

Clinical Cancer Genetics Clinical Referral Guidelines

Pediatric/Rare Diseases Center 2011 Update

Referrals to the appropriate clinics are strongly encouraged for the following indications:

Retinoblastoma

- Any pediatric patient diagnosed with unilateral, bilateral, or trilateral retinoblastoma, regardless of family history
- Personal or family history of a known RB1 mutation

Li-Fraumeni syndrome/Other Rare Tumors

- Pediatric patients with any of the following, regardless of family history:
 - Adrenal tumor
 - Choroid plexus tumor
 - Embryonal rhabdomyosarcoma (before age 6)
 - More than 1 primary tumor
 - Bilateral or multifocal tumors
 - Rhabdoid tumor
 - Colon cancer
 - A childhood tumor plus congenital anomalies
- Pediatric patients with childhood cancer/tumor AND:
 - 1st degree relative* with a childhood cancer
 - 1st or 2nd degree relative**, diagnosed <50 years of age with:
 - Brain tumor
 - Breast cancer
 - Sarcoma
 - Unusually early onset cancer
- Personal or family history of a known p53 mutation, an INI-1 mutation, or other hereditary cancer syndrome

Neurofibromatosis

- Upon request by Dr. John Slopis
- Pediatric patients with a brain tumor and café au lait spots for evaluation of NF1 and/or biallelic mismatch repair mutations
- Personal or family history of a known NF1/NF2 mutation or a SPRED1 mutation

MEN/Endo/Other

- Medullary thyroid cancer
- Parathyroid tumor
- Pheochromocytoma/paraganglioma
- CNS or retinal hemangioblastoma
- Pituitary adenoma with a family history of hyperparathyroidism, pituitary adenoma, or pancreatic tumor
- Adrenal tumor
- Personal or family history of MEN1, MEN2, VHL, Carney complex, hyperparathyroidism-jaw tumor syndrome or other endocrine tumor predisposition syndrome