



University Medical Center Nijmegen
 120 DNA-diagnostics
 PO Box 9101
 6500 HB Nijmegen
street address:
 Adelbertusplein 1
 6525 EK Nijmegen
 The Netherlands

Head: Dr. H. Scheffer
 Mrs. Dr. E.H. Hoefsloot
 Dr. F.A. Hol
 Mrs. Dr. M.J.L. Ligtenberg
 Dr. E.A. Sistermans

Fax *31(0)24-3616658
 Tel *31(0)24-3613799
 E-mail: dnadiagn@antrg.umcn.nl
www.dnadiagnostieknijmegen.nl

Patient information / Please fill in completely

Name+initials Date of birth Sex	
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INVOICE WILL BE SENT TO THE REFERRING PHYSICIAN

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

PLEASE USE A SEPARATE FORM FOR EACH BLOOD OR DNA SAMPLE

► **Sample :** extracted DNA blood, date of withdrawal:

► **Conditions and Shipment**

We prefer extracted DNA, but if you choose to send in blood make sure the following applies:

1. 10 to 20 ml EDTA Blood in unbreakable tubes (neonates at least 5 ml).
2. Please put name, date of birth and sex on each tube
3. Ship samples at room temperature. **Do not freeze blood samples!**
4. Samples should arrive in our lab within 4 days after withdrawal

In case of further questions please contact us at phone +31243613799 or mail to dnadiagn@antrg.umcn.nl

► **Reason for referral :**

- Confirmation of diagnosis
- Carrier testing
- Other :

► **Remarks / relevant clinical information :**

To be filled in by our staff

Date of receipt:

Remarks:

- Are there other affected family members? Yes / No
- Has material from family member(s) been sent in previously? Yes / No
- If yes, please draw pedigree on the last page**

► Reporting of results

Prenatal diagnosis	2 -3 weeks
Testing of a known mutation in the family	6 weeks
Testing for of a yet unknown mutation in the family	see table (months)

Disease	Gene	Reporting of Results (months)
○ Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency	CYP21A2	6
○ Adrenoleukodystrophy	ABCD1	6
○ ADULT syndrome	TP63	6
○ AEC syndrome (Hay-Wells)	TP63	6
○ Angelman syndrome ¹	methylation ¹	3
○ Angioneurotic edema	SERPING2	6
○ Bannayan-Riley-Ruvalcaba syndroom	PTEN	6
○ Bartter syndrome, type II	KCNJ1	6
○ Brachydactyly, type b	ROR2	3
○ Branchiootorenal dysplasia (BOR syndrome)	EYA1	6
○ Canavan disease	ASPA	6
○ Cerebrotendinous xanthomatosis	CYP27A1	6
○ Choroideremia	CHM	6
○ Complex I deficiency, autosomal recessive	NDUV1	6
○ Complex I deficiency, autosomal recessive	NDUV2	6
○ Complex I deficiency, autosomal recessive	NDUFS1	6
○ Complex I deficiency, autosomal recessive	NDUFS2	6
○ Complex I deficiency, autosomal recessive	NDUFS4	6
○ Complex I deficiency, autosomal recessive	NDUFS7	6
○ Complex I deficiency, autosomal recessive	NDUFS8	6
○ Complex II deficiency, autosomal recessive	SDHA	6
○ Complex III deficiency, autosomal recessive	BCS1L	6
○ Complex IV deficiency, autosomal recessive	SCO1	6
○ Complex IV deficiency, autosomal recessive	SCO2	6
○ Complex IV deficiency, autosomal recessive	COX10	6
○ Complex IV deficiency, autosomal recessive	SURF1	6
○ Cowden syndrome	PTEN	6
○ Cystathionine beta-synthase (CBS) deficiency	CBS	6
○ Cystinosis, nephropathic	CTNS	6
○ Deafness, autosomal dominant type 2 (DFNA2)	KCNQ4	3 ²
○ Deafness, autosomal dominant type 9 (DFNA9)	COCH	3 ²
○ Deafness, autosomal dominant type 6/14 (DFNA6/14) (LFSNHL)	WFS1	3
○ Deafness, autosomal recessive type 1 (DFNB1)	GJB2	6
○ Deafness, autosomal recessive type 4 (DFNB4)	DFNB1	3
○ Deafness, X-linked type 1 (DFN1)	DDP	3
○ Deafness, X-linked type 3 (DFN3)	POU3F4	3
○ Dent disease (nephrolithiasis type 1)	CLCN5	6
○ Diabetes insipidus, nephrogenic, autosomal dominant/recessive	AQP2	6
○ Diabetes insipidus, nephrogenic, X-linked	AVPR2	6
○ Diabetes insipidus, central	AVP	6
○ EEC syndrome	TP63	6
○ Epidermolysis bullosa dystrophica ⁵	COL7A1	6 ⁵
○ Fragile X syndrome	FMR1	2
○ Friedreich ataxia ³	FRDA	3 ³
○ Gastric cancer (E-cadherin)	CDH1	6
○ Gitelman syndrome	SLC12A3	6
○ Glucose transporter deficiency type 1	SLC2A1	3
○ Hyper IgD syndrome	MVK	3
○ Ichthyosis, lamellar	TGM1	6
○ Ichthyosis, X-linked ⁴	STS	3 ⁴
○ Kallmann syndrome	KAL1	6
○ Kearns-Sayre syndrome	mtDNA	3
○ Leber hereditary optic atrophy (LHON)	mtDNA	3
○ Leigh syndrome, mitochondrial	mtDNA	3
○ Leigh syndrome, autosomal recessive	SURF1	consult
○ Leopard Syndrome	PTPN11	6
○ Limb-mammary syndrome	TP63	6
○ MELAS syndrome	mtDNA	3
○ MERFF syndrome	mtDNA	3
○ Methylenetetrahydrofolate-5,10 reductase deficiency	MTHFR	6
○ Mevalonate kinase deficiency	MVK	3
○ Mitochondrial disease (myopathy, encephalopathy, etc)	mtDNA	3
○ Mohr-Tranebjaerg syndrome (DFN1)	DDP	3
○ Multiple synostoses syndrome (SYNS1)	NOG	3
○ Muscle Eye Brain disease (MEB)	POMGNT1	6
○ Myoadenylate deaminase deficiency	AMPD1	6
○ Myofosforilase deficiëntie (McArdle disease)	PHKB	6
○ Myotonic dystrophy type 1	DMPK	3
○ Myotonic dystrophy type 2 (proximal myotonic myopathy)	ZNF9	3
○ Nail-Patella syndrome	LMX1b	6
○ Neuropathy, ataxia, and retinitis pigmentosa (NARP)	mtDNA	3
○ Nijmegen breakage syndrome	NBS1	6
○ Noonan syndrome	PTPN11	6
○ Norrie disease	NDP	6

O	Oculopharyngeal muscular dystrophy	OPMD	3
O	Pancreatitis, hereditary	PCTT	3
O	Pelizaeus-Merzbacher disease	PLP1	6
O	Pendred syndrome	SLC26A4	6
O	Pyruvaat dehydrogenase complex deficiency	PDHA1	6
O	Pyruvaat dehydrogenase complex deficiency	PHE3	6
O	Polyposis coli juvenile	PTEN	6
O	Polyposis coli, juvenile	MADH4	consult
O	Prader-Willi syndrome	methylation	3
O	Pyruvaat dehydrogenase complex deficiency	PDHA1	6
O	Pyruvaat dehydrogenase complex deficiency	PHE3	6
O	Renal tubular acidosis with progressive nerve deafness	ATP6B1	6
O	Rett syndrome	MECP2	6
O	Robinow syndrome, autosomal recessive	ROR2	3
O	Spastic paraplegia type 2, X-Linked	SPG2	6
O	Spastic paraplegia type 4, autosomal dominant	SPG4	6
O	Spastic paraplegia type 7, autosomal recessive	PARAPLEGIN	6
O	Spondyloepiphyseal dysplasia tarda, X-linked	SEDL	3
O	Split hand/foot malformation (SHFM)	TP63	6
O	Symphalangism, proximal	NOG	3
O	Torsion Dystonia, early onset	DYT1	3
O	Treacher Collins-Francheschetti syndrome	TCOF	6
O	Tyrosine hydroxylase	TH	6
O	Usher Syndrome, type 1b	MYO7A	3 ²
O	Usher Syndrome, type 2a	USH2A	3 ²
O	Vitamin E, familial isolated deficiency of (AVED)	TTPA	6
O	Waardenburg syndrome type 1	PAX3	6
O	Waardenburg Syndrome, type 2a	MITF	6
O	Waardenburg Syndrome, type 3	PAX3	6
O	Walker-Warburg syndrome	POMT1	6
O	Wolfram syndrome (DIDMOAD)	WFS1	3

Footnotes

1. Mutation analysis of UBE3A possible after consultation
2. Testing of frequent mutations: 3 months; remainder of the gene after consultation
3. Analysis of repeatexpansions: 3 months; sequencing FRDA gene 6 months
4. Deletion testing only
5. Include blood of both parents

