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**Patient information**

Patient Sticker / Please fill out completely

Name + initials

Date of birth  
(d/m/y)

Sex

INVOICE WILL BE SENT TO THE REFERRING PHYSICIAN

Physician	:		City	:	
Hospital/institution	:		Country	:	
Department	:				
Address	:		Telephone	:	
ZIP code	:		Email	:	

PLEASE USE A SEPARATE FORM FOR EACH PATIENT !

► **Sample (blood is strongly preferred)**

- blood  DNA (~50 µg)  other
- date of withdrawal:* *extracted from (tissue):* *namely:*

► **Conditions and Shipment**

- 2 x10 ml EDTA Blood in plastic tubes (no glass). Neonates at least 3 ml.
- Put name, gender and date of birth on each blood- or DNA tube. Improperly labelled samples will be refused.
- Ship samples at room temperature. **Do not freeze blood samples!**
- Samples should arrive in our lab within 5 days after withdrawal.
- If other material than blood (incl. DNA) is used, the handling time may be longer than indicated.
- In case you send in extracted DNA please make sure that amount of DNA is sufficient (~50 ug)

► **Informed consent. Please indicate whether the patient agrees.**

- The patient or his/her legal representative does not object to further use of the material for research in line with the current diagnostic question, and would like to be informed about the possible outcomes if they are relevant for the patient.
- The patient or his/her legal representative does object to further use of the material for research in line with the current diagnostic question.

► **Reason for referral**

- Confirmation of diagnosis
- Exclusion of diagnosis
- Carrier testing
- Analysis of a known familial mutation
- Prenatal testing (only after consultation)
- Other :

*To be filled out by our staff*

Datum ontvangst:

Opmerkingen:

Paraaf Sec.	
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► **Relevant clinical information / Remarks**

- Are there other affected family members? Yes / No  
 Has material from family member(s) been sent in previously? Yes / No

**If so:**

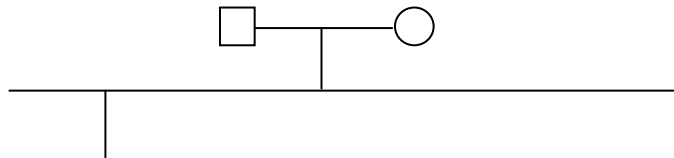
- Name previously tested family member:
- Date of birth previously tested family member:
- Relation to current patient:
- Date of birth parents of the patient, father: mother:
- Number of siblings of patient (see pedigree):
- Our Reference previous request (if known):
- Gene involved and mutation (if known):

*DNA Diagnostics Nijmegen reserves the right not to accept the request if this information is missing*

**Please mark individual of the present request with arrow (→)**

Designate affected family members as ■ / ●.

Indicate previous sent family samples with name and date of birth.



► **Pedigree information:**

Nr.	Name	M/F	Date of birth

## Turn around time (TAT)

Prenatal diagnosis	2 - 3 weeks
Testing of a known mutation in the family	4 weeks
Testing for of a yet unknown mutation in the family	see table (months)

