Recommendations for genetic testing for paraganglioma/pheochromocytoma

Genes in the boxes are most likely to account for the clinical picture described, but clinical presentation for hereditary paraganglioma/pheochromocytoma syndromes can be highly variable; a genetics consultation is recommended.

Clinical evaluation

Directing testing for known familial variant (Familial Mutation, Targeted Sequencing)

Family history consistent with a parent-of-origin effect – predisposition to paraganglioma/pheochromocytoma only with paternal transmission

The following genes can be ordered as part of Cancer Panel, Hereditary, Sequencing and Deletion/Duplication: MAX, RET, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL

Notes:
- Gene(s) most likely to account for phenotype
- Supporting literature is limited
- Rare

References: