Oxford University Hospitals NHS
NHS Foundation Trust

Oxford Testing Criteria:

Hereditary forms of Phaeochromocytoma and Paraganglioma (PGL)

Gene testing available:	**OMIM number(s):**
RET, VHL, SDHB, SDHC, SDHD, SDHAF2, TMEM127, MAX SDHA, FH	(Phaeochromocytoma 171300) (Paragangliona 1 168000) (Carney Complex Type 1 – 160980) **OMIM number(s):** 164761, 608537, 185470, 602413, 602690, 613019, 613403, 154950

Patient name:	**Date of birth:**
Patient postcode:	**NHS number:**
Name of referrer: **Title/Position:**	

Referrals will only be accepted from one of the following:	
Referrer	**Tick if this refers to you.**
Consultant Clinical Geneticist	
Consultant Endocrinologist	

Criteria One of the following:	**Tick if this patient meets criteria**
PGL in neck or elsewhere	
PHEO with family history of PGL	
PHEO <45 yrs of age	
PHEO over 45yrs of age*	
PHEO with other syndromic features# e.g. MEN2, VHL – **specify**	
Malignant PHEO over 45yrs of age	
Bilateral PHEO / Multiple tumours	
Confirmation of affected status in a family with known mutation	
Predictive test for known familial mutation	

- indicates testing for genes specific to stated syndrome

*****Indicates testing for only *TMEM127* and *SDHB* as patients with a later age of diagnosis are more likely to have mutations in these genes than mutations in the other genes (Yao *et al* 2010, Jafri *et al*, 2012)

Gene Tests required	**Tick if required or specify****
ALL genes (VHL, RET (exons 10-11), SDHB, SDHC, SDHD, SDHA, SDHAF2, MAX, TMEM127) FH	
Selected genes (Please specify)	

If the sample does not fulfil the clinical criteria or you are not one of the specified types of referrer and you feel testing should be performed please contact the laboratory to discuss.
Please refer to our website for further referral information: www.ouh.nhs.uk/geneticslab

Contact Details: Oxford Genetics Laboratories, Oxford University Hospitals NHS Foundation Trust, The Churchill Hospital, Oxford, OX3 7LE.
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