



Radboud University Nijmegen Medical Center

848 DNA-diagnostics
PO Box 9101
6500 HB Nijmegen
The Netherlands
street address:
Geert Grooteplein 10
6525 GA Nijmegen
The Netherlands

Head: Dr. H. Scheffer
Mrs. Dr. E.H. Hoefsloot
Mrs. Dr. M.J.L. Ligtenberg
Dr. E.J. Kamsteeg
Dr. M.R. Nelen
Mrs. Dr. H.G. Intema

Fax +31(0)24-3616658
Tel +31(0)24-3613799
E-mail: dna@umcn.nl
http://www.dnadiagnostieknijmegen.nl/en/dd_index_en.php

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 (~20 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 (~20 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 relevant.
- concerning hereditary diseases in general (anonymously)

- The patient or his/her legal representative does object to further use of the material for scientific research.

► **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:

<i>Paraaf Sec.</i>	
--------------------	--

► **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:

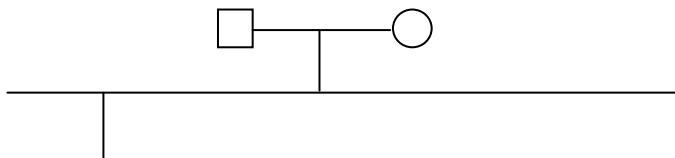
- Name previously tested family member:
- Date of birth previously tested family member:
- Relation to current patient:
- Date of birth parents of the patient, father: mother:
- Number of siblings of patient (see pedigree):
- Our Reference previous request (if known):
- Gene involved and mutation (if 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 I deficiency ⁶		
Achromatopsia			□ <i>NDUFA1</i> □ <i>NDUFS2</i> □ <i>NDUFS6</i> □ <i>NDUFV1</i>		2
□ type 2, <i>CNGA3</i>	<i>CNGA3</i>	2	□ <i>NDUFB11</i> □ <i>NDUFS3</i> □ <i>NDUFS7</i> □ <i>NDUFV2</i>		2
□ type 3, <i>CNGB3</i>	<i>CNGB3</i>	2	□ <i>NDUFS1</i> □ <i>NDUFS4</i> □ <i>NDUFS8</i> □ <i>MIMITIN</i>		2
Adrenal hyperplasia, congenital			Complex II deficiency ⁶		
□ 21-hydroxylase deficiency	<i>CYP21A2</i>	2	□ <i>SDHA</i>	<i>SDHA</i>	2
□ 11-b hydroxylase deficiency	<i>CYP11B1</i>	2	□ <i>SDHB</i>	<i>SDHB</i>	2
□ 3-b hydroxysteroid dehydrog. def. (type II)	<i>HSD3B2</i>	2	Complex III deficiency ⁶		
□ p450c17 and p450c21 deficiency	<i>POR</i>	2	□ <i>BCS1L</i>	<i>BCS1L</i>	2
□ Adrenoleukodystrophy	<i>ABCD1</i>	2	□ <i>UQCRB</i>	<i>UQCRB</i>	2
□ ADULT syndrome	<i>TP63</i>	2	Complex IV deficiency ⁶		
□ AEC syndrome (Hay-Wells)	<i>TP63</i>	2	□ <i>SCO1</i> □ <i>COX10</i> □ <i>SURF1</i>		2
