Paraganglioma Pheochromocytoma Genetic Testing

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

Syndromic (personal or family history of below findings)

- Neurofibromas, optic glioma, Lisch nodules, café au lait spots
- Medullary thyroid cancer, parathyroid hyperplasia/adenoma
- Renal cysts/carcinoma, hemangioblastoma, endolymphatic sac tumor
- Erythrocytosis
- Parathyroid, pituitary, and/or gastro-entero-pancreatic tumors; facial angiofibromas, collagenomas

Single paraganglioma

- NF1
- SDHB immunohistochemistry
- SDHB*, SDHC*, SDHD*, SDHA, SDHAF2
- Absent expression

Multiple paragangliomas

- SDHB*, SDHD, SDHC, MAX, RET, TMEM127, VHL
- SDHB*, SDHD, VHL
- SDHB*, TMEM127
- SDHD*, SDHC*, SDHAF2, SDHA, SDHB, TMEM127
- SDHB*, SDHD*, TMEM127, VHL

Bilateral pheochromocytoma

- VHL

Increased norepinephrine

Increased epinephrine

References:

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