

MITOCHONDRIAL DIAGNOSTICS (BIOCHEMISTRY & GENETICS)

Stichting Klinisch-Genetisch Centrum Nijmegen
Radboud University Nijmegen Medical Center
830 Lab. Genetic, Endocrine & Metabolic Disorders
Geert Grooteplein 10, NL-6525 GA Nijmegen
phone: +31 24 3614428 fax: +31 24 3618900
www.ncmd.nl

Dr. L. v.d. Heuvel, DNA diagnostics
Bert.vandenHeuvel@radboudumc.nl
Dr. R. Rodenburg, mitochondrial diagnostics
Richard.Rodenburg@radboudumc.nl
Prof. Dr. J. Smeitink, mitochondrial medicine
Jan.Smeitink@radboudumc.nl



Patient

Name

Date of Birth

Gender: M / F

Patient is deceased, date

Patient does not give permission for long term storage of samples for the purpose of additional diagnostic or scientific research of the sample(s) at a later stage.

Billing address

Referring physician

Name

Hospital

Department

Address

Phone

FAX

E-mail

Send report to: referring physician

other:

Requested investigation:

Complete biochemical and genetic work-up

Biochemical diagnostics

Genetic diagnostics

Specimen:

Muscle Skin / fibroblasts Blood Other:

Date collected:

Time collected:

(For muscle biopsies) anesthesia: Local General

biopsy taken from: *Musculus*

To be filled out by lab employee:

Ontvangst datum:
tijdstip:

Opmerkingen:

Conditions and shipment:

Please check our website www.ncmd.nl

Protocols can also be requested by phone at +31 24 3619673

Clinical information (please complete both sides of this form):

Biometrics

length: cm P 3 10 50 90

weight: kg P 3 10 50 90

weight to height P 3 10 50 90

head circumf.: cm P 3 10 50 90

General physical abnormalities / restcategory

263 failure to thrive

118 SIDS / 117 near SIDS

120 respiratory insufficiency

229 abnormal breathing pattern

140 lipomas

161 dysmorphic features:

199 other:

Central nervous system

200 mental retardation

281 congenital 282 decline

225 dementia

220 microcephaly

221 macrocephaly

280 impaired consciousness / 216 coma

217 lethargy

210 epilepsy / epileptiform EEG

223 behavioral abnormalities / 267 autism

219 unusual crying

201 motor retardation

283 congenital 284 decline

251 hypertonia / 206 spasticity

252 hypotonia

Central nervous system (continued)

- 228 extrapyramidal signs
- 230 dystonia
- 214 ataxia
- 291 myoclonus
- 299 other:
- 115 speech disturbance
- 227 pyramidal signs
- 235 hemiparesis
- 232 stroke-like episodes
- 292 migraine
- 293 non-migraineous headache
- 299 other:

Muscle + periferal nervous system

- 294 myopathy (not incl. eye muscles)
- 256 exercise intolerance
- 260 muscle cramps
- 295 muscle pain without cramps
- 252 hypotonia
- 253 muscular dystrophy
- 254 muscle weakness
- 257 rhabdomyolysis
- 268 polyneuropathy
- 299 other:

Eyes and hearing

- 114 loss of hearing / deafness
- 906 ptosis
- 940 ophthalmoplegia
- 904 strabismus
- 903 nystagmus
- 901 cataract
- 902 corneal disturbance
- 999 other:

Heart and circulation

- 171 cardiomyopathy
- 151 conduction defects / arrhythmias
- 109 hypertension
- 110 hypotension
- 199 other:

Gastrointestinal tract and liver

- 302 feeding problems
- 301 diarrhea
- 331 cachexia
- 300 (cyclic) vomiting
- 320 pseudo-obstruction / 321 ileus
- 101 hepatomegaly
- 108 icterus
- 399 other:

Medication

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Kidneys

- 403 renal insufficiency
- 406 tubulopathy
- 401 polyuria
- 400 kidney stones
- 499 other:

Blood and immune system

- 603 anemia
- 607 leucopenia
- 606 trombocytopenia
- 102 splenomegaly
- 601 immunodeficiency / 600 recurrent infections
- 699 other:

Genetics

- 800 consanguinity
 - 802 sib of SIDS
 - 804 abortus
 - 820 positive familie anamnesis
 - 821 similar phenotype
 - 822 different phenotype:
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- family member(s) (in case material has been sent to us before):
- name/d.o.b.:
- family relation to patient:

Laboratory investigations

- 760 diabetes mellitus
- 720 hypoglycemia
- 722 lactate acidemia
- 723 acidosis
- 721 ketosis
- 726 elevated CK:
- 725 elevated ASAT / ALAT
- 724 elevated ammonia
- 750 elevated alanine:
- 751 elevated lactate / pyruvate ratio:
- 731 abnormal vitamine status:
- 752 abnormal urinary organic acids:
- 799 other:

Histological examination muscle biopsy

- 4000 ragged-red fibers
- 4001 COX negative fibers
- 4099 other:

Imaging

- 501 skeletal disturbed:
- 204 CT / MRI brain:
- 599 other:

Clinical differential diagnosis

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