Oxford Molecular Genetics Laboratory
Genetics Laboratories, Churchill Hospital, Old Road, Headington, Oxford, OX3 7LE
www.ouh.nhs.uk/geneticslab

SURF1 DEFICIENCY
- OMIM 185620 (*SURF1)*

INTRODUCTION

SURF1 (also known as Surfeit 1) encodes the SURF1 protein which is essential for assembly and maintenance of complex IV of the mitochondrial respiratory chain (also known as cytochrome c oxidase or COX). Loss of function mutations in *SURF1* are associated with autosomal recessive SURF1 deficiency, which is the most common cause of complex IV deficient Leigh syndrome (OMIM #256000) and the most common single cause of Leigh syndrome in the UK.

This service is provided in collaboration with Dr Garry Brown, alongside our other mitochondrial disease services, and is NHS Highly Specialised Services (HSS) funded for NHS referrals from England and Scotland.

TESTING

All samples MUST be accompanied by a completed Mitochondrial proforma (click here)

Diagnostic:	Clinically affected patients
Carrier or Presymptomatic:	Relatives of clinically affected patients
Prenatal:	At risk of having an affected child

REFERRALS

- From Hospital Consultants, mainly Clinical Genetics, Neurology, Paediatrics, Metabolic Medicine
- Prenatal referrals are only accepted from Clinical Genetics and / or Prenatal Diagnosis. They must be discussed with the laboratory and arranged in advance.

STRATEGY

- Sequencing of the coding region of *SURF1*

TECHNICAL INFORMATION

- Sanger sequencing of the exons 1-9 of *SURF1*

TARGET REPORTING TIMES

High priority diagnostic tests:	14-28 calendar days
Routine diagnostic tests:	56 calendar days
Carrier/Presymptomatic tests:	14 calendar days
Prenatal testing (includes maternal contamination check):	3 calendar days

N.B. Details are correct for the date of printing only – last updated 14/07/2016