Radboudumc

REQUEST FORM
Biochemistry & Genetics Mitochondrial Diagnostics

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Investigations are performed by the Clinical Genetics Centre Nijmegen.

Patient

Family name: _____

First name: _____

Middle name: _____

Date of Birth: DD / MM / YY Deceased : ☐ Yes, date _____

Gender: M / F

MRN: _____

Address: _____

ZIP code and City: _____

Referring physician

Name: _____ Phone: _____

Hospital: _____ Fax: _____

Specialty: _____ Email: _____

Department : _____ CC result to: _____

Address: _____

Billing Address: _____

Requested investigations

Please provide clinical data (see next page)

☐ **Complete biochemical and genetic work-up** ☐ **Biochemical diagnostics** ☐ **Genetic diagnostics**

☐ **Special request:** _____

Specimen

☐ **Muscle** Date collected _____ Time collected _____

☐ **Skin/fibroblasts** Date collected _____

☐ **Blood** Date collected _____

☐ **Other**: _____ Date collected _____ Time collected _____

(For muscle bopsies): anesthetics ☐ local ☐ general

Biopsy taken from: *Musculus* _____

To be filled out by lab employee:

Date received: _____
Time of receipt: _____
Remarks:

Medication

☐ Antibiotics: _____

☐ Anticonvulsive drugs: _____

☐ Other: _____

Date received:
Time:

Initials employee:

Medical information (essential for optimal diagnostics)

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 / residual category

263 ☐ failure to thrive
118 ☐ SIDS / 117 ☐ near SIDS
120 ☐ respiratory insufficiency
243 ☐ abnormal breathing
140 ☐ lipomas
161 ☐ dysmorphic features:
102 ☐ splenomegaly
103 ☐ premature
111 ☐ hair abnormalities
116 ☐ strange odour*
124 ☐ skin abnormalities
106 ☐ hydrops fetalis
170 ☐ coarse face
172 ☐ macroglossia
173 ☐ pain in the extremities
174 ☐ gingiva hyperplasia
176 ☐ angiokeratomas
199 ☐ other:

Central nervous system

200 ☐ intellectual disability
281 ☐ congenital / 282 ☐ decline
225 ☐ dementia
220 ☐ microcephaly
221 ☐ macrocephaly
280 ☐ disturbed awareness / 216 ☐ coma
217 ☐ lethargy
210 ☐ epilepsy / epileptiform EEG
223 ☐ behavioral abnormalities / 267 ☐ autism
269 ☐ automutilation
219 ☐ unusual crying
201 ☐ motor retardation
283 ☐ congenital 284 ☐ decline
251 ☐ hypertonia / 206 ☐ spasticity
252 ☐ hypotonia
242 ☐ extrapyramidal signs
244 ☐ dystonia
214 ☐ ataxia
291 ☐ myoclonus
299 ☐ other:
202 ☐ regression in development
264 ☐ developmental delay
270 ☐ leukodystrophy
271 ☐ cerebellar atrophy
272 ☐ spinal muscular atrophy
115 ☐ speech disorder
241 ☐ pyramidal signs
235 ☐ hemiparesis
232 ☐ stroke-like episodes
292 ☐ migraine
293 ☐ non-migraineous headache
299 ☐ other:

Muscle + peripheral nervous system

294 ☐ myopathy (excl. 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 ☐ hearing loss / deafness
906 ☐ ptosis
940 ☐ ophthalmoplegia
904 ☐ strabismus
903 ☐ nystagmus
901 ☐ cataract
902 ☐ corneal disturbances
908 ☐ retinal abnormalities
900 ☐ retinitis pigmentosa
905 ☐ lens luxation
907 ☐ cherry red spot
930 ☐ (vertical) supranuclear gaze palsy
999 ☐ other:

Heart and circulatory system

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

Digestive system and liver

302 ☐ feeding problems
301 ☐ diarrhea
331 ☐ cachexia
300 ☐ vomiting
320 ☐ pseudo-obstruction / 321 ☐ ileus
101 ☐ hepatomegaly
108 ☐ icterus
308 ☐ short bowel
310 ☐ protein-losing enteropathy
399 ☐ other:

Kidneys

403 ☐ renal insufficiency
406 ☐ tubulopathy
401 ☐ polyuria
400 ☐ kidney stones
402 ☐ strange odour / color urine *
499 ☐ other:

Blood and immune system

603 ☐ anemia
607 ☐ leucopenia
606 ☐ thrombocytopenia
102 ☐ splenomegaly
601 ☐ immunodeficiency
600 ☐ recurrent infections
122 ☐ thrombosis
699 ☐ other:

Genetics

800 ☐ consanguinity
802 ☐ sib from SIDS
804 ☐ abortion
820 ☐ positive family anamnesis
821 ☐ similar phenotype
822 ☐ different phenotype:
details family member (s) in the case material has been sent to us before:
name/dob:
family relation to patient:

Laboratory research

760 ☐ diabetes mellitus
720 ☐ hypoglycemia
722 ☐ lactic acidemia
723 ☐ acidosis
721 ☐ ketosis
726 ☐ increased CK
725 ☐ increased ASAT / ALAT
724 ☐ increased ammonia
750 ☐ increased alanine
751 ☐ increased lactate / pyruvate ratio
731 ☐ abnormal vitamin status*
732 ☐ cholesterol / triglycerides status abnormal *
733 ☐ hormones are abnormal
740 ☐ lymphocytes with vacuoles
741 ☐ foam cells in the bone marrow
717 ☐ increased urine mucopolysaccharides
752 ☐ abnormal urine organic acids*
799 ☐ other:

Histological examination muscle biopsy

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

Imaging investigations

501 ☐ skeletal abnormalities
204 ☐ CT / MRI brain
510 ☐ dysostosis multiplex
599 ☐ other:

* Please specify below

Further specification of clinical data, clinical differential

Instructions

Mitochondrial diagnostic tests can be performed on a variety of samples: biopsies from muscle, liver, heart, as well as on cultured fibroblasts from a skin biopsy. Coenzyme Q10 can also be examined in blood samples. Prenatal diagnostic enzyme testing is possible after consultation of our laboratory. Mitochondrial molecular genetic tests can be performed on the same tissue samples, but also on blood and urine. The decision on which sample to send in depends on the clinical phenotype of the patient. For an advice about which type of sample to send in, please contact us.
Conditions of shipment: Please check www.rcmm.info or www.raboudumc.nl/labgk Protocols can also be requested by phone at +31 (0) 24 3614567