□ Alpers syndrome	<i>POLG</i>	6	□ <i>SCO2</i> □ <i>COX15</i> □ <i>LRPPRC</i>		2
□ Amyotrophic lateral sclerosis 4, juvenile (ALS4)	<i>SETX</i>	2	□ Complex V deficiency, autosomal recessive ⁶	<i>ATP12</i>	2
Anemia			Complex (oxphos) deficiency, combined		
□ Hypochromic microcytic	<i>SLC11A2</i>	6	□ <i>GFM1</i>	<i>GFM1</i>	2
□ Refractory iron deficiency ¹	<i>TMPRSS6</i>	6	□ <i>MRPS16</i>	<i>MRPS16</i>	2
□ Sideroblastic anemia, X-linked	<i>ALAS2</i>	6	□ <i>PUS1</i>	<i>PUS1</i>	2
□ Sideroblastic anemia with ataxia, X-linked	<i>ABCB7</i>	6	□ <i>Twinkle</i>	<i>Twinkle</i>	2
□ Sideroblastic anemia, autosomal recessive	<i>SLC25A38</i>	6	□ Congenital anomalies of the kidney and urinary tract (CAKUT)	<i>HNF1B</i>	2
Angelman syndrome			□ Congenital indifference to pain	<i>SCN9A</i>	3
□ Methylation		2	Costello syndrome		
□ <i>UBE3A</i>		2	□ <i>HRAS</i>	<i>HRAS</i>	1
□ Arts Syndrome	<i>PRPS1</i>	2	□ <i>KRAS</i>	<i>KRAS</i>	1
□ Aromatic L-aminoacid decarboxylase def. (AADC)	<i>DDC</i>	2	□ <i>PTEN</i>	<i>PTEN</i>	2
Ataxia			□ Cowden disease		
□ Friedreich ataxia (FRDA)	<i>FXN</i>	2	Cranioectodermal dysplasia (Sensenbrenner syndrome)		
□ Ataxia + isolated vitamin E def. (AVED)	<i>TTPA</i>	2	□ type 1, <i>IFT122</i> □ type 2, <i>WDR35</i>		2
□ Ataxia-oculomotor apraxia type 1 (AOA1)	<i>APTX</i>	2	Cutis laxa type 1, autosomal recessive		
□ Ataxia-oculomotor apraxia type 2 (AOA2)	<i>SETX</i>	2	□ <i>FBLN5</i> □ <i>EFEMP2</i>		2
□ Cerebellar ataxia, aut. recessive	<i>ANO10</i>	2	Cutis laxa type 2B, autosomal recessive		
□ Spastic ataxia, Charlevoix-Saguenay (ARSACS)	<i>SACS</i>	2	□ <i>PYCR1</i>	<i>PYCR1</i>	2
□ Spinocerebellar ataxia with axonal neuropathy	<i>TDP1</i>	2	Cysteinuria		
□ Bannayan-Riley-Ruvalcaba syndrome	<i>PTEN</i>	2	□ <i>SLC3A1</i> □ <i>SLC7A9</i>		2
□ Bardet-Biedl syndrome ¹⁰ , or specific gene :.....		3	□ Cystinosis, nephropathic	<i>CTNS</i>	2
Bartter syndrome			Deafness, autosomal dominant		
□ type 1, <i>SLC12A1</i>	<i>SLC12A1</i>	2	□ type 2 (DFNA2) ²	<i>KCNQ4</i>	2
□ type 2, <i>KCNJ1</i>	<i>KCNJ1</i>	2	□ type 5 (DFNA5) ²	<i>DFNA5</i>	2
□ type 3, <i>CLCNKB</i>	<i>CLCNKB</i>	2	□ type 9 (DFNA9) ²	<i>COCH</i>	2
□ type 4, <i>BSND</i>	<i>BSND</i>	2	□ type 6/14 (DFNA6/14)	<i>WFS1</i>	2
