



Accreditation number 142 - EN/ISO 15189



**Request for DNA testing**

Academic Medical Centre  
Dept. of Clinical Genetics, M1 – 107  
Meibergdreef 9  
1105 AZ AMSTERDAM – The Netherlands  
Postbus 22660  
1100 DD AMSTERDAM – The Netherlands

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Dr. E.J.W. Redeker      Dr. R.H. Lekanne dit Deprez  
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Tel.: +31 20 566 78 99  
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**Ophthalmogenetics:**

The Netherlands Institute for Neuroscience (NIN)

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**PATIENT ID CARD (THE NEXT FIELDS ARE MANDATORY) PLEASE FILL IN:**

**Patient name:** \_\_\_\_\_

**Date of birth:** \_\_\_\_\_

**Sex:** \_\_\_\_\_

**Home address:** \_\_\_\_\_

**Town/country:** \_\_\_\_\_

**Telephone:** \_\_\_\_\_

**Billing address:** \_\_\_\_\_

**Local ID:** \_\_\_\_\_

**REFERRING DOCTOR (THE NEXT FIELDS ARE MANDATORY) PLEASE FILL IN:**

**Referring doctor:** \_\_\_\_\_ **Telephone/tracer:** \_\_\_\_\_

**Hospital:** \_\_\_\_\_ **E-mail:** \_\_\_\_\_

**Department:** \_\_\_\_\_ **Date of sampling:** \_\_\_\_\_

**Address:** \_\_\_\_\_ **Copies to:** \_\_\_\_\_

**Town/Country:** \_\_\_\_\_

Have family members been referred to our lab before?  Yes  No

**Name:** \_\_\_\_\_

**Relation to the patient:** \_\_\_\_\_ **Family number (only when known):** P \_\_\_\_\_

**Consanguinity in the family:**  Yes (Indicate in pedigree, page 4)  No

**TYPE OF REQUEST:**

- Confirmation of clinical diagnosis
- Confirmation of clinical suspicion
- Carrier detection (recessive disease)
- Pre symptomatic test
- Genotyping testing for future prenatal test
- Other:

Research project;  
code: \_\_\_\_\_

**URGENT – EXCLUSIVELY AFTER CONTACTING:**

[kg\\_dna@amc.uva.nl](mailto:kg_dna@amc.uva.nl)

- No
- Yes

Result before: \_\_\_\_\_

Pre-natal analysis: 2 – 3 weeks

Familial mutation: 6 weeks

- DNA storage for future test;  
**Please indicate test (Mandatory field):**

**TO BE FILLED IN BY DNA-LABORATORY EMPLOYEES**

<b>Material</b> <input type="checkbox"/> BL <input type="checkbox"/> CV <input type="checkbox"/> FI <input type="checkbox"/> AMN <input type="checkbox"/> DNA <input type="checkbox"/> Biopsy	<input type="checkbox"/> P	<b>Date sample received</b>	<b>Test number</b>	<b>Family number</b>
	<input type="checkbox"/> PR			P
<b>Quantity:</b>	KG	<b>Initials acceptance</b>	<b>Initials registration</b>	<b>DNA-number</b>
				D / /

You can download our request form on: [www.dnadiagnostiek.nl](http://www.dnadiagnostiek.nl)

