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Zentrum für Humangenetik · D-93042 Regensburg

Request for DNA testing

Zentrum für Humangenetik Regensburg

Version: 19.11.2013 B

Patient data

Last Name, First Name _____ DOB _____

Street _____

ZIP code _____ City/Country _____

☐ female ☐ male

Ethnic origin _____

Billing information

☐ VISA/ MasterCard (see also separate form)
☐ E112 (applicable within European Union)
☐ confirmation of payment from government office / insurance company (attach copy)
☐ bill hospital (provide exact billing address)

Clinical Data

Clinical Diagnosis: _____

Family History: ☐ Positive ☐ Negative ☐ Unknown

If positive, please provide pedigree

☐ Infectious sample: HIV, Hepatitis or other, please state _____

Referring Institution

Name of referring physician (please print) _____

Phone _____

Email address _____

Signature referring physician _____

Signature of patient (in case of minors, the parents):
With this signature I confirm, that I was fully informed about the nature and extend of the planned DNA diagnostic testing. I explicitly consent to the testing and the blood taking.

City, date _____ Signature (patient/parents) _____

Alternatively, confirmation of the referring physician:
Herewith, I confirm that the patient has consented to the DNA testing and the blood taking after the nature and extend of the DNA testing was explained.

City, date _____ Signature (patient/parents) _____

Request for diagnostic genetic testing[α]

Please send 5 – 10 ccm EDTA blood

Craniofacial and Skeletal Disorders

- ☐ Achondrogenesis IA (*TRIP11*)
- ☐ Antley-Bixler syndrome (*POR*, *FGFR2*)
- ☐ Basal cell nevus syndrome (*PTCH1*[1], on request *PTCH2*)
- ☐ Branchiootorenal syndrome (*EYA1*[1], *SIX1*, *SIX5*)
- ☐ Branchiooculofacial syndrome (*TFAP2A*)
- ☐ Craniofrontonasal syndrome (*EFNB1*[1])
- ☐ EEC3 syndrome/split-hand/foot malformation 4 (*p63* partial)
- ☐ Ellis-van Creveld syndrome[2] (*EVC*, *EVC2*)
- ☐ *FGFR* associated Craniosynostosis, syndromal: Pfeiffer, Crouzon, Apert, Jackson-Weiss, Muenke Syndrome (*FGFR1*, *2* and *3* partial)
- ☐ *FGFR* associated skeletal disorders: Hypo- and Achondroplasia, Thanatophoric dysplasia (*FGFR3* partial); Osteoglophonic Dysplasia (*FGFR1*)
- ☐ *FLNA* associated OPD spectrum skeletal disorders: FMD, OPD1+2, MNS, TOD (*FLNA* partial)
- ☐ *FLNB* associated skeletal disorders: BD, AOI, AOIII, Larsen syndrome, SCT
- ☐ *Gli3* associated disorders[1]: Pallister-Hall syndrome, Greig syndrome
- ☐ Lacrimoauriculodentodigital Syndrome (*FGFR2*, *FGFR3, FGF10*)
- ☐ Mandibulofacial dysostosis with microcephaly (*EFTUD2*)
- ☐ Microphthalmia syndromal (*SOX2*[1]*, OTX2*)
- ☐ Microtia, syndromal with cleft palate (*HOXA2*)
- ☐ Miller-Syndrome (*DHODH*)
- ☐ Nager-Syndrome (*SF3B4*)
- ☐ Otofaciocervical syndrome (*EYA1*[1])
- ☐ Saethre-Chotzen syndrome (*FGFR3*: p.Pro250Arg, *TWIST*[1])
- ☐ Simpson-Golabi-Behmel syndrome (*GPC3*[1])
- ☐ *SLC26A2* associated skeletal dysplasias: ACG1B, AOII; DTD, EDM4
- ☐ Treacher Collins-Franceschetti syndrome (*TCOF1*[1], *POLR1D, POLR1C*)
- ☐ Van der Woude/Popliteal Pterygium syndrome (*IRF6*[1])

