

GENETICS LABORATORY REPORT

1 { **Patient Name:** Jean HELIX
Date of Birth: 12/06/1997
Gender: Female
NHS No.: 123 456 7890

2 { Lab. No.: EX1600001
Sample Received: 01/01/2016
Sample Type: DNA extracted from peripheral blood

1 { Referred by: Consultant Diabetologist, City Hospital, London
Date of Report: 22/01/2016

GENETIC TESTING FOR MATURITY ONSET DIABETES OF THE YOUNG

Reason for Request

3 { Jean Helix was diagnosed with diabetes at the age of 16 years. GAD auto-antibodies were not detected. She was initially treated with insulin and is currently treated with a sulphonylurea. There is a family history of diabetes affecting Jean's father. Mutation analysis of the *HNF1A* and *HNF4A* genes has been requested.

Test methodology

4 { 1. Analysis of the promoters, coding regions and exon/intron boundaries of the *HNF1A* (NM_000545.6) and *HNF4A* (NM_175914.4) genes by Sanger sequencing.

5 { Result:	Heterozygous mutation identified
6 { Mutation details	Gene : <i>HNF1A</i> Location : Exon 7 DNA Description : c.1340C>T Protein Description : p.Pro447Leu (p.P447L) Consequence : Missense

Interpretation

7 { Jean Helix is heterozygous for a previously reported *HNF1A* missense mutation, p.Pro447Leu (Hansen *et al* 1997 *Diabetes* **46**: 726-730). This mutation is predicted to be pathogenic and the result confirms a diagnosis of maturity-onset diabetes of the young (MODY), subtype HNF1A. Mutation testing is recommended for other affected family members in order to confirm the aetiology of their diabetes. First-degree relatives who are not known to have diabetes may be tested initially by performing an oral glucose tolerance test (OGTT). Referral to Clinical Genetics is recommended for family members with normal OGTT who request predictive molecular genetic testing. Each of this patient's offspring will be at 50% risk of inheriting this mutation and developing diabetes.

Results are dependent on samples being correctly labelled and family relationships as indicated. **Please note that any remaining DNA will be stored in the laboratory.**

Signed:

Clinical Scientist

Signed:

Consultant Clinical Scientist