□ Basal laminar Drusen	<i>CFH</i>	2	□ type 8/12 (DFNA8/12)	<i>TECTA</i>	2
□ Best Disease	<i>VMD2</i>	2	□ type 10 (DFNA10)	<i>EYA4</i>	2
□ Bloom's syndrome	<i>BLM</i>	2	□ type 13 (DFNA13)	<i>COL11A2</i>	2
□ Börjeson-Forssman-Lehman syndrome	<i>PHF6</i>	2	□ type 15 (DFNA15)	<i>POU4F3</i>	2
Brachydactyly, type b			□ type 20/26 (DFNA20/26)	<i>ACTG1</i>	2
□ <i>ROR2</i>	<i>ROR2</i>	2	Deafness, autosomal recessive		
□ <i>NOG</i>	<i>NOG</i>	2	□ type 1 (DFNB1) (connexin 26)	<i>GJB2/6</i>	2
□ Branchio-oculo-facio syndrome (BOFS)	<i>TFAP2A</i>	2	□ type 8 (DFNB8)	<i>TMPRSS3</i>	2
Branchiootorenal dysplasia (BOR syndrome)			□ type 9 (DFNB9)	<i>OTOF</i>	2
□ <i>EYA1</i>	<i>EYA1</i>	2	□ type 21 (DFNB21)	<i>TECTA</i>	2
□ <i>SIX1</i>	<i>SIX1</i>	2	□ type 79 (DFNB79)	<i>TPRN</i>	2
□ <i>SIX5</i>	<i>SIX5</i>	2	Deafness, X-linked		
Cardio-Facio-Cutaneous syndrome (CFC)			□ type 1 (DFN1)	<i>TIMM8A</i>	2
□ <i>BRAF</i>	<i>BRAF</i>	2	□ type 3 (DFN3)	<i>POU3F4</i>	2
□ <i>KRAS</i>	<i>KRAS</i>	2	□ type 4 (DFN4)	<i>SMPX</i>	2
□ <i>MAP2K1</i>	<i>MAP2K1</i>	2	□ Dent disease (nephrolithiasis type 1)	<i>CLCN5</i>	2
□ <i>MAP2K2</i>	<i>MAP2K2</i>	2	Diabetes insipidus,		
□ Central Areolar Choroid Dystrophy	<i>PRPH2</i>	2	□ central	<i>AVP</i>	2
Centronuclear myopathy, autosomal dominant			□ nephrogenic, X-linked	<i>AVPR2</i>	2
□ <i>DNM2</i>	<i>DNM2</i>	2	□ nephrogenic, autosomal dominant/recessive	<i>AQP2</i>	2
□ <i>MYF6</i>	<i>MYF6</i>	2	□ Doyme honeycomb retinal dystrophy (DHRD) ²	<i>EFEMP1</i>	2
□ Cerebrotendinous Xanthomatosis	<i>CYP27A1</i>	2	Dystonia		
□ CHARGE syndrome	<i>CHD7</i>	2	□ Torsion (early onset) (DYT1)	<i>TOR1A</i>	2
□ Chloride diarrhea	<i>SLC26A3</i>	2	□ Dopa Responsive, autosomal dom. (DYT5)	<i>GCH1</i>	6
□ Choroideremia	<i>CHM</i>	2	□ type 6, aut. dom. (DYT6)	<i>THAP1</i>	2
□ Chromosome 9q deletion syndrome	<i>EHMT1</i>	2	□ type 18, aut. dom. (DYT18, Glut-1)	<i>SLC2A1</i>	2
□ Clouston Syndrome (Hidrotic ectoderm. dyspl.)	<i>GJB6</i>	2	□ Dopa resp., AR, (tyrosine hydroxylase def.)	<i>TH</i>	6
□ Coffin-Lowry syndrome	<i>RPS6KA3</i>	2	□ Dopa resp., AR (sepiapterin reductase def.)	<i>SPR</i>	2
			□ Paroxysmal nonkinesigenic dyskinesia type 1, AD	<i>PNKD</i>	2