Neurodegenerative Disorders

- ☐ NGS panel **Spastic Paraplegia** (37 HSP genes + 32 overlapping movement disorders)[HSP, 3]
- ☐ Andermann syndrome/Agenesis of the corpus callosum with peripheral neuropathy (*KCC3* = *SLC12A6*[2])
- ☐ CADASIL syndrome (*NOTCH3*[1])
- ☐ Cerebral cavernous malformations (*CCM1*[1], *CCM2*[1],*CCM3*[1])
- ☐ Frontotemporal Dementia +/- Parkinsonism (*MAPT*[1])
- ☐ Leukoencephalopathy diffuse with Spheroids (*CSF1R*)
- ☐ Metachromatic leukodystrophy (*ARSA*)
- ☐ Spastic Paraplegia 1, X-linked/MASA syndrome (*L1CAM*[1]) HSP
- ☐ Spastic Paraplegia 3a, autosomal dominant (*Atlastin*[1]) HSP
- ☐ Spastic Paraplegia 4, autosomal dominant (*Spastin*[1]) HSP
- ☐ Spastic Paraplegia 31, autosomal dominant (*REEP1*[1]) HSP
- ☐ Spastic Paraplegia 5a, autosomal recessive (*CYP7B1*[2]) HSP
- ☐ Spastic Paraplegia 7, autosomal recessive (*Paraplegin*[2]) HSP
- ☐ Spastic Paraplegia 11, autosomal recessive (*Spatacsin*[2]) HSP
- ☐ Spastic Paraplegia 15, autosomal recessive (*Spastizin*[2]) HSP
- ☐ Spastic Paraplegia 20/Troyer Syndrome, autosomal recessive (*Spartin*[2]) HSP
- ☐ Spastic Paraplegia complicated, autosomal-recessive[2] Linkage analysis for SPG5a, 7, 11, 14, 15, 20, 21 HSP, 26

Ektodermal Dysplasias

- ☐ EEC3 syndrome (*p63* partial)
- ☐ Ectodermal an-/hypohidrotic dysplasia, autosomal dominant or recessive (*EDAR*[1])
- ☐ Ectodermal an-/hypohidrotic dysplasia, X-linked (*EDA*[1])
- ☐ Hay-Wells Syndrome/AEC Syndrome (*p63* partial)

Brain Malformations/ Muscular Dystrophies

- ☐ NGS panel **HPE** (15 HPE-associated genes + 16 overlapping phenotypes)[HPE, 4]
- ☐ NGS Panel **Neuronal migration disorders** (107 genes associated with lissencephaly, Double cortex, mikrocephaly, PMG)[NM, 4]
- ☐ Andermann syndrome/Agenesis of the corpus callosum with peripheral neuropathy (*KCC3* = *SLC12A6*[2])
- ☐ *ARX*opathies NM: XLAG, Partington syndrome, XMESID, ISSX, X-linked West syndrome (*ARX*)
- ☐ Cerebral cavernous malformations (*CCM1*[1], *CCM2*[1],*CCM3*[1])
- ☐ Complex cortical dysplasia with other brain malform. NM (*TUBB3*)
- ☐ Cong. muscular dystrophies WWS/MEB[1],[2, NM] (*FKRP,LARGE,FKTN, POMT1*, *POMT2*, *POMGnT1, ISPD, COL4A1, GTDC2, TMEM5, B3GNT1, DAG1)*
- ☐ Double cortex/lissencephaly X-linked NM (*DCX*[1])
- ☐ *FOXG1*-associated encephalopathy/Rett syndrome, cong. variant NM
- ☐ Heterotaxia with cardiac and brain malformations (*ZIC3*, *NODAL*)
- ☐ Holoprosencephaly[1,HPE](*SHH*,*SIX3*,*ZIC2*,*TGIF*,*Gli2*,*PTCH*,*NODAL*)
- ☐ Hydranencephaly/ proliferative vasculopathy (*FLVCR2*)
- ☐ Hydrocephalus, X-linked NM, HSP (*L1CAM*[1])
- ☐ Infection-induced acute encephalopathy-3 (*RANBP2* partial)
- ☐ Limb-girdle muscle dystrophies[2, NM]: *FKRP, POMT1, FKTN, POMT2, POMGnT1)*
- ☐ Lissencephaly, autosomal dominant NM (*LIS1*[1]*=PAFAH1B1*, *TUBA1A, TUBG1*)
- ☐ Mental retard., X-linked, with cerebellar hypoplasia (*OPHN1*[1]) NM
- ☐ Microcephaly, primary autosomal recessive[2, NM]: MCPH5 (*ASPM*[1]), MCPH2 (*WDR62*), MCPH1 (*Microcephalin*)
- ☐ Periventricular nodular heterotopia NM (*FLNA*[1], *ARFGEF2*[2])
- ☐ Polymicrogyria, bilateral asymmetric NM (*TUBB2B*)
- ☐ Polymicrogyria, bilateral NM (*GPR56*[2], *SRPX2, TUBA8*)
- ☐ Porencephalie (*COL4A1*)
- ☐ Schizencephaly HPE (*SHH*[1], *SIX3*[1], *EMX2*)
- ☐ Septooptic dysplasia HPE (*HESX1*, *SHH*[1], *SIX3*[1])

