <input type="checkbox"/> Vitreoretinopathopathy, autosomal dominant (<i>BEST1</i>) <input type="checkbox"/> Bietti crystalline corneoretinal dystrophy (<i>CYP4V2</i>) <input type="checkbox"/> Central areolar choroidal dystrophy (<i>PRPH2</i>) <input type="checkbox"/> Choroideremia (<i>CHM</i>) <input type="checkbox"/> Doyme honeycomb retinal dystrophy (<i>EFEMP1</i>) <input type="checkbox"/> Fundus albipunctatus (<i>PRPH2, RDH5, RHO, RLBP1</i>) <input type="checkbox"/> Gyrate atrophy (<i>OAT</i>) <input type="checkbox"/> Hypotrichosis, congenital, with juvenile macular dystrophy (<i>CDH3</i>) <input type="checkbox"/> Leber congenital amaurosis (<i>LCA5</i>) <input type="checkbox"/> Night blindness, congenital stationary, autosomal dominant (<i>GNAT1</i>) <input type="checkbox"/> Night blindness, congenital stationary, X-linked (<i>NYX</i>) <input type="checkbox"/> Norrie disease (<i>NDP</i>) <input type="checkbox"/> Optic atrophy, autosomal dominant (<i>OPA1</i>) <input type="checkbox"/> Retinitis pigmentosa, autosomal dominant (<i>RHO, RP1, PRPH2</i>) <input type="checkbox"/> Retinitis pigmentosa, X-linked (<i>RP2, RPGR includes ORF15</i>) <input type="checkbox"/> Retinoschisis, X-linked, juvenile (<i>RS1</i>) <input type="checkbox"/> Sorsby fundus dystrophy (<i>TIMP3</i>) <input type="checkbox"/> Cone dystrophy with night blindness and supernormal rod responses (<i>KCNV2</i>)	<div style="background-color: #cccccc; padding: 2px;">• Affymetrix-based resequencing array (RetChip v1.0)</div> <input type="checkbox"/> Stargardt disease (<i>ABCA4, CNGB3, ELOVL4</i>) <input type="checkbox"/> cone rod dystrophy (<i>ABCA4, AIPL1, CERKL, CNGB3, CRX, GUCA1A, GUCY2D, KCNV2, PROM1, PRPH2, RDH5, RIMS1, RPGR, RPGRIP1, SEMA4A</i>) <input type="checkbox"/> Exudative vitreoretinopathy (<i>FZD4, LRP5, NDP</i>) <input type="checkbox"/> Retinitis pigmentosa (includes adRP, arRP, XLRP, LCA and CSNB) (<i>ABCA4, AIPL1, CA4, CEP290, CERKL, CNGA1, CNGB1, CRB1, CRX, FSCN2, GRK1, GUCA1B, GUCY2D, IMPDH1, LRAT, MERTK, NR2E3, NRL, PDE6A, PDE6B, PRCD, PROM1, PRPF3, PRPF31, PRPF8, PRPH2, RDH12, RGR, RHO, RLBP1, ROM1, RP1, RP2, RP9, RPE65, RPGR, RPGRIP1, SAG, SEMA4A, TTC8, TTPA, TULP1, USH2A</i>) <div style="background-color: #cccccc; padding: 2px;">• APEX-Chip, diagnostics of known mutations (Asper Biotech, Tartu Estland)</div> <input type="checkbox"/> Leber congenital amaurosis (<i>AIPL1, CRB1, CRX, GUCY2D, LRAT, TULP1, MERTK, CEP290, RDH12, RPGRIP1, LCA5, RPE65, SPATA7</i>) <input type="checkbox"/> Usher syndrome (<i>CDH23, MYO7A, PCDH15, USH1C, USH1G, USH2A, GPR98, CLRN1, DFNB31</i>) <input type="checkbox"/> Bardet-Biedl syndrome, McKusick-Kaufman syndrome, Borjeson-Forsman-Lehmann syndrome, Alström syndrome, Albright osteodystrophy (<i>BBS1, BBS2, BBS3, BBS4, BBS5, BBS6, BBS7, BBS8, BBS9, BBS10, BBS11, BBS12, BBS13, PHF6, ALMS1, GNAS1</i>)
Hereditary cancer syndromes	Others
<input type="checkbox"/> Cowden syndrome (<i>PTEN</i>) <input type="checkbox"/> Familial adenomatous polyposis (<i>APC</i>) <input type="checkbox"/> Familial breast and ovarian cancer (<i>BRCA1, BRCA2</i>) <input type="checkbox"/> Hereditary melanoma (<i>CDKN2A</i>) <input type="checkbox"/> Hereditary non-polyposis colorectal cancer (<i>MLH1, MSH2, MSH6, PMS2</i>) <input type="checkbox"/> MYH-associated polyposis (<i>MUTYH</i>) <input type="checkbox"/> Li-Fraumeni syndrome (<i>TP53</i>) <input type="checkbox"/> Von-Hippel-Lindau syndrome (<i>VHL</i>)	<input type="checkbox"/> Failure of tooth eruption, primary (<i>PTHR1</i>) <input type="checkbox"/> _____ <input type="checkbox"/> _____
⁶ Institute of Human Genetics, University Regensburg, Prof. Weber in cooperation with Medical Practice of Human Genetics Dr. Schönbuchner ³ diagnostics via direct Sanger sequencing and MLPA ⁴ sequence variants identified by array diagnostics are verified by direct Sanger sequencing (accredited DIN EN ISO 15189)	