Disease	Gene	TAT (months)	Disease	Gene	TAT (months)
Early infantile epileptic encephalopathy (EIEE)					
□ type 1, <i>ARX</i>	<i>ARX</i>	2	□ Kearns-Sayre syndrome	mtDNA	3
□ type 2, <i>CDKL5</i>	<i>CDKL5</i>	2	□ Kleefstra syndrome	<i>EHMT1</i>	2
□ type 3, <i>SLC25A22</i>	<i>SLC25A22</i>	2	□ Leber Congenital Amaurosis (LCA) ¹⁰		3
□ type 4, <i>STXBP1</i>	<i>STXBP1</i>	2	or test for specific gene:		
□ type 6, <i>SCN1A</i>	<i>SCN1A</i>	2	□ <i>AIP1</i>	<i>AIP1</i>	2
□ <i>MEF2C</i>	<i>MEF2C</i>	2	□ <i>CRB1</i>	<i>CRB1</i>	2
□ EEC syndrome	<i>TP63</i>	2	□ <i>GUCY2D</i>	<i>GUCY2D</i>	2
□ Epidermolysis bullosa dystrophica	<i>COL7A1</i>	2	□ <i>RPE65</i>	<i>RPE65</i>	2
□ Erythralgia, primary or idiopathic	<i>SCN9A</i>	3	□ Leber hereditary optic atrophy (LHON)	mtDNA	3
Exudative Vitreoretinopathy			□ Leigh syndrome, mitochondrial	mtDNA	3
□ <i>FZD4</i>	<i>FZD4</i>	2	□ Leigh syndrome, autosomal rec., (French-Canadian)	<i>LRPPRC</i>	2
□ <i>LRP5</i>	<i>LRP5</i>	2	□ Leiomyomatosis + renal cell cancer, autosomal dom.	<i>FH</i>	2
□ <i>TSPAN12</i>	<i>TSPAN12</i>	2	LEOPARD syndrome		
□ <i>ZNF408</i>	<i>ZNF408</i>	2	□ <i>PTPN11</i>	<i>PTPN11</i>	2
□ <i>NDP</i> (X-linked)	<i>NDP</i>	2	□ <i>RAF1</i>	<i>RAF1</i>	2
□ Faciogenital dysplasia (Aarskog-Scott syndrome)	<i>FGD1</i>	2	Liddle syndrome		
□ Familial platelet disorder (with propensity to acute myelogenous leukemia)	<i>RUNX1</i>	2	□ <i>SCNN1B</i> + <i>SCNN1G</i> ⁸		2
□ Feingold syndr. (OculoDigitoEsophagoDuodenal)	<i>MYCN</i>	2	□ <i>SCNN1B</i> (only for known mutations in family ¹²)		
□ Fragile X syndrome	<i>FMR1</i>	2	□ <i>SCNN1B</i> (only for known mutations in family ¹²)		
□ Frank-Ter Haar syndrome	<i>SH3PXD2B</i>	2	□ Limb-mammary syndrome	<i>TP63</i>	2
□ Fumarase deficiency	<i>FH</i>	2	Lujan-Fryns syndrome		
□ Gastric cancer (E-cadherin)	<i>CDH1</i>	2	□ <i>MED12</i>	<i>MED12</i>	2
□ Geroderma osteodysplasticum	<i>GORAB</i>	2	□ <i>UPF3B</i>	<i>UPF3B</i>	2
□ Gilbert syndrome	<i>UGT1A1</i>	2	□ <i>ZDHHC9</i>	<i>ZDHHC9</i>	2
□ Gitelman syndrome	<i>SLC12A3</i>	2	□ Mabry syndrome	<i>PIGV</i>	2
□ Glucocorticoid-remediable aldosteronism (GRA)	<i>HSD11B2</i>	2	□ MACS syndrome (macrocephaly, alopecia, cutis laxa, scoliosis)	<i>RIN2</i>	2
□ Glucose transporter type 1 deficiency syndrome	<i>SLC2A1</i>	2	□ Medullary cystic kidney disease	<i>UMOD</i>	2
□ Glucosuria, renal (GLYS1)	<i>SLC5A2</i>	2	Meier-Gorlin syndrome		
□ Glycogen storage disease type IV	<i>GBE1</i>	2	□ <i>CDC6</i>	<i>CDC6</i>	2
Hemochromatosis, hereditary			□ <i>CDT1</i>	<i>CDT1</i>	2
□ type 1, (standard mutations: C282Y/H63D/S65C)	<i>HFE</i>	1	□ <i>ORC1L</i>	<i>ORC1L</i>	2
□ type 1, <i>HFE</i> (whole gene)	<i>HFE</i>	6	□ <i>ORC4L</i>	<i>ORC4L</i>	2
□ type 2A, <i>HJV</i> juvenile	<i>HFE2</i>	6	□ <i>ORC6L</i>	<i>ORC6L</i>	2
□ type 2B, juvenile	<i>HAMP</i>	6	□ MELAS syndrome	mtDNA	3
□ type 3	<i>TFR2</i>	6	Mental retardation, X-linked		
