

Krankenkasse bzw. Kostenträger		MVZ Labor Prof. Seelig		Tagesnummer (bitte frei lassen)	
Name, Vorname des Versicherten		Kriegsstr. 99, 76133 Karlsruhe Postfach 56 09, 76038 Karlsruhe Telefon (07 21) 8 50 00 - 0 Fax (07 21) 85 00 01 99			
geb. am		Probenentnahme Mann <input type="checkbox"/> Frau <input type="checkbox"/>		Materialien	
Kassen-Nr.		Datum: _____		<input type="checkbox"/> Vollblut <input type="checkbox"/> CSF <input type="checkbox"/> Liquor <input type="checkbox"/> Bi <input type="checkbox"/> Biopsie <input type="checkbox"/> Vollblut, zentr <input type="checkbox"/> Syn <input type="checkbox"/> Synovia <input type="checkbox"/> St <input type="checkbox"/> Stuhl <input type="checkbox"/> S <input type="checkbox"/> HB <input type="checkbox"/> HP <input type="checkbox"/> Hep.-Blut <input type="checkbox"/> A <input type="checkbox"/> Abstrich <input type="checkbox"/> EB <input type="checkbox"/> EDTA-Blut <input type="checkbox"/> HP <input type="checkbox"/> Hep.-Plasma <input type="checkbox"/> HCY <input type="checkbox"/> HCY-Z-Gel <input type="checkbox"/> EP <input type="checkbox"/> EDTA-Plasma <input type="checkbox"/> U24 <input type="checkbox"/> 24h-S-Urin <input type="checkbox"/> FB <input type="checkbox"/> NaF-Blut <input type="checkbox"/> CB <input type="checkbox"/> EDTA-Plasma <input type="checkbox"/> U <input type="checkbox"/> Urin <input type="checkbox"/> HL <input type="checkbox"/> Hämolytat <input type="checkbox"/> CP <input type="checkbox"/> Citrat-Blut <input type="checkbox"/> S+L <input type="checkbox"/> Serum+Liquor <input type="checkbox"/> Spu <input type="checkbox"/> Sputum <input type="checkbox"/> Ma <input type="checkbox"/> Mat. angeben:	
Versicherten-Nr.		Körpergröße: _____ [cm]			
Status		Körpergewicht: _____ [kg]			
Betriebsstätten-Nr.		Urinvolumen: _____ [mL]			
Arzt-Nr.		Sammelperiode: _____ [Std]			
Datum					
<input type="checkbox"/> Privat amb. <input type="checkbox"/> Kostenträger <input type="checkbox"/> Ambulante Patienten <input type="checkbox"/> Gutachten / <input type="checkbox"/> BG-Patient <input type="checkbox"/> Privat stat. <input type="checkbox"/> Krankenhaus <input type="checkbox"/> Überweisungsschein genügt <input type="checkbox"/> Aktenzeichen <input type="checkbox"/> BG-Anschrift angeben				* 4 - 8 °C, ** -20 °C, *** lichtgeschützt	

Klinische Fragestellung:	Dringend erforderlich: =>	Absender (Stempel)
	Befundanschrift und Rechnungsanschrift	