Metabolic Disorders

- ☐ NGS panel **Liver disorders/Cholestasis** (84 genes)[CHOL, 3]
- ☐ Cystic Fibrosis (*CFTR*: 36 mutations, on request sequencing)
- ☐ Glucose-6-Phosphat Dehydrogenase-Deficiency (*G6PD*)
- ☐ IPEX syndrome (Immunodysregulation, Polyendocrinopathy, and Enteropathy, X-linked: *FOXP3*)
- ☐ Intrahepatic cholestasis of pregnancy (*ABCB4*, *ATP8B1*)
- ☐ Progressive familial/Benign recurrent intrahepatic cholestasis[2, CHOL] (*ATP8B1*, *ABCB11*, *ABCB4*)
- ☐ Surfactant metabolism dysfunction (*SFTPB, ABCA3, SFTPC, CSF2RA)*
- ☐ Trimethylaminuria *(FMO3)*

Reproductive Genetics

- ☐ Adrenal Hyperplasia, 11β-hydroxylase deficiency (*CYP11B1*)
- ☐ Adrenal Hyperplasia, 21-hydroxylase deficiency (*CYP21A2*[1])
- ☐ Adrenal Hyperplasia, 3β-hydroxysteroid dehydrogenase deficiency (*HSD3B2*)
- ☐ Adrenocortical insufficiency, *NR5A1* associated (*NR5A1*)
- ☐ Androgen insensitivity syndrome (*AR*[1])
- ☐ Kallmann syndrome (*KAL1*[1], *FGFR1*[1], *PROK2*[1], *PROKR2*[1])
- ☐ Leydig cell hypoplasia/LH resistance (*LHCGR*)
- ☐ Ovarian hyperstimulation syndrome, spontaneous (*FSHR*)

[α] Human Genetics Laboratory PD Dr. Hehr
[2] optional prior linkage analysis for suitable families
[1] in addition MLPA to detect exon deletions/duplications
[3] Targeted NGS disease panels currently not accredited

Request for diagnostic genetic testing[β]

Please send 5 - 10 ml EDTA-blood

Retinal disorders[β]