□ type 4	<i>SLC40A1</i>	6	□ with epilepsy	<i>ARX</i>	2
□ type 5, <i>FTH</i> (IRE)	<i>FTH</i>	6	□ with α -thalassemia ¹³	<i>ATRX</i>	2
□ hypochromic microcytic anemia	<i>SLC11A2</i>	6	□ Börjeson-Forssman-Lehman syndrome	<i>PHF6</i>	2
□ hyperferritinemia cataract syndrome	<i>FTL</i> (IRE)	6	□ with cerebellar atrophy	<i>OPHN1</i>	2
Hemolytic uremic syndrome			□ Christianson-type	<i>SLC9A6</i>	2
□ <i>CFH</i>	<i>CFH</i>	2	□ with microcephaly	<i>PQBP1</i>	2
□ <i>CFI</i>	<i>CFI</i>	2	□ with growth hormone deficiency	<i>SOX3</i>	2
□ <i>MCP</i>	<i>MCP</i>	2	□ with infantile spasms	<i>CDKL5</i>	2
□ Hypercalcemia, autosomal dominant (ADH)	<i>CASR</i>	2	□ syndromic (Cabezas syndrome, MRX15)	<i>CUL4B</i>	2
□ Hyperphosphatemia with mental retardation	<i>PIGV</i>	2	□ syndromic (Siderius)	<i>PHF8</i>	2
□ Hypocalciuric hypercalcemia, familial, type I (FHH)	<i>CASR</i>	2	□ syndromic, <i>ZDHHC9</i> -related	<i>ZDHHC9</i>	2
Hyperhomocysteinemia			□ syndromic (MRXS14)	<i>UPF3B</i>	2
□ cystathione beta-synthase deficiency) ²	<i>CBS</i>	3	□ MERFF syndrome	mtDNA	3
□ methylenetetrahydrofolate reductase def. ²	<i>MTHFR</i>	3	□ Merosin-deficient congenital muscular dystrophy (MDC1A)	<i>LAMA2</i>	2
Hypomagnesemia			□ Metachondromatosis	<i>PTPN11</i>	2
□ type 1 (HOMG1)	<i>TRPM6</i>	2	□ Methylmalonic acidemia, type C (CblC) + homocystinuria	<i>MMACHC</i>	6
□ type 2 (HOMG2)	<i>FXYD2</i>	2	□ Microcephaly, primary, autosomal recessive, type 5	<i>ASPM</i>	2
□ type 3 (HOMG3)	<i>CLDN16</i>	2	□ Mitochondriopathy (myopathy, encephalopathy, etc) ²	mtDNA	3
□ type 4 (HOMG4)	<i>EGF</i>	2	Mitochondrial DNA depletion syndrome		
□ type 5 (HOMG5)	<i>CLDN19</i>	2	□ <i>DGUOK</i>	<i>DGUOK</i>	2
□ <i>HNF1B</i>	<i>HNF1B</i>	2	□ <i>MPV17</i>	<i>MPV17</i>	2
□ <i>KCNA1</i>	<i>KCNA1</i>	2	□ <i>SUCLA2</i>	<i>SUCLA2</i>	2
Ichthyosiform erythroderma, nonbullous			□ <i>TK2</i>	<i>TK2</i>	2
□ <i>ALOX12B</i>		2	□ Mitochondrial Neurogastrointestinal Encephalopathy (MNGIE)	<i>ECGF1</i>	2
□ <i>ALOXE3</i>		2	□ Mohr-Tranebjaerg syndrome (DFN1)	<i>DDP</i>	2
Ichthyosis, lamellar			□ Motor and sensory neuropathy type 2C (HMSN2C)	<i>TRPV4</i>	2
□ type 1 (LI 1)	<i>TGM1</i>	2	□ Mowat-Wilson syndrome	<i>ZEB2</i>	2
□ type 2 (LI 2)	<i>ABCA12</i>	2	□ Multiple endocrine neoplasia type 2 (MEN2)	<i>RET</i>	2
□ type 3 (LI 3)	<i>CYP4F22</i>	2	Multiple synostoses syndrome (SYNS1)		
□ Ichthyosis, X-linked ⁴	<i>STS</i>	2	□ <i>GDF5</i>	<i>GDF5</i>	2
□ Joubert syndrome, type 10	<i>OFD1</i>	2	□ <i>NOG</i>	<i>NOG</i>	2
Kallmann syndrome			□ Muscle Eye Brain disease (MEB) ¹¹	<i>POMGnT1</i>	2
□ type 1, <i>KAL1</i>	<i>KAL1</i>	2	Myofibrillar myopathy		
□ type 2, <i>FGFR1</i>	<i>FGFR1</i>	2	□ <i>BAG3</i>	<i>BAG3</i>	2
□ type 3, <i>PROKR2</i>	<i>PROKR2</i>	2	□ <i>FLNC</i>	<i>FLNC</i>	2
□ type 4, <i>PROK2</i>	<i>PROK2</i>	2	□ Myophosphorylase deficiency (McArdle disease)	<i>PYGM</i>	2
□ type 5, <i>CHD7</i>	<i>CHD7</i>	2			
□ type 6, <i>FGF8</i>	<i>FGF8</i>	2			

