

REQUEST FORM Metabolite diagnostics

Radboudumc

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www.radboudumc.nl/laboratoriumvoordagnostiek



Diagnostics is performed by the
Clinical Genetics Center Nijmegen

Patient

Family name: _____
First name: _____
Middle name: _____
Date of Birth: DD / MM / YY Patient deceased : Yes, date _____
Gender: M / F
Your Reference: _____
Address: _____
ZIP code and City: _____

Referring physician

Name: _____ Phone: _____
Hospital: _____ Fax: _____
Specialty: _____ Email: _____
Department : _____ CC result to: Referring physician
Address: _____ Other: _____
Billing address: _____

Requested Investigations

Attention: Please fill out clinical data at page 2.

- Metabolic screening (metabolite diagnostics)**
Reports with results generally within ~3-4 weeks upon receipt, depending on number of tests.
- Control of treatment** (please specify which IEM): _____
- Special request** (please specify which test): _____
- Psychiatric disorders (DNA and metabolite diagnostics); see page 2 "Instructions" (code 1082)**
- Patient does not give permission for long term storage for additional diagnostics or anonymous research of this body material at a later stage (code 1010)

Specimen

For metabolite diagnostics always send urine and heparin plasma.

- Heparin plasma*** Date _____ Time _____ Crisis
- Urine*** Date _____ Collected during _____ hours Quantity _____ ml Crisis
- Liquor/CSF (frozen)** Date _____ Time _____
- Other** (please specify): _____ Date _____ Time _____

* For Porphyria Diagnostics; shield specimens from light by aluminium foil.

Nutritional status

- Fasting for _____ hours
- Parenteral nutrition
- Protein intake _____ g/kg body weight
- Special diet, please specify: _____

Medication

- Antibiotics, please specify: _____
- Anticonvulsiva, please specify: _____
- Other, please specify: _____

As nutrition and medication can influence the results: please specify.

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 autotumultation
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: _____

Clinical differential diagnosis:

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

Specification of clinical data

Instructions

For enzyme and/or DNA diagnostics, please use their specific request forms, which can be downloaded from www.radboudumc.nl/labgk and www.dnadiagnostieknijmegen.nl

- In case of metabolite screening, please send an urine sample of 10-20 ml and 1-2 ml heparin plasma (further info: tel +31 (0)24 3614567).
- Generally, a portion of urine is sufficient (not 24h urine!) Urine should be frozen to prevent bacterial overgrowth.
- Samples drawn/collected in a clinical or metabolic crises are preferred.
- Please send urine, plasma and liquor samples on dry ice (by courier); if necessary, freeze at -20° C until shipment.
- For psychiatric disorders:
 - 2 x 10 ml EDTA whole blood (purple cap)
 - 2 ml heparin plasma
 - 10 ml urine

To be filled out by lab employee:

Date received: _____

Reception time: _____

Remarks: _____