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**Sending**

See page 4

**Packaging Requirements for Sample Transportation**

See page 4

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Test / Gene	Turnaround Time	Test / Gene	Turnaround Time
<b>Cardiologic</b>			
<input type="checkbox"/> Primary electrical disease; <i>please select</i>		<input type="checkbox"/> Wolff-Parkinson-White syndrome	
<input type="checkbox"/> Brugada syndrome ( <i>SCN5A</i> )	6	<input type="checkbox"/> <i>LAMP2</i>	3
<input type="checkbox"/> <i>JLN1 (KCNQ1)</i>	3	<input type="checkbox"/> <i>PRKAG2</i>	3
<input type="checkbox"/> <i>JLN2 (KCNE1)</i>	3	<input type="checkbox"/> Other cardiologic:...	
<input type="checkbox"/> <i>LQT1 (KCNQ1)</i>	3	<b>Diverse</b>	
<input type="checkbox"/> <i>LQT2 (KCNH2)</i>	3	<input type="checkbox"/> Angelman syndrome; <i>please select</i> :	
<input type="checkbox"/> <i>LQT3 (SCN5A)</i>	6	<input type="checkbox"/> Methylation analysis	3
<input type="checkbox"/> <i>LQT5 (KCNE1)</i>	3	<input type="checkbox"/> Mutation analysis ( <i>UBE3A</i> )	3
<input type="checkbox"/> <i>LQT6 (KCNE2)</i>	3	<input type="checkbox"/> <i>UPD15 / Deletion</i>	2
<input type="checkbox"/> <i>LQT7 (Timothy syndrome) (CACNA1C)</i>	2	<input type="checkbox"/> Hereditary Angioedema (HAE) ( <i>SERPING1</i> )	3
<input type="checkbox"/> <i>LQT8 (Andersen syndrome) (KCNJ2)</i>	3	<input type="checkbox"/> Azoospermia (Y-deletions)	2
<input type="checkbox"/> Atrial Standstill ( <i>SCN5A</i> )	6	<input type="checkbox"/> Barth syndrome ( <i>TAZ</i> )	3
<input type="checkbox"/> Sick sinus syndrome ( <i>SCN5A</i> )	6	<input type="checkbox"/> Beckwith-Wiedemann syndrome	
<input type="checkbox"/> Conduction defect ( <i>SCN5A</i> )	6	<input type="checkbox"/> Methylation analysis ( <i>LIT1 &amp; H19</i> )	3
 		<input type="checkbox"/> Mutation analysis ( <i>CDKN1C</i> )	3
<input type="checkbox"/> <i>SQT1 (KCNH2)</i>	3	<input type="checkbox"/> <i>UPD11p15</i>	2
<input type="checkbox"/> <i>SQT2 (KCNQ1)</i>	3	<input type="checkbox"/> Carvajal syndrome ( <i>DSP</i> )	6
 		<input type="checkbox"/> Cherubism ( <i>SH3BP2</i> )	3
<input type="checkbox"/> <i>LQT MLPA (KCNQ1, KCNH2, KCNE1, KCNE2)</i>	2	<input type="checkbox"/> Cholestasis; <i>please select</i>	
<input type="checkbox"/> <i>LQT panel</i>	6	<input type="checkbox"/> <i>BRIC1 (ATP8B1)</i>	4,5
<i>(KCNQ1, KCNH2, SCN5A, KCNE1, KCNE2, MLPA)</i>		<input type="checkbox"/> <i>PFIC1 (ATP8B1)</i>	4,5
<input type="checkbox"/> <i>CPVT; please select</i>		<input type="checkbox"/> <i>BRIC2 (ABCB11)</i>	4,5
<input type="checkbox"/> <i>CASQ2</i>	3	<input type="checkbox"/> <i>PFIC2 (ABCB11)</i>	4,5
<input type="checkbox"/> <i>RYR2</i>	6	<input type="checkbox"/> <i>LPAC syndrome (ABCB4)</i>	4,5
<input type="checkbox"/> <i>ARVD; please select</i>		<input type="checkbox"/> <i>ICP (ABCB4)</i>	4,5
<input type="checkbox"/> <i>DSC2</i>	4,5	<input type="checkbox"/> <i>PFIC3 (ABCB4)</i>	4,5
