

### Hereditary Breast and Ovarian Cancer (BRCA1, BRCA2)

Any personal or family history of:

#### **Breast Cancer**

- Diagnosed < age 40
- In > 2 close relatives < 50
- Bilateral cancer diagnosed at any age
- In a male diagnosed at any age

#### **Breast and Ovarian Cancer**

- Both cancers in one woman
- One of either cancer in two close relatives, diagnosed at any age

#### **Ovarian Cancer**

In two close relatives, diagnosed at any age

In Ashkenazi Jewish families:

#### **Breast Cancer**

- Diagnosed < age 50
- Any age if family history of breast and/or ovarian cancer

#### **Ovarian Cancer**

- Personal or family history of breast and/or ovarian cancer

### Familial Adenomatous Polyposis (FAP)

Any person presenting with polyposis and/or early onset colorectal cancer (< age 40) should be referred for genetic cancer risk assessment and possible genetic testing.

### Hereditary NonPolyposis Colon Cancer (HNPCC)

Any family that meets the following criteria should be considered an appropriate referral for diagnostic services and genetic testing:

#### **Amsterdam Criteria (3-2-1 rule)**

- 3 relatives with colorectal cancer (One is a first degree relative of the other two.)
- 2 successive generations affected
- 1 colorectal cancer diagnosed < 50

#### **Bethesda Criteria**

- Families that meet Amsterdam Criteria
- Any colorectal cancer or endometrial cancer diagnosed < age 45
- Individuals who have two HNPCC-related cancers (endometrial, ovarian, small bowel, renal pelvis, ureter, gastric, hepatobiliary, synchronous/metachronous colorectal cancers); OR
- Individuals with colorectal cancer and a first-degree relative with colorectal cancer and/or HNPCC-related extracolonic cancer and/or a colorectal adenoma. One of the cancers must have been diagnosed at age < 45 years and the adenoma at age < 40 years; OR
- Individuals with colorectal cancer or endometrial cancer diagnosed at age < 45 years; OR
- Individuals with right-sided colon cancer with an undifferentiated pattern (solid/cribiform) diagnosed at < 45 years; OR
- Individuals with signet ring cell type colorectal cancer (> 50% signet ring cells) diagnosed at < 45 years; OR
- Individuals with colorectal adenoma diagnosed at < 40 years

### Li-Fraumeni Syndrome

Any personal or family history or early onset breast cancer (diagnosed < age 35) plus:

- Bone or soft tissue sarcoma
- Brain cancer
- Leukemia
- Adrenocortical carcinoma

### Cowden Syndrome

Any personal or family history of:

- Premenopausal breast cancer or
- Follicular/papillary thyroid cancer

Plus personal or family history of:

- Other breast or thyroid disease (benign or malignant)
- Multiple hamartomatous lesions of skin, mucous membrane
- Cutaneous facial trichilemmomas
- Macrocephaly
- Uterine fibroids/carcinoma

### Multiple Endocrine Neoplasia, Type 1 (MEN1)

Any personal or family history of parathyroid hyperplasia plus a personal or family history of:

- Pituitary tumors (prolactinoma)
- Pancreatic tumors (gastrinoma, insulinoma)

### Multiple Endocrine Neoplasia, Type 2 (MEN2)

Any personal or family history of medullary thyroid cancer, any age with or without features of MEN 2:

- Parathyroid hyperplasia
- Pheochromocytoma
- Marfanoid habitus
- Ganglioneuromatosis
- Mucosal neuroma

### Hereditary Melanoma

Any individual with:

- Family history of melanoma in two or more close relatives
- Personal history of multiple primary melanomas, with or without family history
- Dysplastic nevi with personal or family history of melanomas