4 a Molekulare Humangenetik

Mat	Stoffwechsel	Mat	Neurologie
EB <input type="checkbox"/> α1-Antitrypsin-Mangel, S-/Z-Allel	EB <input type="checkbox"/> Mukoviszidose, CFTR	EB <input type="checkbox"/> CADASIL, NOTCH3	EB <input type="checkbox"/> Spinocerebelläre Ataxie 1, SCA1
EB <input type="checkbox"/> Carnitin Palmitoyltransferase 2-Mangel, CPT2	EB <input type="checkbox"/> Pankreatitis hereditäre, PRSS1	EB <input type="checkbox"/> Chorea hereditäre benigne, TITF1	EB <input type="checkbox"/> Spinocerebelläre Ataxie 2, SCA2
EB <input type="checkbox"/> Cholestase intrahepatische familiäre Typ 1, ATP8B1	EB <input type="checkbox"/> Pankreatitis hereditäre chronische, SPINK1	EB <input type="checkbox"/> Chorea Huntington, HTT	EB <input type="checkbox"/> Spinocerebelläre Ataxie 3, SCA3
EB <input type="checkbox"/> Cholestase intrahepatische familiäre Typ 2, ABCB11	EB <input type="checkbox"/> Phenylketonurie, PAH	EB <input type="checkbox"/> Creutzfeld-Jakob Erkrankung, PRNP	EB <input type="checkbox"/> Spinocerebelläre Ataxie 6, SCA6
EB <input type="checkbox"/> Crigler-Najjar-Syndrom, UGT1A1	EB <input type="checkbox"/> Proteinose alveoläre congenitale, SFTPB	EB <input type="checkbox"/> Dentatorubropallidolusian Atrophie DRPLA	EB <input type="checkbox"/> Tuberöse Hirnsklerose 1, TSC1
EB <input type="checkbox"/> Fruktoseintoleranz hereditäre, ALDOB	EB <input type="checkbox"/> Pyruvatkinase-Mangel, PKLR	EB <input type="checkbox"/> Dystonie Parkinson, ATP1A3	EB <input type="checkbox"/> Tuberöse Hirnsklerose 2, TSC2
EB <input type="checkbox"/> Galaktosämie, GALT	Mat Diabetes mellitus	EB <input type="checkbox"/> Dystonie Parkinson, ATP1A3	EB <input type="checkbox"/> Paralyse periodische hypokaliämische, SCN4A
EB <input type="checkbox"/> 21-Hydroxylase-Mangel, CYP21A2	EB <input type="checkbox"/> MODY Typ 1, HNF4A	EB <input type="checkbox"/> Epilepsie generalisierte, SCN1A	EB <input type="checkbox"/> Paralyse periodische hyperkaliämische, CACNA1S
EB <input type="checkbox"/> Hyperoxalurie Typ 1, AGXT1	EB <input type="checkbox"/> MODY Typ 2, GCK	EB <input type="checkbox"/> Episodische Ataxie 1, KCNA1	EB <input type="checkbox"/> Torsionsdystonie generalisierte, DYT1
EB <input type="checkbox"/> Hyperparathyreoidismus familiärer, CASR	EB <input type="checkbox"/> MODY Typ 3, TCF1	EB <input type="checkbox"/> Episodische Ataxie 2, CACNA1A	EB <input type="checkbox"/> Morbus Alexander, GFAP
EB <input type="checkbox"/> Hyperthermie maligne, RYR1	EB <input type="checkbox"/> MODY Typ 4, IPF1	EB <input type="checkbox"/> Friedreich Ataxie, FRDA	Mat Mentale Retardierung
EB <input type="checkbox"/> Hypoparathyreoidismus familiärer, CASR	EB <input type="checkbox"/> MODY Typ 5, TCF2	EB <input type="checkbox"/> Hereditäre motorisch sensible Neuropathie 1 / CMT1A, PMP22	EB <input type="checkbox"/> Angelman Syndrom, UBE3A
