



Accreditation number 142 - EN/ISO 15189



Request for DNA testing

Academic Medical Centre
Dept. of Clinical Genetics, M1 – 107
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Ophthalmogenetics:

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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

- 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

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

You can download our request form on: www.dnadiagnostiek.nl

Material

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Sending

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Packaging Requirements for Sample Transportation

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Test / Gene	Turnaround Time	Test / Gene	Turnaround Time
Cardiologic			
<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	Diverse	
<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 (Andersen syndrome) (KCNJ2)</i>	2	<input type="checkbox"/> Hereditary Angioedema (HAE) (<i>SERPING1</i>)	3
<input type="checkbox"/> <i>LQT8 (Timothy syndrome) (CACNA1C)</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 & H19</i>)	3
<input type="checkbox"/> <i>SQT1 (KCNH2)</i>	3	<input type="checkbox"/> Mutation analysis (<i>CDKN1C</i>)	3
<input type="checkbox"/> <i>SQT2 (KCNQ1)</i>	3	<input type="checkbox"/> <i>UPD11p15</i>	2
<input type="checkbox"/> <i>LQT MLPA (KCNQ1, KCNH2, KCNE1, KCNE2)</i>	2	<input type="checkbox"/> Carvajal syndrome (<i>DSP</i>)	6
<input type="checkbox"/> <i>LQT panel</i> (<i>KCNQ1, KCNH2, SCN5A, KCNE1, KCNE2, MLPA</i>)	6	<input type="checkbox"/> Cherubism (<i>SH3BP2</i>)	3
<input type="checkbox"/> <i>CPVT; please select</i>		<input type="checkbox"/> Cholestasis; <i>please select</i>	
<input type="checkbox"/> <i>CASQ2</i>	3	<input type="checkbox"/> <i>BRIC1 (ATP8B1)</i>	4,5
<input type="checkbox"/> <i>RYR2</i>	6	<input type="checkbox"/> <i>PFIC1 (ATP8B1)</i>	4,5
<input type="checkbox"/> <i>ARVD; please select</i>		<input type="checkbox"/> <i>BRIC2 (ABCB11)</i>	4,5
<input type="checkbox"/> <i>DSC2</i>	4,5	<input type="checkbox"/> <i>PFIC2 (ABCB11)</i>	4,5
<input type="checkbox"/> <i>DSG2</i>	4,5	<input type="checkbox"/> <i>LPAC syndrome (ABCB4)</i>	4,5
<input type="checkbox"/> <i>DSP</i>	6	<input type="checkbox"/> <i>ICP (ABCB4)</i>	4,5
<input type="checkbox"/> <i>JUP</i>	4,5	<input type="checkbox"/> <i>PFIC3 (ABCB4)</i>	4,5
<input type="checkbox"/> <i>TMEM43</i>	4,5	<input type="checkbox"/> Cornelia de Lange syndrome	
<input type="checkbox"/> <i>PKP2 (+ MLPA)</i>	4,5	<input type="checkbox"/> <i>NIPBL</i>	6
<input type="checkbox"/> <i>RYR2</i>	6	<input type="checkbox"/> <i>SMC1A</i>	4,5
<input type="checkbox"/> <i>ARVD panel; please select</i> (<i>PKP2, DSP, DSG2, DSC2, JUP, TMEM43 + MLPA</i>)	4,5	<input type="checkbox"/> <i>Cyclic/congenital neutropenia (ELA2)</i>	3
<input type="checkbox"/> <i>Carney complex type 1 (PRKAR1A)</i>	3	<input type="checkbox"/> <i>Denys-Drash syndrome (WT1)</i>	3
<input type="checkbox"/> <i>Danon disease (LAMP2)</i>	3	<input type="checkbox"/> <i>Emery Dreifuss muscular dystrophy</i>	
<input type="checkbox"/> <i>Holt-Oram syndrome (TBX5)</i>	3	<input type="checkbox"/> <i>EMD (STA)</i>	3
<input type="checkbox"/> <i>Cardiomyopathy; please select.</i>		<input type="checkbox"/> <i>LMNA (+MLPA)</i>	3
<input type="checkbox"/> Hypertrophic		<input type="checkbox"/> <i>Fabry disease (GLA)</i>	3
<input type="checkbox"/> Hypertrophic + <input type="checkbox"/> Conduction disease		<input type="checkbox"/> <i>Familial Hypercholanemia (FHCA) (BAAT)</i>	3
<input type="checkbox"/> Non compaction		<input type="checkbox"/> <i>Familial hypercholesterolemia (LDLR/APOB)</i>	3
<input type="checkbox"/> Restrictive		<input type="checkbox"/> <i>Fragile X syndrome (FMR1)</i>	2
<input type="checkbox"/> <i>MYBPC3</i>	6	<input type="checkbox"/> <i>Frasier syndrome (WT1)</i>	3
<input type="checkbox"/> <i>MYH7</i>	6	<input type="checkbox"/> <i>Gaucher disease (GBA)</i>	3
<input type="checkbox"/> <i>TNNT2</i>	3	<input type="checkbox"/> <i>Glucocorticoid Remediab Aldosteronism (CYP11B1/CYP11B2)</i>	3
<input type="checkbox"/> <i>TPM1</i>	3	<input type="checkbox"/> <i>Goltz-Gorlin Syndrome (PORCN)</i>	3
<input type="checkbox"/> <i>TNNI3</i>	3	<input type="checkbox"/> <i>Hemihypertrophy</i>	
<input type="checkbox"/> <i>GLA</i>	3	<input type="checkbox"/> Methylation analysis (<i>LIT1 & H19</i>)	3
<input type="checkbox"/> <i>EMD (STA)</i>	3	<input type="checkbox"/> <i>UPD11p15</i>	2
<input type="checkbox"/> <i>LAMP2</i>	3	<input type="checkbox"/> <i>Hemofilia A (HEMA) (F8)</i>	4,5
<input type="checkbox"/> <i>LMNA (+ MLPA)</i>	3	<input type="checkbox"/> <i>Hyper IgE syndrome (STAT3)</i>	3
<input type="checkbox"/> <i>PRKAG2</i>	3	<input type="checkbox"/> <i>Lipodystrophy (LMNA) (+ MLPA)</i>	3
<input type="checkbox"/> <i>TAZ</i>	3	<input type="checkbox"/> <i>Methemoglobinemia (DIA1)</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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Diverse			
<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	Ophthalmogenetics	
<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	
Oncogenetics		<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>PMS2</i>	3	<input type="checkbox"/> <i>MYOC</i>	
<input type="checkbox"/> <i>PMS2</i>	3	<input type="checkbox"/> <i>OPTN</i>	
Endocrinology		<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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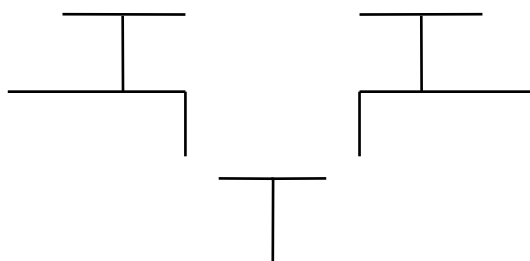
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Pedigree:

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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