Androgen Insensitivity syndrome (AIS)
OMIM 300068

Gene: Androgen receptor (AR)  Locus: Xq11-q12  OMIM: 313700

SERVICE: mutation and dosage analysis of the androgen receptor (AR) gene

TESTING: Diagnostic*: clinically affected patients with XY karyotype
Carrier: female relatives of clinically affected patients (known AR mutation)

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

REFERRALS: from Clinical Geneticists, Paediatric/Adult Endocrinologists, Gynaecologists
The laboratory does NOT accept referrals directly from patients

TARGET REPORTING TIME AND COSTS
(Patients outside of the UK National Health Service are subject to 20% surcharge and payment must be agreed prior to testing)

<table>
<thead>
<tr>
<th>Service</th>
<th>Time</th>
<th>Cost</th>
</tr>
</thead>
<tbody>
<tr>
<td>Diagnostic: sequencing</td>
<td>8 weeks</td>
<td>£393</td>
</tr>
<tr>
<td>Dosage:</td>
<td>2 weeks</td>
<td>£157</td>
</tr>
</tbody>
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<thead>
<tr>
<th>Service</th>
<th>Time</th>
<th>Cost</th>
</tr>
</thead>
<tbody>
<tr>
<td>Carrier: sequence one exon</td>
<td>2 weeks</td>
<td>£157</td>
</tr>
</tbody>
</table>

TECHNICAL INFORMATION
- PCR and fluorescent sequence analysis of exons 1-8 and splice site boundaries of the AR gene
- Multiplex ligation dependent probe amplification analysis of exons 1-8 of the AR gene

More than 95% of patients with complete androgen insensitivity (CAIS) and approximately 20% of patients with partial androgen insensitivity (PAIS) have point mutations in the androgen receptor (AR) gene.

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
- 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.

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Genetics Laboratories
Molecular Genetics, Box 158
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

http://www.addenbrookes.org.uk/moleculargenetics
UKGTN ‘Testing criteria’ template

Patient name:
Patient address:
Name of referrer:
Title/Position:
Department/Hospital:
Contact email/telephone number:

Name of Disease/test:

Androgen Insensitivity syndrome (AIS)

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

<table>
<thead>
<tr>
<th>Referrer</th>
<th>Tick if this refers to you.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Consultant Clinical Geneticist</td>
<td></td>
</tr>
<tr>
<td>Consultant Paediatric Endocrinologist</td>
<td></td>
</tr>
<tr>
<td>Consultant Adult Endocrinologist</td>
<td></td>
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<tr>
<td>Consultant Gynaecologist</td>
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</tbody>
</table>

For information regarding AIS research studies or further interpretation of unclassified variants, please contact Professor Ieuan Hughes (Tel: 01223 336885; iah1000@medschl.cam.ac.uk)

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

<table>
<thead>
<tr>
<th>Criteria (all required)</th>
<th>Tick if this patient meets criteria</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. Normal testosterone production</td>
<td></td>
</tr>
<tr>
<td>2. Absent Mullerian structures</td>
<td></td>
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<tr>
<td>3. XY karyotype</td>
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</table>

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