



Prof. Dr. med. Jürgen Kohlhase
Center for Human Genetics
Heinrich-von-Stephan-Str. 5
79100 Freiburg, Germany
Tel. +49-761-896454-0
Fax +49-761-896454-9
jkohlhase@humangenetik-freiburg.de
<http://www.humangenetik-freiburg.de>

Request form - Molecular genetic diagnostics (see reverse side for tests)

Your reference no.:
Name,
First name:
DOB (dd/mm/yyyy):
Address:

Send report to

Sender

Referring lab

Other person
(Please specify)

Patient data

Sex: male female pregnancy at present (patient or related person at risk) no yes gest. week:

History, clinical features, results of previous tests:

Family data / pedigree

Similar diseases, abortions/ stillbirths, mental or physical impairments, metabolic diseases, cystic fibrosis

Informed consent:

I give my consent that my/ my child's blood/ DNA sample will be examined for genetic changes (mutations) in the gene(s)/ with the procedure specified below related to the diseases/ clinical features described above.

Herewith I declare that I have been informed about the chances and limitations of the requested testing procedure. I was informed in detail about the consequences resulting from the test results. I agree that the sample may be stored in order to allow repetition of the tests or further related tests in the future. All data about me/ my child are subject to medical confidentiality. They can be disclosed to family members or their doctors only with my permission, but not to third parties. I'm entitled to revoke this consent at any time.

I agree that my / my child's test results/ clinical data may be used in scientific publications in anonymized form yes no

Sender (name, full postal address)

Place/ date

Signature patient/ parent

Please turn and fill out

Place/ date

Signat. physician/ genet. counselor

print name physic./ counselor

Requests for testing (update 07/04/2009) Please indicate tests required . Underline single genes ordered, if limited request. If the gene of choice is not listed, please write request on front page.

