

**Glucocorticoid-remediable aldosteronism (GRA) OMIM: 103900**  
**Gene: CYP11B1/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 mutation)**  
\*samples will only be accepted with a completed 'testing criteria' form (see attached)

**REFERRALS:** **Clinical Geneticists, Renal Physicians, Pharmacologists & Endocrinologists**  
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)

<b>Diagnostic:</b>	<b>2 weeks</b>	<b>£99</b>
<b>Presymptomatic:</b>	<b>2 weeks</b>	<b>£145</b>

## TECHNICAL INFORMATION

- Unequal crossover of CYP11B1/CYP11B2 upstream of exon 5

## 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  
Level 6, Addenbrooke's Treatment Centre  
Addenbrooke's Hospital  
Cambridge CB2 0QQ  
Tel: +44 (0) 1223 348866  
Fax: +44 (0) 1223 348870  
Email: [becky.treacy@addenbrookes.nhs.uk](mailto:becky.treacy@addenbrookes.nhs.uk)

Website: [www.cuh.org.uk/genetics-labs](http://www.cuh.org.uk/genetics-labs)



**Accredited Medical Laboratory**  
**Reference No: 1275**

## UKGTN testing criteria

UK Genetic Testing Network

<b>Name of Disease:</b>	<b>Glucocorticoid remediable aldosteronism</b>
	<b>Familial Hyperaldosteronism Type 1  </b>
<b>Name of Gene(s):</b>	<b>CYP11B2 (cytochrome P450, family 11, subfamily B, polypeptide 2)</b>
	<b>CYP11B1 (cytochrome P450, family 11, subfamily B, polypeptide 1)</b>

<b>Patient name:</b>	<b>Date of birth:</b>
<b>Patient postcode:</b>	<b>NHS number:</b>
<b>Name of referrer:</b>	
<b>Title/Position:</b>	
<b>Department/Hospital:</b>	
<b>Contact email/telephone number:</b>	

**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.
<b>Consultant Clinical Geneticist</b>	<input type="checkbox"/>
<b>Consultant Nephrologist</b>	<input type="checkbox"/>
<b>Consultant Endocrinologist</b>	<input type="checkbox"/>
<b>Consultant Clinical Pharmacologist</b>	<input type="checkbox"/>

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

Criteria	Tick if this patient meets criteria
<b>Hypertension and primary hyperaldosteronism and &gt; 2 of the following:</b>	<input type="checkbox"/>
<b>1) Dominant family history of hypertension</b>	<input type="checkbox"/>
<b>2) Family history of CVA under 60y</b>	<input type="checkbox"/>
<b>3) Urine 18 hydroxysteroid excess</b>	<input type="checkbox"/>
<b>4) Positive steroid suppression test?</b>	<input type="checkbox"/>
<b>5) On <math>\geq 3</math> drugs for hypertension</b>	<input type="checkbox"/>
<b>6) Hypokalemia (esp. if on thiazide)</b>	<input type="checkbox"/>
<b>OR at risk relatives with a known familial mutation</b>	<input type="checkbox"/>

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