<input type="checkbox"/> <i>DSG2</i>	4,5	<input type="checkbox"/> Cornelia de Lange syndrome	
<input type="checkbox"/> <i>DSP</i>	6	<input type="checkbox"/> <i>NIPBL</i>	6
<input type="checkbox"/> <i>JUP</i>	4,5	<input type="checkbox"/> <i>SMC1A</i>	4,5
<input type="checkbox"/> <i>TMEM43</i>	4,5	<input type="checkbox"/> Cyclic/congenital neutropenia ( <i>ELA2</i> )	3
<input type="checkbox"/> <i>PKP2 (+ MLPA)</i>	4,5	<input type="checkbox"/> Denys-Drash syndrome ( <i>WT1</i> )	3
<input type="checkbox"/> <i>RYR2</i>	6	<input type="checkbox"/> Emery Dreifuss muscular dystrophy	
 		<input type="checkbox"/> <i>EMD (STA)</i>	3
<input type="checkbox"/> <i>ARVD panel; please select</i>	4,5	<input type="checkbox"/> <i>LMNA (+MLPA)</i>	3
<i>(PKP2, DSP, DSG2, DSC2, JUP, TMEM43 + MLPA)</i>		<input type="checkbox"/> Fabry disease ( <i>GLA</i> )	3
<input type="checkbox"/> <i>Carney complex type 1 (PRKAR1A)</i>	3	<input type="checkbox"/> Familial Hypercholanemia (FHCA) ( <i>BAAT</i> )	3
<input type="checkbox"/> <i>Danon disease (LAMP2)</i>	3	<input type="checkbox"/> Familial hypercholesterolemia ( <i>LDLR/APOB</i> )	3
<input type="checkbox"/> <i>Holt-Oram syndrome (TBX5)</i>	3	<input type="checkbox"/> Fragile X syndrome ( <i>FMR1</i> )	2
<input type="checkbox"/> <i>Cardiomyopathy; please select.</i>		<input type="checkbox"/> Frasier syndrome ( <i>WT1</i> )	3
<input type="checkbox"/> Hypertrophic		<input type="checkbox"/> Gaucher disease ( <i>GBA</i> )	3
<input type="checkbox"/> Hypertrophic + <input type="checkbox"/> Conduction disease		<input type="checkbox"/> Glucocorticoid Remediab Aldosteronism ( <i>CYP11B1/CYP11B2</i> )	3
<input type="checkbox"/> Non compaction		<input type="checkbox"/> Goltz-Gorlin Syndrome ( <i>PORCN</i> )	3
<input type="checkbox"/> Restrictive		<input type="checkbox"/> Hemihypertrophy	
<input type="checkbox"/> <i>MYBPC3</i>	6	<input type="checkbox"/> Methylation analysis ( <i>LIT1 &amp; H19</i> )	3
<input type="checkbox"/> <i>MYH7</i>	6	<input type="checkbox"/> <i>UPD11p15</i>	2
<input type="checkbox"/> <i>TNNT2</i>	3	<input type="checkbox"/> Hemofilia A (HEMA) ( <i>F8</i> )	4,5
<input type="checkbox"/> <i>TPM1</i>	3	<input type="checkbox"/> Hyper IgE syndrome ( <i>STAT3</i> )	3
<input type="checkbox"/> <i>TNNI3</i>	3	<input type="checkbox"/> Lipodystrophy ( <i>LMNA</i> ) (+ MLPA)	3
<input type="checkbox"/> <i>GLA</i>	3	<input type="checkbox"/> Methemoglobinemia ( <i>DIA1</i> )	3
<input type="checkbox"/> <i>EMD (STA)</i>	3		
<input type="checkbox"/> <i>LAMP2</i>	3		
<input type="checkbox"/> <i>LMNA (+ MLPA)</i>	3		
<input type="checkbox"/> <i>PRKAG2</i>	3		
<input type="checkbox"/> <i>TAZ</i>	3		
<input type="checkbox"/> <i>SCN5A</i>	6		
<input type="checkbox"/> <i>Carvajal syndrome (DSP)</i>	6		
<input type="checkbox"/> <i>Congenital heart defects (i.e. Tetralogy of Fallot, ASD, VSD); please select:</i>			
<input type="checkbox"/> <i>GATA4</i>	3		
<input type="checkbox"/> <i>NKX2-5</i>	3		