Disease	Gene	TAT (months)	Disease	Gene	TAT (months)
□ Aarskog-Scott syndrome	<i>FGD1</i>	2	□ Complex V deficiency, autosomal recessive <sup>6</sup>	<i>ATP12</i>	6
Achromatopsia			Complex (oxphos) deficiency, combined		
□ type 2, <i>CNGA3</i>	<i>CNGA3</i>	2	□ <i>GFM1</i> □ <i>MRPS16</i> □ <i>PUS1</i> □ <i>Twinkle</i>		6
□ type 3, <i>CNGB3</i>	<i>CNGB3</i>	2	□ Congenital indifference to pain	<i>SCN9A</i>	3
Adrenal hyperplasia, congenital			Costello syndrome		
□ 21-hydroxylase deficiency	<i>CYP21A2</i>	2	□ <i>HRAS</i>	<i>HRAS</i>	1
□ 11-b hydroxylase deficiency	<i>CYP11B1</i>	2	□ <i>KRAS</i>	<i>KRAS</i>	1
□ 3-b hydroxysteroid dehydrog. def. (type II)	<i>HSD3B2</i>	2	□ Cowden disease	<i>PTEN</i>	2
□ p450c17 and p450c21 deficiency	<i>POR</i>	2	Cysteinuria		
□ Adrenoleukodystrophy	<i>ABCD1</i>	2	□ <i>SLC3A1</i> □ <i>SLC7A9</i>		2
□ ADULT syndrome	<i>TP63</i>	2	□ Cystinosis, nephropathic	<i>CTNS</i>	2
□ AEC syndrome (Hay-Wells)	<i>TP63</i>	2			
□ Alpers syndrome	<i>POLG</i>	6	Deafness, autosomal dominant		
□ Amyotrophic lateral sclerosis 4, juvenile (ALS4)	<i>SETX</i>	2	□ type 2 (DFNA2) <sup>2</sup>	<i>KCNQ4</i>	2
□ Anemia, hypochromic microcytic	<i>SLC11A2</i>	6	□ type 9 (DFNA9) <sup>2</sup>	<i>COCH</i>	2
□ Anemia, refractory iron deficiency <sup>1</sup>	<i>TMPRSS6</i>	6	□ type 6/14 (DFNA6/14)	<i>WFS1</i>	2
Angelman syndrome			□ type 8/12 (DFNA8/12) <sup>2</sup>	<i>TECTA</i>	2
□ Methylation		2	Deafness, autosomal recessive		
□ <i>UBE3A</i>		2	□ type 1 (DFNB1)	<i>GJB2/6</i>	2
□ Arts Syndrome	<i>PRPS1</i>	2	□ type 8/12 (DFNB8/12)	<i>TMPRSS3</i>	2
□ Aromatic L-aminoacid decarboxylase def. (AADC)	<i>DDC</i>	2	□ type 21 (DFNB21) <sup>2</sup>	<i>TECTA</i>	2
□ Ataxia, Friedreich (FRDA) <sup>3</sup>	<i>FXN</i>	2	Deafness, X-linked		
□ Ataxia + isolated vitamin E deficiency (AVED)	<i>TTPA</i>	2	□ type 1 (DFN1)	<i>TIMM8A</i>	2
□ Ataxia-oculomotor apraxia type 2 (AOA2)	<i>SETX</i>	2	□ type 3 (DFN3)	<i>POU3F4</i>	2
□ Ataxia, spastic, Charlevoix-Saguenay (ARSACS) <sup>5</sup>	<i>SACS</i>	2	□ Dent disease (nephrolithiasis type 1)	<i>CLCN5</i>	2
			Diabetes insipidus,		
□ Bannayan-Riley-Ruvalcaba syndrome	<i>PTEN</i>	2	□ central	<i>AVP</i>	2
□ Bardet-Biedl syndrome <sup>10</sup>			□ nephrogenic, X-linked	<i>AVPR2</i>	2
Bartter syndrome			□ nephrogenic, autosomal dominant/recessive	<i>AQP2</i>	2
□ type 1, <i>SLC12A1</i>	<i>SLC12A1</i>	2	Dystonia		
□ type 2, <i>KCNJ1</i>	<i>KCNJ1</i>	2	□ Torsion (early onset) (DYT1)	<i>TOR1A</i>	2
□ type 3, <i>CLCNKB</i>	<i>CLCNKB</i>	2	□ Dopa Responsive, autosomal dom. (DYT5)	<i>GCH1</i>	6
□ type 4, <i>BSND</i>	<i>BSND</i>	2	□ type 6, aut. dom. (DYT6)	<i>THAP1</i>	2
