Genetic Counseling for Cancer Risk – Indications for Referral

Hereditary Breast Ovarian Cancer Syndrome (*BRCA1*/*BRCA2*)

Any personal or family history of:

Breast Cancer
- diagnosed before age 45
- in 2 or more close relatives
- bilateral breast cancer
- male breast cancer

Breast and Ovarian Cancer
- both cancers in the same woman
- one of each cancer diagnosed in two close relatives

Ovarian Cancer
- diagnosed at any age

In Ashkenazi Jewish families:

Breast Cancer
- diagnosed before age 60
- diagnosed at any age if any additional family history of breast or ovarian cancer

Ovarian Cancer
- diagnosed at any age

Li-Fraumeni Syndrome

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

- soft tissue sarcoma
- early onset breast cancer
- acute leukemia
- osteosarcoma
- adrenocortical tumor
- brain tumor

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:

- breast cancer
- non-medullary thyroid cancer
- endometrial carcinoma
- macrocephaly
- Lhermitte-Duclos disease (LDD)
- benign breast disease or thyroid lesions
- gastrointestinal hamartomas
- mental retardation

Hereditary Non-Polyposis Colon Cancer (HNPCC)

Any family that meets the following criteria:

Amsterdam II Criteria (the 3-2-1 rule) – must meet all criteria:

- ***3*** or more relatives with an HNPCC-associated cancer[1] (one has to be a first-degree relative of the other two)
- ***2*** successive generations are affected
- ***1*** HNPCC-associated cancer diagnosed before age 50

Bethesda Criteria – only need to meet one criterion:

- colorectal cancer diagnosed before age 50
- presence of synchronous or metachronous HNPCC-associated tumors,[2] regardless of age
- colorectal cancer with MSI-high histology diagnosed before age 60
- colorectal cancer diagnosed in a patient with 1 or more first-degree relatives with an HNPCC-associated tumor, with one of the cancers being diagnosed before age 50
- colorectal cancer diagnosed in a patient with 2 or more first- or second-degree relatives with HNPCC-associated tumors, regardless of age

1. colorectal cancer, cancer of the endometrium, small bowel, ureter or renal pelvis
2. 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 or more adenomatous polyps of the colon and/ or rectum with or without a family history of colorectal cancer

Multiple Endocrine Neoplasia, Type 1 (MEN1)

Any personal or family history of parathyroid hyperplasia plus pituitary tumors and/or pancreatic tumors

Multiple Endocrine Neoplasia, Type 2 (MEN2)

Any personal or family history of medullary thyroid cancer, with or without other features of MEN2 including: parathyroid hyperplasia, pheochromocytoma, marfanoid habitus, ganglioneuromatosis and mucosal neuromas

Hereditary Melanoma

Any individual with:

- a family history of melanoma in two or more close relatives with or without a family history of pancreatic cancer
- a personal history of multiple primary melanomas
- dysplastic nevi and a family history of melanoma with or without a history of pancreatic cancer