

Glucocorticoid-remediable aldosteronism (GRA)

OMIM 103900

Gene: CYP11B2/CYP11B2

Locus: 8q21

OMIM: 610613/124080

SERVICE: unequal crossover of CYP11B2/CYP11B2

TESTING: Diagnostic: clinically affected patients
Presymptomatic: patients at risk of developing GRA (known familial mutation)

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

REFERRALS: from Consultant Clinical Geneticists, Renal Physicians, Pharmacologists & 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	£133
Presymptomatic:	2 weeks	£133

TECHNICAL INFORMATION

- Unequal crossover of CYP11B1/CYP11B2 upstream of exon 5

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:

Glucocorticoid remediable aldosteronism

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 Nephrologist	
Consultant Endocrinologist	
Consultant Clinical Pharmacologist	

**Minimum criteria required for testing to be appropriate as stated in the Gene Dossier:
(More than 3 required)**

Criteria	Tick if this patient meets criteria
Hypertension	
Primary hyperaldosteronism	
Dominant family history of hypertension	
Family history of CVA under 60y	
Urine 18 hydroxysteroid excess?	
Positive steroid suppression test?	

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

UKGTN 'Testing criteria' template