□ Basal laminar Drusen	<i>CFH</i>	2	□ type 18, aut. dom. (DYT18, Glut-1)	<i>SLC2A1</i>	2
□ Best Disease	<i>VMD2</i>	2	□ Dopa Resp., AR, (Tyrosine hydroxylase def.)	<i>TH</i>	6
□ Börjeson-Forssman-Lehman syndrome	<i>PHF6</i>	2			
Brachydactyly, type b			□ EEC syndrome	<i>TP63</i>	2
□ <i>ROR</i> □ <i>NOG</i>		2	□ Epidermolysis bullosa dystrophica <sup>5</sup>	<i>COL7A1</i>	2
□ Branchio-oculo-facio syndrome (BOFS)	<i>TFAP2A</i>	2	□ Erythralgia, primary or idiopathic	<i>SCN9A</i>	3
Branchiootorenal dysplasia (BOR syndrome)			Exudative Vitreoretinopathy		
□ <i>EYA1</i> □ <i>SIX5</i> □ <i>SIX1</i>		2	□ <i>FZD4</i>	<i>FZD4</i>	2
			□ <i>LRP5</i>	<i>LRP5</i>	2
Cardio-Facio-Cutaneous syndrome (CFC)			□ <i>TSPAN12</i>	<i>TSPAN12</i>	2
□ <i>BRAF</i>	<i>BRAF</i>	2	□ <i>NDP</i> (X-linked)	<i>NDP</i>	2
□ <i>KRAS</i>	<i>KRAS</i>	2			
□ <i>MAP2K1</i>	<i>MAP2K1</i>	2	□ Faciogenital dysplasia (Aarskog-Scott syndrome)	<i>FGD1</i>	2
□ <i>MAP2K2</i>	<i>MAP2K2</i>	2	□ Familial platelet disorder	<i>RUNX1</i>	2
□ Central Areolar Choroid Dystrophy	<i>PRPH2</i>	2	(with propensity to acute myelogenous leukemia)		
□ Cerebrotendinous Xanthomatosis	<i>CYP27A1</i>	2	□ Feingold syndr. (OculoDigitoEsophagoDuodenal)	<i>MYCN</i>	2
□ CHARGE syndrome	<i>CHD7</i>	2	□ Fragile X syndrome	<i>FMR1</i>	2
□ Choroideremia	<i>CHM</i>	2	□ Frank-Ter Haar syndrome	<i>SH3PXD2B2</i>	2
□ Chromosome 9q deletion syndrome	<i>EHMT1</i>	2	□ Fumarase deficiency	<i>FH</i>	2
□ Clouston Syndrome (Hidrotic ectoderm. dyspl.)	<i>GJB6</i>	2			
Complex I deficiency <sup>6</sup>			□ Gastric cancer (E-cadherin)	<i>CDH1</i>	2
□ <i>NDUFA1</i> □ <i>NDUFS2</i> □ <i>NDUFS6</i> □ <i>NDUFV1</i>		6	□ Gilbert syndrome	<i>UGT1A1</i>	2
□ <i>NDUFB11</i> □ <i>NDUFS3</i> □ <i>NDUFS7</i> □ <i>NDUFV2</i>		6	□ Gitelman syndrome	<i>SLC12A3</i>	2
□ <i>NDUFS1</i> □ <i>NDUFS4</i> □ <i>NDUFS8</i> □ <i>MIMITIN</i>		6	□ Glucocorticoid-remediable aldosteronism (GRA)	<i>HSD11B2</i>	2
Complex II deficiency <sup>6</sup>			□ Glucose transporter deficiency type 1	<i>SLC2A1</i>	2
□ <i>SDHA</i> □ <i>SDHB</i>		6	□ Glucosuria, renal (GLYS1)	<i>SLC5A2</i>	2
Complex III deficiency <sup>6</sup>					
□ <i>BCS1L</i> □ <i>UQCRCB</i>		6	Hemochromatosis, hereditary		
Complex IV deficiency <sup>6</sup>			□ type 1, <i>HFE</i> (standard mutations: C282Y/H63D/S65C) <sup>9</sup>		1
□ <i>SCO1</i> □ <i>COX10</i> □ <i>SURF1</i>		6	□ type 1, <i>HFE</i> (whole gene) <sup>9</sup>		6
□ <i>SCO2</i> □ <i>COX15</i> □ <i>LRPPRC</i>		6	□ type 2A, juvenile, <i>HJV</i> □ type 3, <i>TFR2</i>		6
			□ type 2B, juvenile, <i>HAMP</i> □ type 4, <i>SLC40A1</i>		6
			□ hypochromic microcytic anemia, <i>SLC11A2</i>		6