- ☐ Achromatopsia, autosomal recessive *(CNGA3, CNGB3, GNAT2*)
- ☐ Atrophia gyrata, autosomal recessive *(OAT)*
- ☐ Bietti crystalline dystrophy, autosomal recessive *(CYP4V2)*
- ☐ Choroideremia, X-chromosomal (*CHM*)[4]
- ☐ Familial exudative vitreoretinopathy (*FZD4, LRP5, NDP, TSPAN12, ZNF408*)
- ☐ Congenital stationary night blindness
  - ○ Autosomal dominant *(GNAT1, RHO, PDE6B)*
  - ○ X-chromosomal *(NYX)*
- ☐ Maculopathies
  - ○ Stargardt disease, autosomal recessive (*ABCA4*)
  - ○ Fundus albipunctatus, autosomal recessive (*RDH5*)
  - ○ Best disease, autosomal dominant *(BEST1)*
  - ○ Adult vitelliform macular dystrophy, autosomal dominant *(BEST1, PRPH2)*
  - ○ Pattern dystrophy, autosomal dominant (*PRPH2*)
  - ○ Bestrophinopathy, autosomal recessive *(BEST1)*
  - ○ Vitreoretinochoroidopathy, autosomal dominant *(BEST1)*
  - ○ Doyne honeycomb dystrophy, autosomal dominant *(EFEMP1)*
  - ○ Choroidal dystrophy, central areolar, autosomal dominant *(PRPH2, GUCY2D)*
  - ○ Sorsby fundus dystrophy, autosomal dominant *(TIMP3)*
  - ○ Macular dystrophy with hypotrichosis, autosomal recessive *(CDH3)*
- ☐ Norrie syndrome, X-chromosomal *(NDP)*
- ☐ Opticus atrophy, autosomal dominant *(OPA1)*[4]
- ☐ Retinitis pigmentosa, autosomal dominant *(PRPF31, PRPH2, RHO, RP1)*
- ☐ Retinitis pigmentosa, X-chromosomal *(RP2, RPGR inkl. ORF 15)*
- ☐ Retinoschisis, X-chromosomal *(RS1)*
- ☐ Usher Syndrom Typ IIA, autosomal recessive *(USH2A)*
- ☐ Cone dystrophy with supernormal rod responses, autosomal recessive *(KCNV2)*

Gene panel diagnostics[5]

- ☐ Gene panel Stargardt disease/macular dystrophy (*ABCA4, CNGB3, ELOVL4*)
- ☐ Gene panel Leber congenital amaurosis (LCA) (*AIPL1*, *CEP290*, *CRB1, CRX*, *GUCY2D*, *RDH12*, *RPE65*, *RPGRIP1*)
- ☐ Gene panel Cone dystrophy/Cone-rod-dystrophy (*ABCA4, ADAM9, AIPL1, CERKL, CDHR1, CNGB3, CRX, C8ORF37, GUCA1A, GUCY2D, KCNV2, PROM1, PRPH2, RDH5, RAB28, RAX2/RAXL1, RIMS1, RPGR inkl. ORF 15, RPGRIP1, SEMA4A*)

Hereditary cancer syndromes[β, 4]

- ☐ Cowden syndrome (*PTEN*), autosomal dominant
- ☐ Familial adenomatous polyposis (FAP1), autosomal dominant (*APC*)
- ☐ Familial adenomatous polyposis 2 (FAP2), autosomal recessive *(MUTYH)*
- ☐ Hereditary melanoma (*CDKN2A*), autosomal dominant
- ☐ Li-Fraumeni syndrome (*TP53*), autosomal dominant
- ☐ Von-Hippel-Lindau syndrome (*VHL*), autosomal dominant
- ☐ Familial breast and ovarian cancer, autosomal dominant[6]
  - ○ *(BRCA1)*
  - ○ *(BRCA2)*
- ☐ Hereditary non-polyposis colorectal cancer (HNPCC), autosomal dominant[6]
  - ○ (*MSH2, MSH6*)
  - ○ *(MLH1, PMS2)*

Sonstige[β]

- ☐ Primary failure of tooth eruption *(PTH1R),* autosomal dominant
- ☐ Familial amyloidosis *(TTR),* autosomal dominant
- ☐ Carrier status (known familial mutation, report required)

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☐ _____

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[β] Institut für Humangenetik der Universität Regensburg, Prof. Dr. B. Weber in Kooperation mit Praxis für Humangenetik, Dr. I. Schönbuchner; tests are performed via Sanger sequencing (accredited *DIN EN ISO 15189*)

[4] + MLPA

[5] Gene panel diagnostics are performed via NGS (not accredited) and Sanger sequencing (accredited *DIN EN ISO 15189*)

[6] Diagnostics are performed via NGS (not accredited) and Sanger sequencing (accredited *DIN EN ISO 15189*)