# OXFORD MEDICAL GENETICS LABORATORIES

## *GENETIC TESTING FOR INHERITED ENDOCRINE NEOPLASIA SYNDROMES AND RELATED ENDOCRINE DISORDERS*

**Patient details**

Surname…… First Name ……

Date of Birth…….. Hospital No…….

Affected □ )
or ) **Please provide clinical details of conditions / complete relevant supplementary referral form**
Asymptomatic □ ) forms available for Hyperparathyroidism/Hypercalcaemia and Hypoparathyroidism

**Test requested**: (tick as required)

Multiple Endocrine Neoplasia type 1 (*MEN1* gene) ☐

Multiple Endocrine Neoplasia type 4 (*CDKN1B* gene) ☐

Multiple Endocrine Neoplasia type 2 circle MEN2a, MEN2B, or FMTC if known (*RET* gene) ☐

Familial Isolated Pituitary Adenoma (*AIP* gene) ☐

Familial Hyperparathyroidism (FIHP) *MEN1* gene ☐ *CASR* gene ☐ *CDC73* gene ☐

Familial Hypocalciuric Hypercalcaemia FHH1 -*CASR* gene ☐ FHH2 -*GNA11 gene* ☐ FHH3 -*AP2S1* gene ☐

Neonatal Severe Hyperparathyroidism (NSHPT) (*CASR* gene) ☐

Autosomal Dominant Hypocalcaemic Hypercalciuria (ADHH) ADH1 -*CASR* gene ☐ ADH2 -*GNA11* gene ☐

Hyperparathyroidism- Jaw Tumour syndrome (HPT-JT) (*CDC73* gene) ☐

**Or** Familial Hyperparathyroidism gene panel ☐

(simultaneous analysis of *MEN1, CASR, CDC73, RET, CDKN1B(p27), CDKN2B(p15), CDKN2C(p18), CDKN1A(p21)*)

Hypoparathyroidism : *AIRE gene* ☐ *GATA3* gene ☐ *PTH* gene ☐ *GCM2* gene ☐ *CASR* gene ☐ *GNA11* gene ☐

**FAMILIAL HISTORY**: YES ☐ NO ☐

Please provide family tree and names (including maiden names) of affected relatives, if known.

Has a pathogenic variant been identified in a family member? YES ☐ NO ☐

If yes, Please provide details of mutation and the name of an affected family member in whom the mutation has been identified.

**Has consent for test been obtained**? YES ☐ NO ☐

**Has Genetic Counselling been arranged**? YES ☐ NO ☐

If yes, please provide details of Geneticist/Counsellor.
Name……..
Address ……..
Telephone…… Fax……..

**Contact details of Requesting Consultant**
Name……..
Address ……..
Telephone…… Fax……..

**Is this address also for Invoice** YES ☐ NO ☐

If No, please provide correct address.

……..

Name of Requester …… Signature…….

Please contact the laboratory for current prices.
Please send **blood** (10mls from adult or 2.5mls from child) with EDTA as the anticoagulant, or **DNA** to: Oxford Molecular Genetics Laboratory, Oxford University Hospitals, The Churchill, Headington, Oxford, OX3 7LE.

**Contact Details for Enquiries:**

**Laboratory:** Treena Cranston, Genetics Laboratories, Oxford University Hospitals, Churchill Hospital, Headington, Oxford, OX3 7LE. Tel: 01865 225594, Fax: 01865 226006.

**Genetic Counselling:** Dr Lisa Walker, Consultant Clinical Geneticist, Dept of Clinical Genetics, Churchill Hospital, Headington, Oxford, OX3 7LE. Tel: 01865 225421, Fax: 01865 226011.

**Clinical and Interpretation of Results:** Professor R V Thakker, University of Oxford, Nuffield Dept Clinical Medicine, OCDEM, Churchill Hospital, Oxford, OX3 7LD. Tel: 01865 857501, Fax: 01865 857502