GENE(S) ALPHABETICAL	SYNDROME (DISEASE)	GENE(S) ALPHABETICAL	SYNDROME (DISEASE)
ACTA2	<input type="checkbox"/> Aortic aneurysm, thoracic familial	KCNV2	<input type="checkbox"/> cone dystrophy
AGL	<input type="checkbox"/> Glycogen storage disease 3	KIF21A	<input type="checkbox"/> CFEOM1
AGS2, AGS3, AGS4, TREX1	<input type="checkbox"/> Aicardi-Goutières syndrome	KIND1	<input type="checkbox"/> Kindler syndrome
APC, MUTYH	<input type="checkbox"/> Adenomat. Polyposis coli	KRAS	<input type="checkbox"/> CFC/ Noonan syndrome
APP	<input type="checkbox"/> Presenile Dementia (Alzheimer type)	KRT5, KRT14	<input type="checkbox"/> Epidermolysis bullosa simplex
ARSA, PSAP	<input type="checkbox"/> Metachromat. leukodystrophy	LAMB3, LAMA3, LAMC2	<input type="checkbox"/> Epidermolysis bullosa junctionalis
ARSB	<input type="checkbox"/> Mucopolysaccharidosis 6	LIFR, HSPG2	<input type="checkbox"/> Schwartz-Jampel syndrome 1+2
ARX	<input type="checkbox"/> Epilepsy, West syndrome, XLAG syndrome	LIS1, DCX, TUBA1A, ARX	<input type="checkbox"/> Lissencephaly
AVPR2, AQP2	<input type="checkbox"/> Diabetes insipidus renalis	LMNA	<input type="checkbox"/> Muscular dystrophy, Progeria
AZF1	<input type="checkbox"/> Azoospermia, Yq11 microdeletion	LMNB1	<input type="checkbox"/> Leukodystrophy, dominant
B3GALTL	<input type="checkbox"/> Peters-Plus syndrome	LMX1B	<input type="checkbox"/> Nail-Patella syndrome
BCKDHA, BCKDHB, DBT, DLD	<input type="checkbox"/> Maple syrup urine disease	MAPT (TAU), PGRN	<input type="checkbox"/> Frontotemporal dementia
BRAF, MEK1, MEK2, KRAS	<input type="checkbox"/> Cardio-Facio-Cutaneous syndrome (CFC)	MECP2	<input type="checkbox"/> Rett syndrome, Xq28 duplication
BRCA1, BRCA2	<input type="checkbox"/> Breast-/Ovarian carcinoma, familial	MED12, UPF3B	<input type="checkbox"/> FG syndrome, Lujan-Fryns syndrome
CATCH22 (DGCR)	<input type="checkbox"/> DiGeorge syndrome	MEK1, MEK2, BRAF, KRAS	<input type="checkbox"/> Cardio-Fazio-Cutanes syndrome
CDH1 (ECAD)	<input type="checkbox"/> Gastric cancer, familial	MET	<input type="checkbox"/> Renal carcinoma, papillary
CDKL5	<input type="checkbox"/> infantile spasms, epilepsy, atyp. Rett syndrome	MLH1, MSH2, MSH6, PMS2	<input type="checkbox"/> Colon carcinoma (HNPCC)
CDKN2A, CDK4	<input type="checkbox"/> Maligne melanoma, familial	MSX2	<input type="checkbox"/> Parietal foramina 1; Craniosynostosis 2
CFTR	<input type="checkbox"/> Cystic fibrosis, CBAVD/CUAVD	MUTYH (MYH)	<input type="checkbox"/> Colon carcinoma/ polyposis coli
CHD7	<input type="checkbox"/> CHARGE syndrome, Kallmann syndrome	MYCN	<input type="checkbox"/> Feingold syndrome
CHN1	<input type="checkbox"/> Duane anomaly isolated	NAGLU	<input type="checkbox"/> Mucopolysaccharidosis 3B
CHRNA2, CHRNA4, CHRNB2	<input type="checkbox"/> Frontal lobe epilepsy	NF1, SPRED1	<input type="checkbox"/> Neurofibromatosis 1, NF1-like syndrome
COL3A1	<input type="checkbox"/> Ehlers-Danlos syndrome Type 4 (vascular)	NHS	<input type="checkbox"/> Nance-Horan syndrome
COL4A1	<input type="checkbox"/> Porencephaly, stroke (early manifesting)	NKX2-5 (CSX), GATA4	<input type="checkbox"/> Congenital heart defect, Atrial septal defect
COL4A3, COL4A4	<input type="checkbox"/> Alport syndrome, autosomal recessive	NR0B1 (DAX1)	<input type="checkbox"/> X-linked congen. adrenal hyperplasia
COL4A5	<input type="checkbox"/> Alport syndrome, X-linked	P53, CHEK2	<input type="checkbox"/> Li-Fraumeni syndrome
COL5A1, COL5A2	<input type="checkbox"/> Ehlers-Danlos syndrome Type 1/2	PAX6	<input type="checkbox"/> Aniridia Type II
COL7A1	<input type="checkbox"/> Epidermolysis bullosa dystrophica	PGRN, MAPT (TAU)	<input type="checkbox"/> Frontotemporal Dementia
COL17A1, LAMB3, LAMA3, LAMC2	<input type="checkbox"/> Epidermolysis bullosa junctionalis	PITX2, FOXC1	<input type="checkbox"/> (Axenfeld)-Rieger syndrome, Iridogoniodysgenesis, Peters syndrome
CREBBP, EP300	<input type="checkbox"/> Rubinstein-Taybi syndrome	PLEC1	<input type="checkbox"/> Musc. dystrophy with Epidermolysis bullosa
CTSK	<input type="checkbox"/> Pyknodysostosis (cathepsine K-deficiency)	PLOD1+2	<input type="checkbox"/> Ehlers-Danlos syndr. 6A+B; Nevo-Bruck syn.
CX26 (GJB2)	<input type="checkbox"/> Deafness, recessive <input type="checkbox"/> Keratitis-Ichthyosis-deafness syn.	PROK2 (KAL4), PROKR2 (KAL3)	<input type="checkbox"/> Kallmann syndrome 3+4
CYP21A2, CYP11B1, HSD3B2	<input type="checkbox"/> Congenital adrenal hyperplasia	PRP	<input type="checkbox"/> Präsenile Dementia (Creutzfeld-Jacob)
DCX, LIS1, TUBA1A, ARX	<input type="checkbox"/> Lissencephaly	PRSS1, SPINK1, CFTR	<input type="checkbox"/> hereditary pancreatitis
DMD	<input type="checkbox"/> Duchenne-Becker muscular dystrophy	PSAP	<input type="checkbox"/> Metachromatic leukodystrophy; Krabbe disease