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<b>Diverse</b>			
<input type="checkbox"/> Niemann-Pick disease Type A & B ( <i>SMPD1</i> )	3	<input type="checkbox"/> Central hypothyroidism	
<input type="checkbox"/> Niemann-Pick disease Type C1 ( <i>NPC1</i> )	4,5	<input type="checkbox"/> <i>HESX1</i>	3
<input type="checkbox"/> Niemann-Pick disease Type C2 ( <i>NPC2</i> )	3	<input type="checkbox"/> <i>LHX3</i>	3
<input type="checkbox"/> Prader-Willi syndrome		<input type="checkbox"/> <i>PROP1</i>	3
<input type="checkbox"/> Methylation analysis	3	<input type="checkbox"/> <i>POU1F1</i>	3
<input type="checkbox"/> UPD15 / deletion	2	<input type="checkbox"/> <i>SOX2</i>	3
<input type="checkbox"/> Premature Ovarian Failure (POF) ( <i>FMR1</i> )	2	<input type="checkbox"/> <i>TRH</i>	3
<input type="checkbox"/> Properdin deficiency( <i>PFC</i> )	3	<input type="checkbox"/> <i>TRHR</i>	3
<input type="checkbox"/> Rett syndrome		<input type="checkbox"/> <i>TSHB</i>	3
<input type="checkbox"/> <i>MECP2</i>	3	<input type="checkbox"/> <i>TSHR</i>	3
<input type="checkbox"/> <i>CDKL5</i>	4,5	<input type="checkbox"/> Albright's Hereditary Osteodystrophy	3
<input type="checkbox"/> Riley-Day syndrome (fam. Dysautonomia) ( <i>IKBKAP</i> )	2	<input type="checkbox"/> Pseudohypoparathyroidism Ia	3
<input type="checkbox"/> Shwachman-Diamond syndrome ( <i>SBDS</i> )	3	<input type="checkbox"/> <i>GNAS</i>	3
<input type="checkbox"/> Silver-Russell syndrome		<input type="checkbox"/> <i>PTHR1</i>	3
<input type="checkbox"/> Methylation 11p15 ( <i>H19</i> )	3	<input type="checkbox"/> McCune-Albright syndrome ( <i>Codon 201 GNAS</i> )	3
<input type="checkbox"/> UPD7	2	<input type="checkbox"/> Hyperthyroidism ( <i>TSHR</i> )	3
<input type="checkbox"/> Simpson-Golabi-Behmel syndrome ( <i>GPC3</i> )	3	<input type="checkbox"/> Thyroid Hormone Resistance ( <i>THR3</i> )	3
<input type="checkbox"/> Skin fragility Woolly Hair syndrome ( <i>DSP</i> )	6	<b>Ophthalmogenetics</b>	
<input type="checkbox"/> Tay-Sachs disease ( <i>HEXA</i> )	3	<input type="checkbox"/> Albinism, X-linked ( <i>OA1</i> )	
<input type="checkbox"/> Wilms Tumor ( <i>WT1</i> )	3	<input type="checkbox"/> Albinism, Recessive	
<input type="checkbox"/> Genotyping concerning: ...	3	<input type="checkbox"/> <i>OCA1 (Tyr)</i>	
<input type="checkbox"/> MLPA concerning: ...		<input type="checkbox"/> <i>OCA2 (Pgene)</i>	
<input type="checkbox"/> Other: ...		<input type="checkbox"/> Aniridia <input type="checkbox"/> Anophtalmia <input type="checkbox"/> Macrophtalmia	
<b>Oncogenetics</b>		<input type="checkbox"/> <i>PAX6 (+ MLPA)</i>	3
<input type="checkbox"/> FAP Familial Adenomatous Polyposis ( <i>APC</i> )	6	<input type="checkbox"/> <i>SOX2 (+ MLPA)</i>	3
<input type="checkbox"/> Multiple colorectal adenomas ( <i>MUTYH</i> )	3	<input type="checkbox"/> Doyme Honeycomb retinal dystrophy ( <i>EFEMP1</i> )	
<input type="checkbox"/> Familial nonpolyposis colon cancer (HNPCC):		<input type="checkbox"/> Best macular dystrophy (Early-onset) ( <i>VMD2</i> )	
<i>please select:</i>		<input type="checkbox"/> Dominant Optic Atrophy ( <i>OPA1</i> )	
<input type="checkbox"/> <i>MLH1</i>	4,5	<input type="checkbox"/> Leber Congenital Amaurosis ( <i>LCA</i> )	
<input type="checkbox"/> <i>MSH2</i>	3	<input type="checkbox"/> Pattern Dystrophy (Dominant) ( <i>RDS</i> )	
<input type="checkbox"/> <i>MSH6</i>	3	<input type="checkbox"/> Primary Glaucoma; <i>please select:</i>	
<input type="checkbox"/> <i>PMS2</i>	3	<input type="checkbox"/> <i>CYP1B1</i>	
		<input type="checkbox"/> <i>MYOC</i>	
		<input type="checkbox"/> <i>OPTN</i>	
<b>Endocrinology</b>		<input type="checkbox"/> Pseudoxanthoma elasticum ( <i>ABCC6</i> )	
<input type="checkbox"/> Hyperinsulinism		<input type="checkbox"/> Retinoschisis, X-linked ( <i>XLRS1</i> )	
<input type="checkbox"/> <i>ABCC8</i>	6	<input type="checkbox"/> Retinitis pigmentosa; <i>please select :</i>	
<input type="checkbox"/> <i>GCK</i>	3	<input type="checkbox"/> Dominant	
<input type="checkbox"/> <i>GLUD1</i>	3	<input type="checkbox"/> Recessive	
<input type="checkbox"/> <i>KCNJ11</i>	3	<input type="checkbox"/> X-Linked	
<input type="checkbox"/> Thyroidal hypothyroidism		<input type="checkbox"/> Stargardt Disease 1 (recessive) ( <i>ABCA4</i> )	
<input type="checkbox"/> <i>DUOX2</i>	6	<input type="checkbox"/> Other ophthalmology...	
<input type="checkbox"/> <i>DUOXA2</i>	3		
<input type="checkbox"/> <i>FOXE1</i>	3		
<input type="checkbox"/> <i>NKX2-1</i>	3		
<input type="checkbox"/> <i>PAX8</i>	3		
<input type="checkbox"/> <i>TPST2</i>	3		
<input type="checkbox"/> <i>TG</i>	6		
<input type="checkbox"/> <i>IYD</i>	3		
<input type="checkbox"/> <i>TPO</i>	3		
<input type="checkbox"/> <i>TSHR</i>	3		

