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

**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 carcinoma
- macrocephaly
- Lhermitte-Duclos disease (LDD)
- benign breast disease or thyroid lesions
- gastrointestinal hamartomas
- autism

**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

**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*

**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

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