

Insulin receptoropathies, autosomal recessive:
Donohue syndrome; Rabson Mendenhall syndrome **OMIM: 246200/262190**
Gene: insulin receptor (INSR) **Locus: 19p13.2** **OMIM: 147670**

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

TESTING: **Diagnostic*:** **clinically affected patients**
 Carrier: **relatives of clinically affected patients (known mutation)**
 Prenatal: **at risk of having an affected child (known mutations)**
 *samples will only be accepted with a completed 'testing criteria' form (see attached)

REFERRALS: **Clinical Geneticists and Paediatricians only**
 The laboratory does **NOT** accept referrals directly from patients

TARGET REPORTING TIME AND COSTS
 (Non UK National Health Service patients are subject a surcharge. Payment must be agreed prior to testing – please include invoice form A)

Diagnostic:	8 weeks	£475 (sequencing + dosage)
Carrier:	2 weeks	£175 (sequence two exons)
Prenatal:	3 days	£350 (including maternal contamination studies)

TECHNICAL INFORMATION

- PCR and fluorescent sequence analysis of exons 1-22 and splice site boundaries of the INSR gene
- Multiplex ligation dependent probe amplification analysis of all exons
- Linkage analysis, if definite clinical diagnosis and sufficient family members available (please contact laboratory)

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
 Tel: +44 (0) 1223 348866
 Fax: +44 (0) 1223 348870
 Email: becky.treacy@addenbrookes.nhs.uk
 Website: www.cuh.org.uk/genetics-labs



Accredited Medical Laboratory
Reference No: 1275

UKGTN testing criteria

Name of Disease:	Donohue syndrome; Leprechaunism Rabson Mendenhall syndrome; Pineal hyperplasia, insulin-resistant diabetes mellitus and somatic abnormalities
Name of Gene:	INSR

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 Paediatrician	<input type="checkbox"/>

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, (typically greater than 500pmol/L in a normoglycemic individual) AND	<input type="checkbox"/>
Syndrome consistent with Donohue or Rabson Mendenhall in infancy/childhood: <ul style="list-style-type: none"> • intrauterine growth retardation/linear growth impairment • fasting or post absorptive hypoglycaemia/ post prandial hyperglycaemia • hypertrichosis • coarse facial features • enlargement of sex hormone-dependent tissues (genitalia, nipples) • lack of adipose tissue 	<input type="checkbox"/>

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