

# Androgen Insensitivity syndrome (AIS)

OMIM 300068

Gene: Androgen receptor (AR)

Locus: Xq11-q12

OMIM: 313700

**SERVICE:** mutation and dosage analysis of the AR gene

**TESTING:** Diagnostic: clinically affected patients with XY karyotype  
Carrier: female relatives of clinically affected patients (known AR mutation)  
Prenatal: at risk of having an affected child (known AR mutation)

**REFERRALS:** from Clinical Geneticists, Paediatricians, Endocrinologists, Gynaecologists

## TARGET REPORTING TIME AND COSTS

(Non NHS patients are subject to 20% surcharge and payment must be agreed prior to testing)

Diagnostic:	8 weeks	£903 (sequencing + dosage)
Carrier:	2 weeks	£195 (sequence one exon)
		£157 (dosage analysis)
Prenatal:	3 days	£555 (sequence one exon)

## 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 less than 50% of patients with partial androgen insensitivity (PAIS) have 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.

## 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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