Disease	Gene	TAT (months)	Disease	Gene	TAT (months)
<input type="checkbox"/> Myotonic dystrophy type 1	<i>DMPK</i>	2	<input type="checkbox"/> Renal tubular acidosis, distal	<i>SLC4A1</i>	2
<input type="checkbox"/> Myotonic dystrophy type 2 (prox. myotonic myopathy)	<i>CNBP(ZNF9)</i>	2	Renal tubular acidosis, distal, + deafness		
<input type="checkbox"/> Nail-Patella syndrome	<i>LMX1B</i>	2	<input type="checkbox"/> <i>ATP6V1B1</i>	<i>ATP6V1B1</i>	2
Nemaline myopathy			<input type="checkbox"/> <i>ATP6V0A4</i>	<i>ATP6V0A4</i>	2
<input type="checkbox"/> type 3	<i>ACTA1</i>	2	<input type="checkbox"/> Renal tubular acidosis with osteopetrosis	<i>CA2</i>	2
<input type="checkbox"/> type 7	<i>CFL</i>	2	<input type="checkbox"/> Retinal cone-dystrophy, type 3B	<i>KCNV2</i>	2
Nephrotic Syndrome,			Retinitis Pigmentosa		
<input type="checkbox"/> Finnish type, congenital	<i>NPHS1</i>	2	<input type="checkbox"/> autosomal recessive ¹⁰ ,		3
<input type="checkbox"/> steroid resistant	<i>NPHS2</i>	2	or test specific gene:.....		2
<input type="checkbox"/> type 3, NPHS3	<i>PLCE1</i>	2	<input type="checkbox"/> autosomal dominant ¹⁰ ,		3
<input type="checkbox"/> incl. diffuse mesangial sclerosis	<i>WT1</i>	2	or test specific gene:.....		2
<input type="checkbox"/> incl. focal segmental glomerulosclerosis	<i>ACTN4</i>	2	Rett syndrome		
<input type="checkbox"/> foc. segm. glomerulosclerosis 2 (FSGS2)	<i>TRPC6</i>	2	<input type="checkbox"/> <i>MECP2</i>	<i>MECP2</i>	2
<input type="checkbox"/> foc. segm. glomerulosclerosis 3 (FSGS3)	<i>CD2AP</i>	2	<input type="checkbox"/> <i>FOXG1</i> (congenital variant)	<i>FOXG1</i>	2
<input type="checkbox"/> foc. segm. glomerulosclerosis 5 (FSGS5)	<i>INF2</i>	2	<input type="checkbox"/> <i>CDKL5</i> (variant with infantile spasms)	<i>CDKL5</i>	2
<input type="checkbox"/> Pierson syndrome, congenital	<i>LAMB2</i>	2	<input type="checkbox"/> <i>MEF2C</i>	<i>MEF2C</i>	2
<input type="checkbox"/> Neuropathy, ataxia, and retinitis pigmentosa (NARP)	mtDNA	3	<input type="checkbox"/> <i>SCN1A</i>	<i>SCN1A</i>	2
<input type="checkbox"/> Nijmegen breakage syndrome	<i>NBN</i>	2	<input type="checkbox"/> <i>SLC25A22</i>	<i>SLC25A22</i>	2
Noonan syndrome			<input type="checkbox"/> <i>STXBP1</i>	<i>STXBP1</i>	2
<input type="checkbox"/> <i>PTPN11</i> + <i>SOS1</i> + <i>KRAS</i> + <i>RAF1</i> ⁸		2	<input type="checkbox"/> Rigid Spine Muscular Dystrophy I	<i>SEPN1</i>	2
<input type="checkbox"/> <i>PTPN11</i>	<i>PTPN11</i>	2	Robinow syndrome		
<input type="checkbox"/> <i>SOS1</i>	<i>SOS1</i>	2	<input type="checkbox"/> autosomal dominant	<i>WNT5A</i>	2
<input type="checkbox"/> <i>KRAS</i>	<i>KRAS</i>	1	<input type="checkbox"/> autosomal recessive	<i>ROR2</i>	2
<input type="checkbox"/> <i>RAF1</i>	<i>RAF1</i>	2	<input type="checkbox"/> Schinzel-Giedion syndrome	<i>SETBP1</i>	2
<input type="checkbox"/> <i>NRAS</i>	<i>NRAS</i>	2	<input type="checkbox"/> SeSAME syndrome	<i>KCNJ10</i>	2
<input type="checkbox"/> <i>SHOC2</i>	<i>SHOC2</i>	2	<input type="checkbox"/> Sideroblastic anemia and spinocerebellar ataxia (ASAT)	<i>ABCB7</i>	2