EB <input type="checkbox"/> Hypoparathyreoidismus familiärer, GATA3	EB <input type="checkbox"/> MODY Typ 6, NEUROD1	EB <input type="checkbox"/> Hereditäre motorisch sensible Neuropathie 1 / CMT1B, MPZ	EB <input type="checkbox"/> DiGeorge-Syndrom, DGS
EB <input type="checkbox"/> Laktat-Dehydrogenase-Mangel, LHDA	EB <input type="checkbox"/> Mitochondrialer Diabetes, MTTL1	EB <input type="checkbox"/> Hereditäre motorisch sensible Neuropathie 2 / CMT2A, MFN2	EB <input type="checkbox"/> Fragiles X-Syndrom, FMR1
EB <input type="checkbox"/> Laktat-Dehydrogenase-Mangel, LDHB	Mat Porphyrie	EB <input type="checkbox"/> Hereditäre motorisch sensible Neuropathie 2 / CMT2B, LMNA	EB <input type="checkbox"/> Noonan-Syndrom, PTPN11
EB <input type="checkbox"/> Laktoseintoleranz hereditäre, LCT	EB <input type="checkbox"/> P. akute intermittierende, HMBS	EB <input type="checkbox"/> Hereditäre motorisch sensible Neuropathie 2 / CMT2D, GARS	EB <input type="checkbox"/> Noonan-Syndrom, SOS1
EB <input type="checkbox"/> Medium-chain acyl-CoA-Mangel, ACADM	EB <input type="checkbox"/> P. chronisch hepatische, UROD	EB <input type="checkbox"/> Hereditäre motorisch sensible Neuropathie 2 / CMT2D, GARS	EB <input type="checkbox"/> Prader-Willi-Syndrom, SNRPN
EB <input type="checkbox"/> Menkes-Syndrom, MNK	EB <input type="checkbox"/> P. congenitale erythroetische, UROS	EB <input type="checkbox"/> Hereditäre Neuopathie mit Druckpresen, PMP22	EB <input type="checkbox"/> RETT-Syndrom, MECP2
EB <input type="checkbox"/> Morbus Fabry, GLA	EB <input type="checkbox"/> P. cutanea tarda, UROD	EB <input type="checkbox"/> Hereditäre Neuopathie mit Druckpresen, PMP22	HB <input type="checkbox"/> Karyotypisierung
EB <input type="checkbox"/> Morbus Gaucher, GBA	EB <input type="checkbox"/> P. variegata, PPOX	EB <input type="checkbox"/> Kallmann-Syndrom 1, KAL1	Mat Demenz
EB <input type="checkbox"/> Morbus Meulengracht, UGT1A1	EB <input type="checkbox"/> P. Doss, ALAD	EB <input type="checkbox"/> Kallmann-Syndrom 2, FGFR2	EB <input type="checkbox"/> APOE
EB <input type="checkbox"/> Morbus Pompe, GAA	EB <input type="checkbox"/> P. erythropoetische, FECH	EB <input type="checkbox"/> Lateralsklerose amyotrophe, SOD1	EB <input type="checkbox"/> PSEN1
EB <input type="checkbox"/> Morbus Sandhoff, HEXB	EB <input type="checkbox"/> Koproporphyrin hereditäre, CPO	EB <input type="checkbox"/> Makula-Degeneration senile, CFH	EB <input type="checkbox"/> PSEN2
EB <input type="checkbox"/> Morbus Tay-Sachs, HEXA		EB <input type="checkbox"/> Optikusneuropathie autosomal dominante, OPA1	
EB <input type="checkbox"/> Morbus Wilson, ATP7B		EB <input type="checkbox"/> Paraplegie spastische 1, L1CAM	
		EB <input type="checkbox"/> Paraplegie spastische 2, PLP	
		EB <input type="checkbox"/> Paraplegie spastische 3, SPG3	
		EB <input type="checkbox"/> Paraplegie spastische 4, SPG4	
		EB <input type="checkbox"/> Spinobulbäre Muskelatrophie Kennedy, SBMA	
		EB <input type="checkbox"/> Spinale Muskelatrophie, SMN1	



