

REQUEST FORM Metabolite diagnostics

Radboudumc

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www.radboudumc.nl/laboratoriumvoordiagnostiek

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: _____
 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

- 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)
Some investigations within this diagnostics are provided by our collaborating laboratories

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 Porphyrria 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: _____

As nutrition and medication can influence the results: please specify

Medication

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

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 80
 Weight: kg P 3 10 50 80
 Weight to height: P 3 10 50 80
 Head circumf: cm P 3 10 50 80

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: _____

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 at the webpage of www.radboudumc.nl/labgk and www.radboudumc.nl/genoomdiagnostiek.

- In case of metabolite screening, please send an urine sample of 10-20 ml and 1-2 ml heparin plasma (further info: phone +31 (0)24 3614567 or email: secretariaat-tml.labgk@radboudumc.nl).
- 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 EDTA-blood (3-6 ml plastic tube (purple cap)) – do not centrifuge
 - 2 ml heparin plasma
 - 10 ml urine