

Renal tubular acidosis, distal, autosomal dominant **OMIM: 179800**

Gene: SLC4A1

Locus: 17q21-q22

OMIM: 109270

SERVICE: **mutation analysis of exons 14, 15 & 20 of SLC4A1 gene**

TESTING: **Diagnostic: clinically affected patients**
Presymptomatic: patients at risk of developing dRTA (known mutation)
*samples will only be accepted with a completed 'testing criteria' form (see attached)

REFERRALS: **from Clinical Geneticists and Renal Physicians only**
The laboratory does NOT accept referrals directly from patients

TARGET REPORTING TIME AND COSTS

(Non UK National Health Service patients are subject to a surcharge. Payment must be agreed prior to testing – please include invoice form A)

Diagnostic:	8 weeks	£175
Presymptomatic:	2 weeks	£145 (sequence one exon)

TECHNICAL INFORMATION

- PCR and fluorescent sequence analysis of exons 14, 15 & 20 and splice site boundaries of the SLC4A1 gene
Point mutations or small insertions/deletions in these exons are responsible for all reported cases.

SAMPLE REQUIREMENTS

- 1-5ml blood in EDTA or 50ul DNA (concentration ~500ng/ul)
- 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
- **Samples and paperwork must include three unique and matching patient identifiers**

SHIPPING DETAILS

- DNA can be sent by first class post
- Blood must be appropriately packaged and preferably sent by courier to arrive as soon as possible.
- Do not freeze prior or during postage.

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.

CONTACT DETAILS

Genetics Laboratories, Box 143
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Cambridge CB2 0QQ
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Website: www.cuh.org.uk/genetics-labs



Accredited Medical Laboratory
Reference No: 1275

UKGTN testing criteria

Name of Disease: **Distal Renal Tubular Acidosis**

Name of Gene: **SLC4A1**

Patient name:

Date of birth:

Patient postcode:

NHS number:

Name of referrer:

Title/Position:

Department/Hospital:

Contact email/telephone number:

Referrals will only be accepted from one of the following:
(Please indicate with a tick which category refers to the referrer).

Referrer	Tick if this refers to you
Consultant Renal Physician	
Consultant Clinical Geneticist	

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

Criteria	Tick if this patient meets criteria
1. Inappropriately alkaline urine in the context of metabolic acidosis, <u>OR</u> 2. failure of acid challenge test AND	
3. evidence of renal tract calcification	

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