

REQUEST FORM

Diagnostics of complement mediated disorders (CMD)

Radboudumc

Laboratory for Diagnostics
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The Netherlands

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www.radboudumc.nl/laboratoriumvoordagnostiek



Investigations are conducted by
the Clinical Genetics Centre
Nijmegen.

Patient

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

Referring physician

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

Indications

- | | | |
|---|--|---|
| <input type="checkbox"/> (suspected) aHUS | <input type="checkbox"/> (suspected) <i>S. pneumoniae</i> HUS | <input type="checkbox"/> Basal Laminar Drusen (BLD) |
| <input type="checkbox"/> aHUS patient with eculizumab | <input type="checkbox"/> (suspected) C3 glomerulopathy | <input type="checkbox"/> Age-related Macular Degeneration (AMD) |
| <input type="checkbox"/> (suspected) STEC-HUS | <input type="checkbox"/> Paroxysmal Nocturnal Hemoglobinuria (PNH) | <input type="checkbox"/> Other: _____ |

Patient does not give permission for long-term storage of this sample material for any additional diagnostic or research use at a later date (code 1010)

Requested Investigations

For blood collection protocols and advice per indication: see the back side

DNA level: Department of Genetics

- Basic gene panel kidney diseases**
- CFH
 - CFI
 - CFB
 - C3
 - CD46 (MCP)
 - MLPA CFH/CFHRs

NGS (ORF) Complement and coagulation genes (WES)

EDTA blood for all panels

- Additional gene panel kidney diseases**
- CFH related proteins (CFHR)
 - CFHR1
 - CFHR2
 - CFHR3
 - CFHR4
 - CFHR5
 - Thrombomodulin (THBD)
 - DGKE

Other genes:

- ADAMTS13
- Vitronectin
- C5
- CD55 (DAF)
- CD59
- PLG
- CFP (properdin)
- MIMACHC

Basic gene panel eye diseases

- CFH
- CFI
- CFB
- C3

Protein level: Department of TML

Complement proteins

- EDTA plasma:
- C5
 - Factor B
 - Factor H
 - Factor I

Activation markers

- EDTA plasma:
- C3bBbP
 - C3bc
 - Bb
 - C4d
 - sC5b-9 (TCC)

(Auto) antibodies

- Serum:
- Antibodies LPS O157/O26/O55/O103
 - Auto-antibodies Factor H
 - C3 nephritic factor
 - C4 nephritic factor

Functional assays

- Serum:
- Factor H function

Eculizumab*

- Serum:
- Eculizumab (Soliris) level**
 - C5-eculizumab complex
 - Eculizumab inhibiting capacity

* C5-eculizumab complex and inhibiting capacity will be determined in case of eculizumab level >8 and < 100 ug/ml or after consultation with Laboratory Specialist
 Clinical Genetics +31(0)24 3614567

** In collaboration with Department of Clinical Pharmacy

Blood collection

Level	Tube type	Minimal volume	Protocol after collection
DNA	EDTA tube for whole blood (purple) or already isolated DNA	5ml (>20 µg)	Not centrifuged, please ship at room temperature .
Protein	Serum tube (yellow or red)	3ml	Allow to clot at room temperature max. 45 min, immediately centrifuge (2000xg, 10 min, 4 °C) and freeze serum at -80 °C. Ship on dry ice.
Protein	EDTA tube for EDTA-plasma (purple)	3ml	Place tube directly on crushed ice , centrifuge (2000xg, 10 min, 4 °C) within 60 min after collection and freeze EDTA plasma at -80 °C. Ship on dry ice.

Advice per indication

Indication

(suspected) aHUS

(6003)

aHUS patient with eculizumab

(6008)

(suspected) C3 glomerulopathy

(6005)

(suspected) STEC-HUS

(6004)

(suspected) *S.pneumoniae* HUS

(6006)

Paroxysmal Nocturnal Hemoglobinuria (PNH)

(6007)

Basal Laminal Drusen (BLD)

Age-related Macular Degeneration (AMD)

Other indications

(6999)

Advice

Basic gene panel kidney diseases, Additional gene panel kidney diseases, Auto-antibodies Factor H, Antibodies LPS O157/O26/O55/O103, TCC, Bb, C3bc, C3bBbP, Factor H, Factor I, Factor B

Eculizumab (Soliris) level, C5, TCC

Basic gene panel kidney diseases, Additional gene panel kidney diseases, Auto-antibodies Factor H, C3 nephritic factor, C4 nephritic factor, TCC, Bb, C3bc, C3bBbP, Factor H, Factor I, Factor B

Basic gene panel kidney diseases, Additional gene panel kidney diseases, Auto-antibodies Factor H, Antibodies LPS O157/O26/O55/O103, TCC, Bb, C3bc, C3bBbP, Factor H, Factor I, Factor B

Basic gene panel kidney diseases, Additional gene panel kidney diseases, Auto-antibodies Factor H, Antibodies LPS O157/O26/O55/O103, TCC, Bb, C3bc, C3bBbP, Factor H, Factor I, Factor B

Eculizumab (Soliris) level, C5, TCC, Bb, C3bc, C3bBbP

Basic gene panel eye diseases, TCC, Bb, C3bc, C3bBbP, Factor H, Factor I, Factor B

Basic gene panel eye diseases, TCC, Bb, C3bc, C3bBbP, Factor H, Factor I, Factor B

Consultation with Laboratory Specialist Clinical Genetics (+31 (0)24-3614567)

Specification of other clinical data

Instructions

- Send request and samples to Radboudumc, Laboratory for Diagnostics, PO Box 9101, Internal Post 815, 6500 HB Nijmegen, The Netherlands
- For urgent request please contact Laboratory Specialist Clinical Genetics (+31 (0)24-3614567)