<input type="checkbox"/> <i>CBL</i>	<i>CBL</i>	2	<input type="checkbox"/> Simpson-Golabi-Behmel syndrome, type 2	<i>OFD1</i>	2
<input type="checkbox"/> Norrie disease	<i>NDP</i>	2	<input type="checkbox"/> Small Patella syndrome	<i>TBX4</i>	2
<input type="checkbox"/> Occult macular dystrophy (OCMD)	<i>RP1L1</i>	2	Spastic paraplegia, autosomal dominant		
<input type="checkbox"/> Oculo-dento-digital dysplasia (ODDD)	<i>GJA1</i>	2	<input type="checkbox"/> type 3A (SPG3A) ⁹	<i>ATL1</i>	2
<input type="checkbox"/> Oculopharyngeal muscular dystrophy	<i>PABPN1</i>	2	<input type="checkbox"/> type 4 (SPG4) ⁹	<i>SPAST</i>	2
Ophthalmoplegia, chronic progressive (PEO)			<input type="checkbox"/> type 6 (SPG6)	<i>NIPA1</i>	2
<input type="checkbox"/> <i>POLG</i>	<i>POLG</i>	2	<input type="checkbox"/> type 8 (SPG8)	<i>KIAA0196</i>	2
<input type="checkbox"/> <i>SLC25A4</i> (ANT1)	<i>SLC25A4</i>	2	<input type="checkbox"/> type 13 (SPG13)	<i>HSPD1</i>	2
<input type="checkbox"/> <i>Twinkle</i>	<i>Twinkle</i>	2	<input type="checkbox"/> type 10 (SPG10)	<i>KIF5A</i>	2
<input type="checkbox"/> Opitz G/BBB syndrome	<i>MID1</i>	2	<input type="checkbox"/> type 17 (Silver syndrome)	<i>BSCL2</i>	2
<input type="checkbox"/> Opitz-Kaveggia syndrome	<i>MED12</i>	2	<input type="checkbox"/> type 31 (SPG31) ⁹	<i>REEP1</i>	2
<input type="checkbox"/> Optic atrophy, deafness, ophthalmoplegia and myopathy	<i>OPA1</i>	2	<input type="checkbox"/> type 42 (SPG42)	<i>SLC33A1</i>	2
<input type="checkbox"/> Orofaciodigital syndrome, type 1	<i>OFD1</i>	2	Spastic paraplegia, autosomal recessive		
<input type="checkbox"/> Pancreatitis, hereditary	<i>PRSS1</i>	3	<input type="checkbox"/> type 5A (SPG5A)	<i>CYP7B1</i>	2
Paraganglioma / Pheochromocytoma, hereditary		2	<input type="checkbox"/> type 7, autosomal recessive	<i>SPG7</i>	2
<input type="checkbox"/> <i>SDHA</i> <input type="checkbox"/> <i>SDHD</i> <input type="checkbox"/> <i>VHL</i>			<input type="checkbox"/> type 11, autosomal recessive	<i>SPG11</i>	2
<input type="checkbox"/> <i>SDHB</i> <input type="checkbox"/> <i>SDHAF2</i> <input type="checkbox"/> <i>RET</i>			<input type="checkbox"/> type 15 (SPG15)	<i>ZFYVE26</i>	2
<input type="checkbox"/> <i>SDHC</i> <input type="checkbox"/> <i>TMEM127</i> <input type="checkbox"/> <i>MAX</i>			<input type="checkbox"/> type 20 (SPG20)	<i>SPG20</i>	2
<input type="checkbox"/> Parkinson's Disease (type 8)	<i>LRRK2</i>	2	<input type="checkbox"/> type 21 (SPG21)	<i>SPG21</i>	2
<input type="checkbox"/> Paroxysmal Extreme Pain disorder (PEPD)	<i>SCN9A</i>	2	<input type="checkbox"/> type 35 (SPG35)	<i>FA2H</i>	2
<input type="checkbox"/> Pendred syndrome	<i>SLC26A4</i>	2	<input type="checkbox"/> type 48 (SPG48)	<i>KIAA0415</i>	2
Pheochromocytoma, hereditary: see paraganglioma			<input type="checkbox"/> Spinocerebellar ataxia, aut. rec., type 1 (SCAR1)	<i>SETX</i>	2
<input type="checkbox"/> Pitt-Hopkins syndrome (PHTS)	<i>TCF4</i>	2	<input type="checkbox"/> Spinal muscular atrophy (SMA), scapulo-peroneal	<i>TRPV4</i>	2
Polycystic liver disease (PCLD)			<input type="checkbox"/> Spondyloepiphyseal dysplasia tarda, X-linked (SEDL)	<i>TRAPPC2</i>	2