Disease	Gene	TAT (months)	Disease	Gene	TAT (months)
Hemolytic uremic syndrome			□ Nail-Patella syndrome	<i>LMX1B</i>	2
□ <i>CFH</i>	<i>CFH</i>	6	Nephrotic Syndrome,		
□ <i>CFI</i>	<i>CFI</i>	6	□ Finnish type, congenital	<i>NPHS1</i>	2
□ <i>MCP</i>	<i>MCP</i>	6	□ steroid resistant	<i>NPHS2</i>	2
□ Hypercalcemia, autosomal dominant (ADH)	<i>CASR</i>	2	□ type 3, NPHS3	<i>PLCE1</i>	2
□ Hypocalciuric hypercalcemia, familial, type I (FHH)	<i>CASR</i>	2	□ incl. diffuse mesangial sclerosis	<i>WT1</i>	2
Hyperhomocysteinemia			□ incl. focal segmental glomerulosclerosis	<i>ACTN4</i>	2
□ cystathione beta-synthase deficiency) <sup>2</sup>	<i>CBS</i>	3	□ foc. segm. glomerulosclerosis 2 (FSGS2)	<i>TRPC6</i>	2
□ methylenetetrahydrofolate reductase def. <sup>2</sup>	<i>MTHFR</i>	3	□ foc. segm. glomerulosclerosis 5 (FSGS5)	<i>INF2</i>	2
□ Hypomagnesemia	<i>FXRD</i>	2	□ Pierson syndrome, congenital	<i>LAMB2</i>	2
Ichthyosis, lamellar			□ Neuropathy, ataxia, and retinitis pigmentosa (NARP)	mtDNA	3
□ type 1 (LI 1)	<i>TGM1</i>	2	□ Nijmegen breakage syndrome	<i>NBS1</i>	2
□ type 2 (LI 2)	<i>ABCA12</i>	2	Noonan syndrome		
□ type 3 (LI 3)	<i>CYP4F22</i>	2	□ <i>PTPN11</i> + <i>SOS1</i> + <i>KRAS</i> + <i>RAF1</i> <sup>8</sup>		2
□ Ichthyosis, X-linked <sup>4</sup>	<i>STS</i>	2	□ <i>PTPN11</i>	<i>PTPN11</i>	2
Kallmann syndrome			□ <i>SOS1</i>	<i>SOS1</i>	2
□ type 1, <i>KAL1</i>	<i>KAL1</i>	2	□ <i>KRAS</i>	<i>KRAS</i>	1
□ type 2, <i>FGFR1</i>	<i>FGFR1</i>	2	□ <i>RAF1</i>	<i>RAF1</i>	2
□ type 3, <i>PROKR2</i>	<i>PROKR2</i>	2	□ <i>NRAS</i>	<i>NRAS</i>	2
□ type 4, <i>PROK2</i>	<i>PROK2</i>	2	□ <i>SHOC2</i>	<i>SHOC2</i>	2
□ Kearns-Sayre syndrome	mtDNA	3	□ Norrie disease	<i>NDP</i>	2
□ Kleeftstra syndrome	<i>EHMT1</i>	2	□ Oculo-dento-digital dysplasia (ODDD)	<i>GJA1</i>	2
□ Leber Congenital Amaurosis (LCA) <sup>10</sup>		3	□ Oculopharyngeal muscular dystrophy	<i>PABPN1</i>	2
□ Leber hereditary optic atrophy (LHON)	mtDNA	3	□ Ophthalmoplegia, chronic progressive (PEO)		
□ Leigh syndrome, mitochondrial	mtDNA	3	□ <i>POLG</i> □ <i>SLC25A4</i> (ANT1) □ <i>Twinkle</i>		2
□ Leigh syndrome, autosomal rec., (French-Canadian)	<i>LRPPRC</i>	2	□ Oritz-Kaveggia syndrome	<i>MED12</i>	2
□ Leiomyomatosis + renal cell cancer, autosomal dom.	<i>FH</i>	2	□ Pancreatitis, hereditary	<i>PRSS1</i>	3
LEOPARD syndrome			□ Parkinson's Disease (type 8)	<i>LRKK2</i>	2
□ <i>PTPN11</i>	<i>PTPN11</i>	2	□ Paroxysmal Extreme Pain disorder (PEPD)	<i>SCN9A</i>	2
□ <i>RAF1</i>	<i>RAF1</i>	2	□ Pendred syndrome	<i>SLC26A4</i>	2
Little syndrome			□ Pitt-Hopkins syndrome (PHS)	<i>TCF4</i>	2
□ <i>SCNN1B</i> + <i>SCNN1G</i> <sup>8</sup>		2	Polycystic liver disease (PCLD)		
□ <i>SCNN1B</i> (only for known mutations in family <sup>12</sup> )			□ <i>PRKCSH</i>	<i>PRKCSH</i>	6
