

**Uromodulin nephropathy**  
**Familial juvenile hyperuricemic nephropathy (FJHN)**  
**Medullary cystic kidney disease (MCKD2)**  
OMIM 162000; 603860

**Gene: UMOD**

**Locus: 16p12.3**

**OMIM: 191845**

**SERVICE:** mutation analysis of exons 4, 5 & 6 of UMOD gene

**TESTING:** Diagnostic: clinically affected patients  
Presymptomatic: patients at risk of developing FJHN/MCKD2 (known UMOD mutation)

**REFERRALS:** from Consultant Clinical Geneticists, Rheumatological and Renal Physicians only

**TARGET REPORTING TIME AND COSTS**

(Non NHS patients are subject to 20% surcharge and payment must be agreed prior to testing)

Diagnostic:	8 weeks	£317
Presymptomatic:	2 weeks	£195 (sequence one exon)

**TECHNICAL INFORMATION**

- PCR and fluorescent sequence analysis of exons 4, 5 & 6 and splice site boundaries of the UMOD gene

Point mutations or small insertions/deletions in these exons are responsible for all reported cases.

**SAMPLE REQUIREMENTS**

- 5ml blood in EDTA or 50ul DNA
- All patient samples must be labelled with **name, date of birth and Hospital/NHS number**

Samples should be accompanied by a FULLY completed request card (available from the laboratory). Please include details of test, family history, patient address & postcode, GP, referring clinician and unit/hospital.

**CONSENT**

It is the responsibility of the referring clinician to ensure consent has been obtained for:

- testing and storage
- the use of the sample and the information generated from it to be shared with members of the patients family and their health professionals

After testing, part of this sample might be used anonymously for the development of new tests and to monitor the quality of laboratory results.

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**Genetics Laboratories**  
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## UKGTN 'Testing criteria' template

**Patient name:**

**Patient address:**

**Name of referrer:**

**Title/Position:**

**Department/Hospital:**

**Name of Disease/test:**

**Uromodulin Nephropathy (UMOD)**

**Familial Juvenile Hyperuricemic nephropathy (FJHN)**

**Medullary Cystic Kidney Disease Type 2 (MCKD2)**

**Referrals only will be accepted from one of the following:**

(Please indicate with a tick which category refers to the referrer).

<b>Referrer</b>	<b>Tick if this refers to you.</b>
Consultant Physician	
Consultant Clinical Geneticist	
Consultant Renal Physician	
Consultant Rheumatological physician	

**Minimum criteria required for testing to be appropriate as stated in the Gene Dossier:**

<b>Criteria</b>	<b>Tick if this patient meets criteria</b>
a) low urate excretion or	
b) hyperuricemia and/or	
c) medullary cysts and/or	
d) renal impairment and/or	
e) gout	
or: b) and c) alone	
or: c) and d) alone	
or: c) and e) alone	

**If the sample does not fulfil these criteria and you still feel that testing should be performed please contact the molecular genetics laboratory**