

REQUEST FORM Biochemistry & Genetics Mitochondrial Diagnostics

Radboudumc

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Translational Metabolic Laboratory

Patient

Family name: _____

First name: _____

Middle name: _____

Date of Birth: DD / MM / YYYY Patient deceased: Yes, date _____

Gender: M / F

Your reference (MRN): _____

Address: _____

ZIP code and City: _____

Referring physician

Name: _____ Phone: _____

Hospital: _____ Fax: _____

Specialty: _____ Email: _____

Department: _____ CC result to: _____

Address: _____

Billing address: _____

Requested investigations

Attention! Please provide clinical data on page 2

Complete biochemical and genetic work-up Biochemical diagnostics Genetic diagnostics

Special request: _____

Specimen	Date collected	Time collected	Protocol after collection
<input type="checkbox"/> Muscle	___/___/___	__:__ hour	Protocol for sending a fresh muscle biopsy / Protocol for sending a frozen muscle biopsy
<input type="checkbox"/> Skin / fibroblasts	___/___/___	__:__ hour	Protocol for sending skin biopsy samples
<input type="checkbox"/> (Morning) Urine (portion 50-100 ml) (for children: collected urine is sufficient) For mtDNA diagnostics	___/___/___	__:__ hour	Cooled (non frozen) urine to be send not on Friday or in weekend; do NOT freeze
<input type="checkbox"/> Heparin blood (10 ml) For CoQ10	___/___/___	__:__ hour	Whole blood to be send at room temperature on collection day not on Friday or in weekend; do NOT centrifuge, do no NOT freeze
<input type="checkbox"/> 2 x EDTA-blood (3-6 ml plastic tube (purple cap)) For DNA diagnostics	___/___/___	__:__ hour	Whole blood to be send at room temperature; do NOT centrifuge, do NOT freeze
<input type="checkbox"/> Other: _____ _____	___/___/___	__:__ hour	

Anesthetics for muscle biopsies: Local General

To be filled out by lab employee:

Date received: _____

Reception time: _____

Signature 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 strange crying
201 motoric 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 cerebellum 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 cornea disturbance
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 case material has been sent to us before:
name/dob.date: _____
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 mucopolysaccharides increased in the urine
752 abnormal urine organic acids
799 other: _____

Histological examination muscle biopsy

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

Visual art investigation

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

* Please specify below

Further specification of clinical data, clinical differential diagnosis

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 heparin whole blood samples (10 ml). 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.radboudumc.nl/labgk. Protocols can also be requested by email: secretariaat-tml.labgk@radboudumc.nl.