□ <i>SCNN1B</i> (only for known mutations in family <sup>12</sup> )			□ <i>SEC63</i>	<i>SEC63</i>	6
□ Limb-mammary syndrome	<i>TP63</i>	2	Polyposis coli, juvenile		
□ Lujan-Fryns syndrome	<i>MED12</i>	2	□ <i>SMAD4</i>	<i>SMAD4</i>	2
□ MELAS syndrome	mtDNA	3	□ <i>BMPR1A</i>	<i>BMPR1A</i>	2
Mental retardation, X-linked			□ Prader-Willi syndrome	methylation	2
□ with epilepsy	<i>ARX</i>	2	□ Pseudohypoadosteronism, type 1	<i>NR3C2</i>	2
□ with $\alpha$ -thalassemia <sup>13</sup>	<i>ATRX</i>	2	□ PTEN Hamartoma Tumor Syndrome (PHTS)	<i>PTEN</i>	2
□ Börjeson-Forssman-Lehman syndrome	<i>PHF6</i>	2	Pyruvate dehydrogenase complex deficiency <sup>6</sup>		
□ with cerebellar atrophy	<i>OPHN1</i>	2	□ PDHA1 □ PDHB □ PDHX		6
□ Christianson-type	<i>SLC9A6</i>	2	□ DLAT □ DLD		6
□ with microcephaly	<i>SQBP1</i>	2	□ Renal tubular acidosis + deafness	<i>ATP6B1</i>	2
□ with growth hormone deficiency	<i>SOX3</i>	2	Rett syndrome		
□ with infantile spasms	<i>CDKL5</i>	2	□ <i>MECP2</i>	<i>MECP2</i>	2
□ syndromic (Cabezas syndrome, MRX15)	<i>CUL4B</i>	2	□ <i>FOXG1</i> (congenital variant)	<i>FOXG1</i>	2
□ syndromic, <i>ZDHC9</i> -related	<i>ZDHC9</i>	2	□ <i>CDKL5</i> (variant with infantile spasms)	<i>CDKL5</i>	2
□ MERFF syndrome	mtDNA	3	□ Rigid Spine Muscular Dystrophy I	<i>SEPN1</i>	2
□ Merosin-deficient congenital muscular dystrophy (MDC1A)	<i>LAMA2</i>	2	Robinow syndrome		
□ Methylmalonic acidemia, type C (CblC)	<i>MMACHC</i>	6	□ autosomal dominant	<i>WNT5A</i>	2
+ homocystinuria			□ autosomal recessive	<i>ROR2</i>	2
□ Microcephaly, primary, autosomal recessive, type 5	<i>ASPM</i>	2	□ Schinzel-Giedion syndrome	<i>SETBP1</i>	2
□ Mitochondriopathy (myopathy, encephalopathy, etc) <sup>2</sup>	mtDNA	3	□ SeSAME syndrome	<i>KCNJ10</i>	2
Mitochondrial DNA depletion syndrome			□ Small Patella syndrome	<i>TBX4</i>	2
□ DGUOK □ MPV17 □ SUCLA2 □ TK2		6	Spastic paraplegia		
□ Mitochondrial Neurogastrointestinal			□ type 4, autosomal dominant (SPG4)	<i>SPAST</i>	2
Encephalopathy (MNGIE)	<i>ECGF1</i>	6	□ type 3A, autosomal dominant (SPG3A )	<i>ATL1</i>	2
□ Mohr-Tranebjaerg syndrome (DFN1)	<i>DDP</i>	2	□ type 31, autosomal dominant (SPG31)	<i>REEP1</i>	2
□ Motor and sensory neuropathy type 2C (HMSN2C)	<i>TRPV4</i>	2	□ type 10, autosomal dominant (SPG10)	<i>KIF5A</i>	2
□ Multiple synostoses syndrome (SYNS1)	<i>NOG</i>	2	□ type 7, autosomal recessive	<i>SPG7</i>	2
□ Muscle Eye Brain disease (MEB) <sup>11</sup>	<i>POMGnT1</i>	2	□ type 11, autosomal recessive	<i>SPG11</i>	2
□ Myophosphorylase deficiency (McArdle disease)	<i>PYGM</i>	2	□ type 17, autosomaal dom. (Silver syndrome)	<i>BSCL2</i>	2
□ Myotonic dystrophy type 1	<i>DMPK</i>	2			
□ Myotonic dystrophy type 2 (prox. myotonic myopathy)	<i>CNBP(ZNF9)</i>	2			

