MITOCHONDRIAL DIAGNOSTICS (BIOCHEMISTRY & GENETICS)

Stichting Klinisch-Genetisch Centrum Nijmegen
Radboud university medical center
Translational Metabolic Laboratory (TML)
Geert Grooteplein 10, NL-6525 GA Nijmegen
phone: +31 24 3614567 fax: +31 24 3618900
www.ncmd.nl

Dr. L. v.d. Heuvel, DNA diagnostics
b.vandenheuvel@radboudumc.nl
Dr. R. Rodenburg, mitochondrial diagnostics
r.rodenburg@radboudumc.nl
Prof. Dr. J. Smeitink, mitochondrial medicine
j.smeitink@radboudumc.nl

Patient
First name*:.....
Middle name*:.....
Family name* :.....
Date of Birth*:.....
Gender*: M / F
MRN Number.....
☐ Patient is deceased, date .....
☐ Patient does not give permission for lang term storage of samples for the purpose of additional diagnostic or scientific research of the sample(s) at a later stage
Billing addres:.....
.....

Referring physician
Name.....
Hospital:.....
Department:.....
Address:.....
Phone:.....
Fax:.....
E-mail:.....
Send report to: ☐ referring physician
☐ other:

• *These items are required*

Requested investigation:
☐ Complete biochemical and genetic work-up ☐ Biochemical diagnostics ☐ Genetical 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 www.nmcd.nl or www.radboudumc.nl/labgk
Protocols can also be requested by phone at +31 24 3614567

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

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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:.....

.....

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 disturbanced:.....
204 ☐ CT / MRI brain:.....
599 ☐ other:.....

Clinical differential diagnosis

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