

Gonadal dysgenesis OMIM: 400044

Gene: SRY

Locus: Yp11.3

OMIM: 480000

SERVICE: mutation analysis of the SRY (sex determining region Y) gene

TESTING: **Diagnostic*:** clinically affected patients with XY karyotype
Family members: relatives of affected patients (with known SRY mutation)
*samples will only be accepted with a completed 'testing criteria' form (see attached)

REFERRALS: **Clinical Geneticists, Paediatric/Adult Endocrinologists, Gynaecologists**
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)

Diagnostic:	8 weeks	£145	(sequence one exon)
		£145	(SRY/AMEL duplex PCR)
Family members:	2 weeks	£145	(sequence one exon)

TECHNICAL INFORMATION

- PCR and fluorescent sequence analysis of single exon and splice site boundaries of the SRY gene

Mutations detected in ~15% of patients with complete gonadal dysgenesis. More than 45 different small mutations (point mutations/small deletions and insertions) described, with the majority being unique to each family

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
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Addenbrooke's Hospital
Cambridge CB2 0QQ
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Website: www.cuh.org.uk/genetics-labs



Accredited Medical Laboratory
Reference No: 1275

UKGTN testing criteria

Name of disease(s):	Gonadal dysgenesis
Name of gene(s):	SRY (sex determining region Y) gene

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

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.
Consultant Clinical Geneticist	<input type="checkbox"/>
Consultant Paediatric Endocrinologist	<input type="checkbox"/>
Consultant Adult Endocrinologist	<input type="checkbox"/>
Consultant Gynaecologist	<input type="checkbox"/>

Minimum criteria required for testing to be appropriate:

Criteria (all required)	Tick if this patient meets criteria
1. 46, XY karyotype AND	<input type="checkbox"/>
2. Normal external female genitalia AND	<input type="checkbox"/>
3. Mullerian structures evident	<input type="checkbox"/>

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