Disease	Gene	TAT (months)
<input type="checkbox"/> Spinocerebellar ataxia, aut. rec., type 1 (SCAR1)	<i>SETX</i>	2
<input type="checkbox"/> Spinal muscular atrophy (SMA), scapuloperoneal	<i>TRPV4</i>	2
<input type="checkbox"/> Spondyloepiphyseal dysplasia tarda, X-linked (SEDL)	<i>TRAPPC2</i>	2
<input type="checkbox"/> Split hand/foot malformation (SHFM)	<i>TP63</i>	2
<input type="checkbox"/> Stargardt disease, type 1	<i>ABCA4</i>	2
<input type="checkbox"/> Symphalangism, proximal	<i>NOG</i>	2
<input type="checkbox"/> Testotoxicosis, familial <sup>2</sup>	<i>LHCGR</i>	2
<input type="checkbox"/> Thiamine-responsive megaloblastic anemia (deafness & diab. mellitus)	<i>SLC19A2</i>	2
<input type="checkbox"/> Thrombotic thrombocytopenic purpura (TTP)	<i>ADAMTS13</i>	6
<input type="checkbox"/> Treacher Collins-Franceschetti syndrome	<i>TCOF1</i>	2
<input type="checkbox"/> Tyrosine hydroxylase deficiency	<i>TH</i>	2
<input type="checkbox"/> Usher syndrome <sup>2,10</sup> , type ..		3
<input type="checkbox"/> Von Willebrand disease type 2	<i>VWF</i>	6
<input type="checkbox"/> Waardenburg syndrome		
<input type="checkbox"/> type 1 & 3, <i>PAX3</i> <input type="checkbox"/> type 2, <i>MITF</i>		2
<input type="checkbox"/> Walker-Warburg (-like) syndrome (consanguinity: yes / no) <sup>7</sup>		2
<input type="checkbox"/> <i>POMT1</i> <input type="checkbox"/> <i>FCMD</i> <input type="checkbox"/> <i>LAMA2</i> (MDC1A)		
<input type="checkbox"/> <i>POMT2</i> <input type="checkbox"/> <i>FKRP</i> <input type="checkbox"/> Homozygosity testing <sup>7</sup>		
<input type="checkbox"/> <i>POMGnT1</i> <input type="checkbox"/> <i>LARGE</i>		
<input type="checkbox"/> West syndrome	<i>ARX</i>	2
<input type="checkbox"/> Wolfram syndrome (DIDMOAD)	<i>WFS1</i>	2