DNASE1, TREX1	<input type="checkbox"/> System. Lupus Erythematosus (SLE)	PSEN1, PSEN2	<input type="checkbox"/> Präsenile Dementia (Alzheimer)
EDA (ED1), EDARADD, EDAR	<input type="checkbox"/> Ektodermal dysplasia	PTCH1	<input type="checkbox"/> Gorlin-(Golz) syndrome (Basa cell nevus synd.)
EMX2	<input type="checkbox"/> Schizencephaly	PTPN11, SOS1, KRAS, RAF1	<input type="checkbox"/> Noonan syndrome
FAM58A	<input type="checkbox"/> STAR syndrome	RECQL4	<input type="checkbox"/> Rothmund-Thomson syndrome; Baller-Gerold syndrome; Rapadilino syndrome
FBN1, TGFB1+2, ACTA2, MYH11	<input type="checkbox"/> Marfan syndrome	RET (MEN2A+ 2B)	<input type="checkbox"/> Multiple Endocrine Neoplasia 2
FGFR1 (KAL2)	<input type="checkbox"/> Kallmann, Antley-Bixler, Jackson-Weiss, Pfeiffer syndrome	SALL1	<input type="checkbox"/> Townes-Brocks syndrome
FKRP	<input type="checkbox"/> Congenital muscular dystrophy Type 1C, Limb-Girdle Muscular Dystrophy 2I	SALL4	<input type="checkbox"/> Okinohiro syndrome
FLG	<input type="checkbox"/> Ichthyosis vulgaris	SCN1A, GABRG2	<input type="checkbox"/> Dravet syndrome (Severe Myoclonic Epilepsy)
FOXC1, PITX2	<input type="checkbox"/> (Axenfeld)-Rieger syndrome, Iridogoniodysgenesis, Peters syndrome	SCN1B, SCN1A, SCN2A, GABRG2, GABRD	<input type="checkbox"/> Epilepsy, generalised, with febrile seizures
FOXP1	<input type="checkbox"/> atyp. Rett syndrome	SDHB	<input type="checkbox"/> hereditary paraganglioma
FOXP2	<input type="checkbox"/> verbal dyspraxia	SGSH	<input type="checkbox"/> Mucopolysaccharidosis 3A (Sanfilippo A)
GABRG2, GABRD, SCN1B, SCN1A, SCN2A	<input type="checkbox"/> Epilepsy, generalised, with febrile seizures	SLC2A1 (GLUT1)	<input type="checkbox"/> Glucose transporter defect (blood-brain)
GALC, PSAP	<input type="checkbox"/> Krabbe disease	SLC26A4	<input type="checkbox"/> Taubheit, recessive (Pendred syndrome)
GALNS	<input type="checkbox"/> Mucopolysaccharidosis 4A	SLURP1	<input type="checkbox"/> Mal de Meleda
GATA4, NKX2-5	<input type="checkbox"/> Congenital heart defect, Atrial septal defect	SOS1, KRAS, RAF1, PTPN11	<input type="checkbox"/> Noonan syndrome
GBE1	<input type="checkbox"/> Glycogen storage disease 4	SPINK5	<input type="checkbox"/> Netherton syndrome
GJA1 (Cx43)	<input type="checkbox"/> Oculodentodigital dysplasia (ODDD)	SOX9	<input type="checkbox"/> Campomelic dysplasia
GLA	<input type="checkbox"/> Fabry disease	SPRED1	<input type="checkbox"/> Neurofibromatosis 1-like syndrome
GLB1	<input type="checkbox"/> Mucopolysaccharidosis 4B	STS	<input type="checkbox"/> Ichthyosis, X-linked
GLI3	<input type="checkbox"/> Greig syndrome, Pallister-Hall syndrome	TBX1	<input type="checkbox"/> DiGeorge- / Velocardiofacial syndrome
GNPTAB	<input type="checkbox"/> Mucopolysaccharidosis 2+3	TBX3	<input type="checkbox"/> Ulnar-Mammary syndrome
GNS	<input type="checkbox"/> Mucopolysaccharidosis 3D	TBX5	<input type="checkbox"/> Holt-Oram syndrome
HESX1	<input type="checkbox"/> Septo-Optische Dysplasie	TGFBR2, TGFB1	<input type="checkbox"/> Marfan-like syndrome/ Loeys-Dietz syndrome
HGSNAT (TMEM76)	<input type="checkbox"/> Mucopolysaccharidosis 3C	TGM1	<input type="checkbox"/> lamellar Ichthyosis
HOXA1	<input type="checkbox"/> Stammhirndysgenese	TNXB, COL3A1	<input type="checkbox"/> Ehlers-Danlos syndrome Typ 3
HRAS	<input type="checkbox"/> Costello syndrome	TP63 (p73L, TP73L)	<input type="checkbox"/> Ektrodaktyly (Split-Hand-Split-Foot disease)
HSPG2, LIFR	<input type="checkbox"/> Schwartz-Jampel syndrome 1, 2	TPM2	<input type="checkbox"/> Arthrogyposis multiplex congenita
IDS	<input type="checkbox"/> Mucopolysaccharidosis 2	UBE3A	<input type="checkbox"/> Angelman syndrome
IDUA	<input type="checkbox"/> Mucopolysaccharidosis 1 (Hurler/Scheie)	UPF3B, MED12	<input type="checkbox"/> FG syndrome, Lujan-Fryns syndrome
INSR	<input type="checkbox"/> insulinres. Diab. mell.; Donohue synd.; HHF5	VHL	<input type="checkbox"/> Von-Hippel-Lindau syndrome
ITGB4, ITGA6	<input type="checkbox"/> Epidermolysis bullosa junct. with pyloric atresia	WNT3	<input type="checkbox"/> Tetraamelia
KAL1, KAL2, KAL3, KAL4, CHD7	<input type="checkbox"/> Kallmann syndrome	WNT7A	<input type="checkbox"/> Fuhrmann syndrome
		Array CGH (105k / 244k Agilent)	<input type="checkbox"/> Malformation-Retardation syndrome

Sample requirements: 5-10 ml EDTA blood or DNA

We perform all genetic tests by PCR and direct DNA sequencing (exceptions: microdeletion syndromes, qPCR or MLPA; CFTR, mutation panel). If deletions are described for any gene, we offer deletion analysis either by Real-Time PCR (Borozdin et al. 2004, 2006; Böhm et al. Hum Mutat 2004, Unger et al. 2008) or MLPA. Further test can be offered upon request (Phone +49-761-8964540 or jkohlhase@humangenetik-freiburg.de).