

Renal Tubular Acidosis, distal, autosomal dominant (dRTA)

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 SLC4A1 mutation)

*samples will only be accepted with a completed 'testing criteria' form (see attached)

REFERRALS: from Consultant Clinical Geneticists and Consultant 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 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

- 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.

**East Anglian Medical Genetics Service
Genetics Laboratories
Molecular Genetics, Box 158
Level 6, Addenbrooke's Treatment Centre
Addenbrooke's Hospital
Cambridge CB2 0QQ**

Tel: 01223 348866

Fax: 01223 348870

Email: becky.treacy@addenbrookes.nhs.uk

UKGTN 'Testing criteria' template

Patient name:

Patient address:

Name of referrer:

Title/Position:

Department/Hospital:

Name of Disease/test:

Distal Renal Tubular Acidosis

Referrals only will 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
1a) Inappropriately alkaline urine in the context of metabolic acidosis or 1b) failure of acid challenge test	
and 2) 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