

Oxford Molecular Genetics Laboratory

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MULTIPLE ENDOCRINE NEOPLASIA TYPE 1 (MEN1) – OMIM +131100 & FAMILIAL ISOLATED HYPERPARATHYROIDISM (FIHP) – OMIM +145000

INTRODUCTION

Multiple endocrine neoplasia is characterised by the occurrence of tumours involving two or more endocrine glands. MEN1 (also known as Wermer syndrome) is clinically variable, dependant on the site of tumours and their products of secretion. Primary hyperparathyroidism is the most common feature affecting >95% of patients. It is an autosomal dominant condition with age related penetrance, however by age 50yrs >95% of individuals have clinical symptoms and ~100% by 80yrs.

The condition is caused by pathogenic mutation in the *MEN1* gene (11q13). Approximately 10% of individuals have no family history of MEN1 and present as new cases due to *de novo* *MEN1* gene mutations.

Pathogenic variants in the *MEN1* gene can also cause FIHP (~20% cases). This is characterised by an autosomal dominant family history of primary hyperparathyroidism.

TESTING

Diagnostic:	Clinically affected patients
Presymptomatic:	Individuals at risk of developing MEN1/FIHP. Clinically unaffected relatives of individuals in whom a <i>MEN1</i> pathogenic mutation has been identified.
Prenatal:	Prenatal testing is not usually requested

REFERRALS

All samples MUST be accompanied by a completed pre-referral form ([click here](#))

- Clinical guidance and advice to direct appropriate gene screening is available from Professor Raj Thakker, Professor of Medicine, OCDEM, Churchill Hospital (rajesh.thakker@ndm.ox.ac.uk).
- UK diagnostic referrals are accepted from Clinical Genetics, Endocrinology and consultants from other relevant specialties.
- Presymptomatic referrals should ideally be referred through/in collaboration with Clinical Genetics departments.
- Requests for presymptomatic testing should either be discussed with the laboratory in advance or be accompanied by details regarding the known pathogenic mutation in the family.
- Prenatal requests are only accepted from Clinical Genetics and must be discussed with the laboratory and arranged in advance.

STRATEGY & TECHNICAL INFORMATION

- Mutation screening in diagnostic referrals is undertaken by fluorescent sequencing of the entire coding region and exon/intron boundaries of the *MEN1* gene (exons 2-10) and dosage analysis by multiplex ligation-dependent probe amplification (MLPA) for large scale rearrangements of the *MEN1* gene.
- When a pathogenic mutation has been identified in an individual, subsequent testing of family members (presymptomatic or diagnostic confirmation) involves testing for the familial mutation only.

TARGET REPORTING TIMES

Diagnostic test:	40 days
Presymptomatic/Familial Mutation test:	10 days

N.B. Details are correct for the date of printing only – last updated 15/07/2015