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#### Sending

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#### Packaging Requirements for Sample Transportation

See page 4

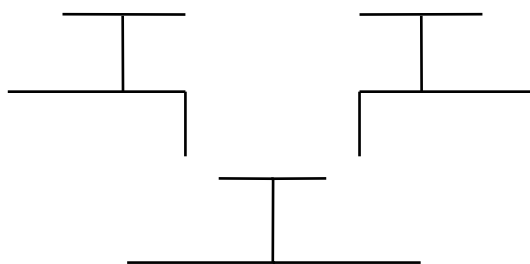
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Pedigree:

Indicate patient with arrow (↗)

Indicate affected people with ■

Indicate carriers with ■□



### **Sending**

Do not freeze the samples. Send at room temperature. EC countries: from Monday to Wednesday by regular post, after Wednesday send by express. Send prenatal material on the same day with a courier. Non EC countries: Send with a courier.

### **Packaging Requirements for Sample Transportation**

Packing Instruction 650 (PI650) applied to UN3373:

<http://www.bwhct.nhs.uk/regionalgenetics/downloads/Packaging%20Requirements%20for%20Sample%20Transportation1.pdf>

Necessary equipment: primary Leak-proof receptacle, absorbent material, safety bag, outer packaging.

(For Belgium: Zie ook de TNT post voorschriften: <http://www.tntpost.nl/zakelijk/brieven/brieven-en-kaarten-sturen/diagnostische-monsters/index.aspx>)

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