<input type="checkbox"/> <i>PRKCSH</i>	<i>PRKCSH</i>	6	<input type="checkbox"/> Split hand/foot malformation (SHFM)	<i>TP63</i>	2
<input type="checkbox"/> <i>SEC63</i>	<i>SEC63</i>	6	<input type="checkbox"/> Stargardt disease, type 1	<i>ABCA4</i>	2
<input type="checkbox"/> Polyglucosan body disease, adult form	<i>GBE1</i>	2	Symphalangism, proximal		
Polyposis coli, juvenile			<input type="checkbox"/> <i>GDF5</i> <input type="checkbox"/> <i>NOG</i>		2
<input type="checkbox"/> <i>SMAD4</i>	<i>SMAD4</i>	2	<input type="checkbox"/> Testotoxicosis, familial ²	<i>LHCGR</i>	2
<input type="checkbox"/> <i>BMPR1A</i>	<i>BMPR1A</i>	2	<input type="checkbox"/> Thiamine-responsive megaloblastic anemia (deafness & diab. mellitus)	<i>SLC19A2</i>	2
Porphyria			<input type="checkbox"/> Thrombotic thrombocytopenic purpura (TTP)	<i>ADAMTS13</i>	6
<input type="checkbox"/> Erythropoietic protoporphyria, X-linked	<i>ALAS2</i>	6	Treacher Collins-Franceschetti syndrome		
<input type="checkbox"/> Prader-Willi syndrome	methylation	2	<input type="checkbox"/> <i>TCOF1</i>	<i>TCOF1</i>	2
Pseudohypoadosteronism type 1			<input type="checkbox"/> <i>POLR1D</i>	<i>POLR1D</i>	2
<input type="checkbox"/> autosomal dominant (<i>NR3C2</i>)	<i>NR3C2</i>	2	<input type="checkbox"/> <i>POLR1C</i>	<i>POLR1C</i>	2
<input type="checkbox"/> autosomal recessive (<i>SCNN1A</i>)	<i>SCNN1A</i>	2	<input type="checkbox"/> Tyrosine hydroxylase deficiency	<i>TH</i>	2
<input type="checkbox"/> autosomal recessive (<i>SCNN1B</i>)	<i>SCNN1B</i>	2	<input type="checkbox"/> Usher syndrome ^{2,10} , type ..		3
<input type="checkbox"/> autosomal recessive (<i>SCNN1G</i>)	<i>SCNN1G</i>	2	<input type="checkbox"/> Von Hippel-Lindau syndrome (VHL)	<i>VHL</i>	2
<input type="checkbox"/> PTEN Hamartoma Tumor Syndrome (PHTS)	<i>PTEN</i>	2	<input type="checkbox"/> Von Willebrand disease type:.....	<i>VWF</i>	3
Pyruvate dehydrogenase complex deficiency ⁶			Waardenburg syndrome		
<input type="checkbox"/> <i>DLAT</i>	<i>DLAT</i>	2	<input type="checkbox"/> type 1 & 3, <i>PAX3</i>	<i>PAX3</i>	2
<input type="checkbox"/> <i>DLD</i>	<i>DLD</i>	2	<input type="checkbox"/> type 2, <i>MITF</i>	<i>MITF</i>	2
<input type="checkbox"/> <i>PDHA1</i>	<i>PDHA1</i>	2	<input type="checkbox"/> type 2d, <i>SNAI2</i>	<i>SNAI2</i>	2
<input type="checkbox"/> <i>PDHB</i>	<i>PDHB</i>	2	<input type="checkbox"/> type 2e, <i>SOX10</i>	<i>SOX10</i>	2
<input type="checkbox"/> <i>PDHX</i>	<i>PDHX</i>	2			

Disease	Gene	TAT (months)	Disease	Gene	TAT (months)
Walker-Warburg (-like) syndrome (consanguinity: yes / no) ⁷		2	<input type="checkbox"/> Warburg micro syndrome	<i>RAB3GAP</i>	2
<input type="checkbox"/> <i>POMT1</i> <input type="checkbox"/> <i>FCMD</i> <input type="checkbox"/> Homozygosity testing ⁷			<input type="checkbox"/> West syndrome	<i>ARX</i>	2
<input type="checkbox"/> <i>POMT2</i> <input type="checkbox"/> <i>FKRP</i>			<input type="checkbox"/> Wolfram syndrome (DIDMOAD)	<i>WFS1</i>	2
<input type="checkbox"/> <i>POMGnT1</i> <input type="checkbox"/> <i>LARGE</i>					

FOOTNOTES

1. Please include serum for hepcidin testing, see 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).