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Molekulare Humangenetik - Seite 1

Weitere Untersuchungen siehe Untersuchungsprogramm.

4 a Molekulare Humangenetik

Mat	Hämатologie	Mat	Onkologie	Mat	Nieren	Mat	Mitochondrien
EB	<input type="checkbox"/> Atransferrinämie congenitale, TF	EB	<input type="checkbox"/> HNPCC, MLH1	EB	<input type="checkbox"/> Alport-Syndrom, COL4A5	EB	<input type="checkbox"/> MELAS-Syndrom, MTTL1
EB	<input type="checkbox"/> Erythrozytose familiäre, EPOR	EB	<input type="checkbox"/> HNPCC, MSH2	EB	<input type="checkbox"/> Hyperkalzämie, hypokalziurische familiäre, CASR	EB	<input type="checkbox"/> MERRF-Syndrom, MTTK
EB	<input type="checkbox"/> Erythrozytose familiäre, EGLN1	EB	<input type="checkbox"/> HNPCC, MSH6	EB	<input type="checkbox"/> Bartter Syndrom 1, SLC12A1	EB	<input type="checkbox"/> Diabetes-Deafness-Syndrom, MTTL1
EB	<input type="checkbox"/> Erythrozytose familiäre, EPAS1	EB	<input type="checkbox"/> HNPCC, PMS1	EB	<input type="checkbox"/> Bartter Syndrom 2, KCNJ1	EB	<input type="checkbox"/> Leber'sche Optikusatrophy, MTND1,4,6,-MTCYB
EB	<input type="checkbox"/> Glukose-6-Phosphat Dehydrogenase-Mangel, G6PD	EB	<input type="checkbox"/> HNPCC, PMS2	EB	<input type="checkbox"/> Diabetes insipidus renalis 1, AVPR2		
EB	<input type="checkbox"/> Hämochromatose 1, HFE	EB	<input type="checkbox"/> LiFraumeni-Syndrom, TP53	EB	<input type="checkbox"/> Diabetes insipidus renalis 2, AQP2	Mat	Immundefekte
EB	<input type="checkbox"/> Hämochromatose 2a, HJV	EB	<input type="checkbox"/> Multiple endokrine Neoplasien 1, MEN1			EB	<input type="checkbox"/> Agammaglobulinämie Bruton, BTK
EB	<input type="checkbox"/> Hämochromatose 2b, HAMP	EB	<input type="checkbox"/> Multiple endokrine Neoplasien 2a, RET	Mat	Skelettmuskel	EB	<input type="checkbox"/> Agammaglobulinämie Non-Bruton, IGMM
EB	<input type="checkbox"/> Hämochromatose 3, TFR2	EB	<input type="checkbox"/> Peutz-Jeghers-Syndrom, STK11	EB	<input type="checkbox"/> Muskeldystrophie Duchenne-Becker, Dystrophingen	EB	<input type="checkbox"/> Agammaglobulinämie Non-Bruton, IGLL1
EB	<input type="checkbox"/> Hämochromatose 4, HFE4	EB	<input type="checkbox"/> Polyposis coli familiäre, APC	EB	<input type="checkbox"/> Myotone Dystrophie 1, DMPK	EB	<input type="checkbox"/> Agammaglobulinämie Non-Bruton, VPRED1
EB	<input type="checkbox"/> Megaloblastische Anämie, CUBN	EB	<input type="checkbox"/> Polyposis coli juvenile, BMPR1A	EB	<input type="checkbox"/> Myotone Dystrophie 2, ZNF9	EB	<input type="checkbox"/> Granulomatose chronische, CYBB
EB	<input type="checkbox"/> Mikrozytäre Anämie congenitale, SLC11A2	EB	<input type="checkbox"/> Polyposis coli familiäre, MUTYH	EB	<input type="checkbox"/> Muskeldystrophie okulopharyngeale, PABPN1		
EB	<input type="checkbox"/> Sideroblastische Anämie, ALAS2	EB	<input type="checkbox"/> Schilddrüsenkarzinom familiäres medulläres, RET	EB	<input type="checkbox"/> Myotonie congenitale Becker, CLCN1	Mat	Bindegewebe - Skelett
EB	<input type="checkbox"/> Sphärozytose hereditäre, ANK1	EB	<input type="checkbox"/> Von-Hippel-Lindau-Syndrom, VHL	EB	<input type="checkbox"/> Myotonie congenitale Thomsen, CLCN1	EB	<input type="checkbox"/> Achondroplasie, FGFR3
EB	<input type="checkbox"/> Thalassämien, Globingene			EB	<input type="checkbox"/> Paramyotonia congenita, SCN4A	EB	<input type="checkbox"/> Kraniosynostosen, FGFR1/2
EB	<input type="checkbox"/> Thrombotische thrombozytopen. Purpura, ADAMTS13	Mat	Periodische Fiebersyndrome	EB	<input type="checkbox"/> Long QT-Syndrom 1, KCNQ1	EB	<input type="checkbox"/> Marfan-Syndrom, FBN1
		EB	<input type="checkbox"/> Hyper-IgD-Syndrom, MVK	EB	<input type="checkbox"/> Long QT-Syndrom 2, KCNH2	EB	<input type="checkbox"/> Progerie, LMNA
Mat	Fertilitätsstörungen	EB	<input type="checkbox"/> Mittelmeerfieber familiäres, MEFV	EB	<input type="checkbox"/> Long QT-Syndrom 3, SCN5A	EB	<input type="checkbox"/> Morbus Osler Typ 1, ENG
EB	<input type="checkbox"/> Azoospermie, AZF-Loci	EB	<input type="checkbox"/> Muckle-Wells-Syndrom, CIAS1	EB	<input type="checkbox"/> Long QT-Syndrom 5, KCNE1	EB	<input type="checkbox"/> Morbus Osler Typ 2, ALK1
EB	<input type="checkbox"/> Congenitale Aplasie des Vas deferens, CFTR	EB	<input type="checkbox"/> Periodisches Fieber familiäres, TNFRSF1A	EB	<input type="checkbox"/> Long QT-Syndrom 6, KCNE2	Mat	Pharmakogenetik
EB	<input type="checkbox"/> Androgenresistenz, AR			EB	<input type="checkbox"/> Kardiomyopathie hypertrophe familiäre, TNNT2	EB	<input type="checkbox"/> 5FU-Toxizität, DPYD
EB	<input type="checkbox"/> Östrogenresistenz, ESR1			EB	<input type="checkbox"/> Long QT-Syndrom 1, KCNQ1	EB	<input type="checkbox"/> Azulfidinsensitivität, TPMT
EB	<input type="checkbox"/> XX-male-Syndrom, SRY			EB	<input type="checkbox"/> Long QT-Syndrom 2, KCNH2	EB	<input type="checkbox"/> Irinotecan-Toxizität, UGT1A1
EB	<input type="checkbox"/> XY-female-Syndrom, SRY			EB	<input type="checkbox"/> Brugada-Syndrom 3, SCN5A	EB	<input type="checkbox"/> Narkose-Verträglichkeit, BCHE
EB	<input type="checkbox"/> Swyer-Syndrom, SRY			EB	<input type="checkbox"/> Long QT-Syndrom 5, KCNE1	EB	<input type="checkbox"/> Warfarin-Resistenz, VKORC1
HB	<input type="checkbox"/> Karyotypisierung			EB	<input type="checkbox"/> Long QT-Syndrom 6, KCNE2		

