

Liddle Syndrome (Pseudoaldosteronism)

OMIM 177200

Gene: SCNN1B; SCNN1G

Locus: 16p13-p12

OMIM: 600760; 600761

SERVICE: mutation analysis of exon 12 in both the SCNN1B and SCNN1G genes

TESTING: Diagnostic*: clinically affected patients
Presymptomatic: patients at risk of developing Liddle syndrome
(known SCNN1B or SCNN1G mutation)

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

REFERRALS: from Consultant Clinical Geneticists, Renal Physicians, Pharmacologists and Endocrinologists

TARGET REPORTING TIME AND COSTS

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

Diagnostic:	8 weeks	£267
Presymptomatic:	2 weeks	£195 (sequence one exon)

TECHNICAL INFORMATION

- PCR and fluorescent sequence analysis of exon 12 and splice site boundaries in both the SCNN1B and SCNN1G genes

Point mutations in this exon in either of these genes 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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UKGTN 'Testing criteria' template

Patient name:

Patient address:

Name of referrer:

Title/Position:

Department/Hospital:

Name of Disease/test:

Liddle syndrome (Pseudoaldosteronism)

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 Endocrinologist	
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. Biochemical evidence of hypokalaemic alkalosis with low renin and aldosterone	
2. Hypertension	

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