

Insulin receptoropathies:

Donohue syndrome; Rabson Mendenhall syndrome; Type A severe insulin resistance
OMIM 246200; 262190; 610549

Gene: insulin receptor (INSR)

Locus: 19p13.2

OMIM: 147670

SERVICE: mutation analysis of the insulin receptor (INSR) gene

TESTING: Diagnostic*: clinically affected patients
Carrier: relatives of clinically affected patients (known INSR mutation)
Prenatal: at risk of having an affected child (known INSR mutations)

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

REFERRALS: from Consultant Clinical Geneticists, Paediatricians and Metabolic Medicine Consultants

TARGET REPORTING TIME AND COSTS

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

Diagnostic:	8 weeks	£1000	(sequencing)
Carrier:	2 weeks	£195	(sequence one exon)
Prenatal:	3 days	£555	

TECHNICAL INFORMATION

- PCR and fluorescent sequence analysis of exons 1-22 and splice site boundaries of the INSR 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.

**East Anglian Medical Genetics Service
Genetics Laboratories
Molecular Genetics, Box 158
Level 6, Addenbrooke's Treatment Centre
Addenbrooke's Hospital
Cambridge CB2 0QQ**

Tel: 01223 348866

Fax: 01223 348870

Email: becky.treacy@addenbrookes.nhs.uk

UKGTN 'Testing criteria' template

Patient name:

Patient address:

Name of referrer:

Title/Position:

Department/Hospital:

Name of Disease/test:

Donohue syndrome; leprechaunism

Rabson Mendenhall syndrome; severe insulin resistance

Type A insulin resistance

Referrals only will 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	
Consultant Paediatrician	
Metabolic Medicine Consultant	

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

Criteria	Tick if this patient meets criteria
Severely elevated plasma insulin	
Syndrome consistent with Donohue or Rabson Mendenhall syndromes in infancy/childhood	
Post pubertal severe insulin resistance with plasma adiponectin >5 mg/l (Cambridge assay)	

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