

Gonadal dysgenesis

OMIM 306100

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)

REFERRALS: from Consultant Clinical Geneticists/Endocrinologists/Paediatricians only

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 (sequence one exon)
Carrier:	2 weeks	£195 (sequence one exon)

TECHNICAL INFORMATION

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

More than 45 different small mutations (point mutations/small deletions and insertions) described, with the majority being private to each family

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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