#### FOOTNOTES

1. Please include serum for hepcidin testing, see [www.hepcidinanalysis.com](http://www.hepcidinanalysis.com)
2. Testing of frequent mutations: remainder of the gene after consultation
3. Analysis of repeat expansions: 1 month; sequencing FXN gene 2 months
4. Deletion testing only
5. Include blood of both parents
6. Only when biochemical analysis of a muscle biopsy has revealed reduced complex activity
7. Homozygosity testing of candidate loci. Only possible when parents are consanguineous !
8. Declaration per gene, all genes are analysed
9. Most mutations are found in these genes
10. Known mutations are analysed by APEX (Asper) micro array analysis, followed by confirmation of detected mutation(s)
11. See Walker-Warburg(-like) for other genes.
12. These genes will only be tested separately if the mutation has already been detected in other family members.
13. Analysis of whole gene only when EDTA blood is provided. In all other cases (including DNA), only hotspot analysis will be performed.

## **Additional information for requesting molecular diagnostic testing at the Division of DNA-Diagnostics of the Clinical Genetics Centre Nijmegen**

### **1 Requests**

- 1.1 In order to prevent delays and errors, requests for molecular diagnostics should be clear and comprehensive. By filling out the request form completely, all necessary details are provided.
- 1.2 By accepting a request for molecular diagnostics, the Division of DNA-Diagnostics commits itself to conduct the requested research with care and expertise, in accordance with the quality guidelines as specified for our laboratory.
- 1.3 Requests may be rejected in case insufficient information is provided to guarantee a result which is in accordance with our quality guidelines. The requesting party is contacted immediately when this applies.
- 1.4 DNA-diagnostics must be able to contact the referring clinician in case of queries regarding the patient or the requested tests.
- 1.5 The invoice will be sent to the referring physician. In case a different billing address should be used, this needs to be indicated clearly on the request form.

### **2 Samples**

- 2.1 The requesting party should make sure that the sample tubes are properly labelled with name, gender and date of birth of the patient, and are accompanied by a completed request form.
- 2.2 A volume of 2 x 10 ml EDTA blood is required per patient (for neonates this is at least 3 ml), which should be shipped by regular mail at room temperature, in plastic (no glass) tubes. Other materials/tissues only after consultation.
- 2.3 When requirements 2.1 and 2.2 are not met, DNA-Diagnostics is not obliged to accept the samples.
- 2.4 When no other arrangements have been made at the time of the request, DNA-Diagnostics will store or dispose of the samples and/or remaining material according to the rules and regulations of the Division. Additional information under 6.

### **3 Testing**

- 3.1 The Division of DNA-Diagnostics determines which procedures, methods and equipment are employed to conduct the requested analysis.
- 3.2 All procedures are carried out according to all applicable standards, rules and regulations. Details can be sent on request.
- 3.3 In case a particular request involves procedures which are outside the scope of expertise and experience of the Department, DNA-Diagnostics will contact the requesting party about outsourcing these activities.
- 3.4 DNA-Diagnostics is not responsible for all activities and storage which occur prior to the acceptance of a sample.

### **4 Results**

- 4.1 Results (test results, advise, information etc.) are provided in writing.
- 4.2 Results are usually provided within the following time frames:
  - Prenatal testing : 2-3 weeks
  - Presymptomatic testing /carrier testing/ confirmation of diagnosis(familial mutation known): 4 weeks
  - Elaborate mutation scanning (familial mutation not known): 2 months

### **5 Patient confidentiality**

- 5.1 The privacy of all patients is guaranteed as stated in the Radboud University Medical Centre rules and regulations on patient confidentiality.

### **6 Use of patient material**

- 6.1 Please note that DNA will be stored from this patient's sample at the Division of DNA-Diagnostics. The sample will be kept indefinitely unless a written request for its disposal is received from the patient or his lawful representatives.
- 6.2 DNA-Diagnostics uses coded patient material for research purposes. Only testing in line with the original request will be carried out. The referring physician will be informed in case this leads to results that are relevant for the patient.
- 6.3 For the development and improvement of new and existing techniques, DNA-Diagnostics uses coded patient material, for control and validation among others. In case the patient objects to the use of the material for this purpose the patient or his lawful representative can contact Dr. H. Scheffer (Division Head).