

REQUEST FORM

Diagnostics of complement mediated disorders (CMD)

Radboudumc

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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

- ☐ (suspected) aHUS ☐ (suspected) *S. pneumoniae* HUS ☐ Basal laminar drusen (BLD)
☐ aHUS patient with eculizumab ☐ (suspected) C3 glomerulopathy ☐ Age-related macular degeneration (AMD)
☐ (suspected) STEC-HUS ☐ Paroxysmal nocturnal hemoglobinuria (PNH) ☐ Other: _____

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

Requested Investigations

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

DNA level:

EDTA blood for all panels

- | | | | |
|---|--|--|--|
| <input type="checkbox"/> Kidney diseases: basic gene panel | <input type="checkbox"/> Kidney diseases: additional gene panel | <input type="checkbox"/> Other genes: | <input type="checkbox"/> Eye diseases: basic gene panel |
| <input type="checkbox"/> CFH | <input type="checkbox"/> CFH related proteins (CFHR) | <input type="checkbox"/> ADAMTS13 | <input type="checkbox"/> CFH |
| <input type="checkbox"/> CFI | <input type="checkbox"/> CFHR1 | <input type="checkbox"/> Vitronectin | <input type="checkbox"/> CFI |
| <input type="checkbox"/> CFB | <input type="checkbox"/> CFHR2 | <input type="checkbox"/> C5 | <input type="checkbox"/> CFB |
| <input type="checkbox"/> C3 | <input type="checkbox"/> CFHR3 | <input type="checkbox"/> CD55 (DAF) | <input type="checkbox"/> C3 |
| <input type="checkbox"/> CD46 (MCP) | <input type="checkbox"/> CFHR4 | <input type="checkbox"/> CD59 | |
| <input type="checkbox"/> MLPA CFH/CFHRs | <input type="checkbox"/> CFHR5 | <input type="checkbox"/> PLG | |
| | <input type="checkbox"/> Thrombomodulin (THBD) | <input type="checkbox"/> CFP | |
| | <input type="checkbox"/> DGKE | | |

☐ NGS (ORF) Complement and coagulation genes (WES)

Protein level:

Activation markers

EDTA plasma:

- ☐ C3bBbP
☐ C3bc
☐ sC5b-9 (TCC)
☐ C4d

Eculizumab*

Serum:

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

(Auto) antibodies

Serum:

- ☐ Antibodies LPS O157
☐ Auto-antibodies Factor H
☐ C3 nephritic factor

Complement factors

EDTA plasma:

- ☐ factor H

* C5-eculizumab complex and inhibiting capacity will be determined if eculizumab level >8 and < 100 ug/ml or after consultation with clinical geneticist +31(0)24 3614567

Blood collection protocols			
level	Collect	Minimal capacity	Protocol after collection
DNA	EDTA tube for whole blood (purple) or already isolated DNA	5ml (>20 µg)	Not centrifuged, please ship at <u>room temperature.</u>
Protein	Clotted tube for serum (yellow or red)	3ml	Max. allow 45 min at room temperature to clot, centrifuge, (2000xg, 10 min, 4 °C) and freeze serum at -80 °C. Ship on dry ice.
Protein	EDTA tube for EDTA-plasma (purple)	3ml	After collection <u>directly on crushed ice</u> , centrifuge (2000xg, 10 min, 4 °C) and freeze EDTA plasma at -80 °C. Ship on dry ice.

Advice per indication	
Indication	Advice
(suspected) aHUS (6003)	DNA Kidney diseases: Basic gene panel and possibly DNA kidney diseases: additional gene panel Auto-Antibodies against Complement Factor H,TCC, C3bc, C3bBbP
aHUS patient with eculizumab (6008)	Ecuzumab (soliris) level, TCC
(suspected) C3 glomerulopathy (6005)	DNA Kidney diseases: Basic gene panel and CFHR5 Auto-Antibodies against Complement Factor H, C3 nephritic factor, TCC, C3bc, C3bBbP
(suspected) STEC-HUS (6004)	DNA Kidney diseases: Basic gene panel Antibodies against LPS O157, Auto-Antibodies against complement Factor H
(suspected) <i>S.pneumoniae</i> HUS (6006)	DNA Kidney diseases: Basic gene panel Antibodies against LPS O157, Auto-antibodies against complement Factor H
Paroxysmal nocturnal heamoglobinuria (PNH) (6007)	Ecuzumab (soliris) level
Basal laminar drusen (BLD)	DNA Eye diseases: Basic gene panel
Age-related macular degeneration (AMD)	DNA Eye diseases: Basic gene panel
Other indications (6999)	Consultation with clinical geneticist (+31 (0)24-3614567)

Specification of other clinical data

- | Instructions |
|--|
| <ul style="list-style-type: none"> For general terms and conditions see application form LMI: general complement provisions Send request and samples to Radboudumc, Laboratory Medicine, 830 TML, M325.04.268 Reception, Geert Grooteplein Zuid 30, 6525 GA Nijmegen, The Netherlands For urgent request please contact clinical genetisist (+31 (0)24-3614567) |