

PLEASE RETURN THIS FORM WITH EDTA BLOOD OR DNA

## Molecular Genetic Testing for Multiple Endocrine Neoplasia Types 1 and 2

Please send **EDTA** blood (20ml adults; 2.5ml children) or DNA to Dr S. Ellard, Molecular Genetics Laboratory, Royal Devon & Exeter NHS Healthcare Trust, Barrack Road, Exeter EX2 5DW

Clinical Head: Professor A.T. Hattersley (01392-403089 or A.T.Hattersley@exeter.ac.uk)  
Principal Clinical Molecular Geneticist: Dr S. Ellard (01392-402910 or S.Ellard@exeter.ac.uk)  
Please fill in as fully as possible and tick boxes where appropriate.

### Patient details

Surname .....  
First name(s) .....  
Date of birth.....  
Hospital No.....

### Requestor details

Clinician name .....  
Telephone .....  
Address for report.....  
Address for invoice.....

### Clinical information

Status:

Age at diagnosis

Affected

Asymptomatic

Multiple Endocrine Neoplasia Type 1

(*MEN1* gene testing)

Multiple Endocrine Neoplasia Types 2A, 2B and  
Familial Medullary Thyroid Carcinoma (FMTC)

(*RET* gene testing)

Hyperparathyroidism  .....  
Pituitary tumour (type)  .....  
Pancreatic tumour (type)  .....  
Other (please state)  .....

MTC  .....  
Hyperparathyroidism  .....  
Pheochromocytoma  .....  
Additional features of MEN2B  .....  
eg Mucosal neuroma, Marfanoid habitus

### Family history?

Yes

No

Please give details:

Affected Grandparent? Father's father  ..... Father's mother  .....  
Mother's father  ..... Mother's mother  .....  
Affected Parent? Father  ..... Mother  .....  
Affected Sibling(s)? .....  
Affected Children? .....  
Other affected relatives (cousins, aunts, uncles) .....  
(NB. A pedigree showing clinical details of affected family members would be very helpful).

Testing for known mutation in family member? Yes

Presymptomatic test? Yes  MEN1 mutation?  RET mutation?

Mutation details .....

Name of affected family member in whom mutation was identified .....

# ***Molecular Genetic Testing for Multiple Endocrine Neoplasia Types 1 and 2***

**Available at the Molecular Genetics Laboratory**

(CPA Accredited Laboratory)

**Royal Devon & Exeter NHS Healthcare Trust**

## ***MEN1 GENE ANALYSIS FOR MULTIPLE ENDOCRINE NEOPLASIA TYPE 1***

- Mutation analysis of the *MEN1* gene by sequencing the coding region (exons 2-10) and splice sites

Germline mutations in the *MEN1* gene are found in approximately 90% of patients with familial MEN1 and 75% of sporadic cases.

*Reporting time:* 1- 6 weeks

*Cost:*                      *Sequencing of exons 2, 3, 4, 5, 6, 7, 8, 9 and 10*                      £400  
   *Testing for known mutation in family member*                      £75

## ***RET GENE ANALYSIS FOR MULTIPLE ENDOCRINE NEOPLASIA TYPES 2A AND 2B (MEN2A AND 2B) AND FAMILIAL MEDULLARY THYROID CARCINOMA (FMTC)***

- Mutation analysis of the RET proto-oncogene by sequencing of exons 10, 11, 13, 14, 15 and 16 as appropriate.

Mutations in the *RET* proto-oncogene (exons 10 and 11) are found in >95% of MEN2A patients. Familial medullary thyroid carcinoma (FMTC) may be caused by mutations in exons 10, 11, 13 or 14. Although most MEN2B patients have the M918T mutation in exon 16, several families have been reported with an exon 15 mutation (A883F). Genetic testing of the *RET* gene is recommended in all cases of medullary thyroid carcinoma, even if they are apparently spontaneous.

*Reporting time:* 1- 6 weeks

*Cost:*                      *Sequencing of exons 10, 11, 13, 14, 15 and 16*                      £250  
   *Sequencing of exons 15 and 16 (MEN2B)*                      £100  
   *Testing for known mutation in family member*                      £75

### **CONTACT DETAILS**

For clinical enquiries please contact: Prof. A.T. Hattersley, Department of Diabetes & Vascular Medicine, School of Postgraduate Medicine & Health Sciences, Barrack Road, Exeter EX2 5AX Tel: 01392 403089. Fax: 01392 403027. Email: A.T.Hattersley@ex.ac.uk

For enquiries regarding laboratory tests please contact: Dr. Sian Ellard, Molecular Genetics Laboratory, RD&E Hospital (Wonford), Barrack Road, Exeter EX2 5DW Tel: 01392 402910 Email: S.Ellard@ex.ac.uk