Dispositionen

Mat	Myokardinfarkt	Mat	Hyperlipidämie	Mat	Thrombophilie	Mat	Osteoporose
EB	<input type="checkbox"/> ACE-Genotyp	EB	<input type="checkbox"/> APOB 100-Mutationen	EB	<input type="checkbox"/> AT III-Mutationen	EB	<input type="checkbox"/> CALCR-Mutation
EB	<input type="checkbox"/> Fibrinogen-Rezeptor-Mutation	EB	<input type="checkbox"/> APOE-Allele	EB	<input type="checkbox"/> Faktor III/Prothrombin-Mutation	EB	<input type="checkbox"/> COL1A1-Mutation
EB	<input type="checkbox"/> Integrin-α2-Mutation	EB	<input type="checkbox"/> APOA5-Mutationen	EB	<input type="checkbox"/> Faktor V Leiden-Mutation	EB	<input type="checkbox"/> ESR1-Mutation
EB	<input type="checkbox"/> MTHFR-Mutation	EB	<input type="checkbox"/> APOC2-Mutationen	EB	<input type="checkbox"/> Faktor V Cambridge-Mutation	EB	<input type="checkbox"/> IL6-Mutation
EB	<input type="checkbox"/> PAI-Mutation	EB	<input type="checkbox"/> CETP-Mutationen	EB	<input type="checkbox"/> Faktor V HongKong-Mutation	EB	<input type="checkbox"/> VDR-Mutation
EB	<input type="checkbox"/> Arteriosklerose-Disposition Selektin E-Gen, S128R, L554F	EB	<input type="checkbox"/> LDLR-Mutationen	EB	<input type="checkbox"/> Faktor V Liverpool-Mutation		
		EB	<input type="checkbox"/> LPL-Mutationen	EB	<input type="checkbox"/> Faktor V Ferrara-Mutation		
				EB	<input type="checkbox"/> PROC-Mutationen		
				EB	<input type="checkbox"/> PROC-Mutationen		

Einwilligungserklärung des Patienten gemäß §8 Abschnitt 2 des Gendiagnostikgesetzes

Mit meiner Unterschrift bestätige ich mein Einverständnis mit der/den angeforderten genetischen Analyse/n. Über Wesen, Bedeutung und Tragweite der Untersuchung bin ich informiert worden. Mir ist bekannt, dass ich die Einwilligung jederzeit widerrufen oder von der Mitteilung des Befundes Abstand nehmen kann. Ich bin einverstanden, dass verbleibendes genetisches Material zum Zwecke der Nachprüfbarkeit der Ergebnisse aufbewahrt wird.

Ort, Datum _____ Unterschrift _____



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Molekulare Humangenetik - Seite 2