Genetic Counseling for Cancer Risk – Considerations for Referral

Hereditary Breast Ovarian Cancer Syndrome (BRCA1/BRCA2)

Any personal or family history of:

Breast Cancer
- diagnosed at or before age 50
- bilateral breast cancer
- triple negative breast cancer
- male breast cancer
- diagnosed at any age if any additional family history of breast, ovarian, pancreatic or prostate cancer in a close relative

Ovarian Cancer
- diagnosed at any age

Prostate Cancer
- metastatic
- high grade, if any additional family history of breast, ovarian, pancreatic or prostate cancer in a close relative

Pancreatic Cancer
- diagnosed at any age if any additional family history of breast, ovarian, pancreatic or prostate cancer in a close relative

Any Cancer
- BRCA1 or BRCA2 mutation detected on somatic tumor sequencing

In Ashkenazi Jewish families:

Breast, Ovarian, High Grade Prostate or Pancreatic Cancer
- Personal or family history at any age

Lynch Syndrome

Any personal or family history of:

Colorectal or Endometrial Cancer
- diagnosed before age 50
- diagnosed at any age if any additional family history of at least one additional close relative with colorectal or other Lynch syndrome associated cancer[1]
- tumor has features of microsatellite instability/mismatch repair deficiency
- synchronous or metachronous Lynch syndrome-associated cancers[1] in the same individual

1. colorectal cancer, cancer of the endometrium, small bowel, stomach, ovary, pancreas, ureter and renal pelvis, biliary tract and brain (usually glioblastoma), as well as sebaceous gland adenomas and keratoachanthomas

Adenomatous Polyposis Syndromes (FAP, AFAP and MAP)

Any person with a total of 10-20 or more adenomatous polyps of the colon and/ or rectum with or without a family history of colorectal cancer

Li-Fraumeni Syndrome

Two or more close relatives with one or more of the following types of neoplasms:

- sarcoma (soft tissue or osteosarcoma)
- early onset breast cancer
- acute leukemia
- adrenocortical tumor
- brain tumor
- pediatric cancers

PTEN Hamartoma Tumor Syndrome (Cowden Syndrome)

Six or more characteristic mucocutaneous lesions (facial trichilemmomas, acral keratoses, papillomatous papules, mucosal lesions)

-or-

A strong personal and/or family history that includes two or more of the following findings:

- breast cancer
- non-medullary thyroid cancer
- endometrial carcincoma
- macrocephaly
- Lhermitte-Duclos disease (LDD)
- benign breast disease or thyroid lesions
- gastrointestinal hamartomas
- autism

Other Hereditary Cancer Syndromes

Personal/family history of rare or multiple tumors, including:

- any multiple or bilateral primary cancers in one individual
- diffuse gastric cancer
- medullary thyroid cancer
- multiple melanomas with or without multiple dysplastic nevi and/or pancreatic cancer
- multiple hamartomatous gastrointestinal polyps (juvenile, Peutz-Jeghers type)
- paraganglioma and/or pheochromocytoma
- renal carcinoma of the following types:
  - type II papillary
  - oncocytic and/or chromophobe
- sex cord-stromal tumors, including Sertoli-Leydig tumors

Reproductive Carrier Screening

Individual identified as a carrier for a single mutation in:

- ATM
- FH
- NBN