Breast and/or Ovarian Genetic Assessment

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INCLUSION CRITERIA[a]

One or more of the following:

- Early-age-onset breast cancer[b]
- Two breast primaries[c] or breast and ovarian cancer[d] in a single individual
  or
  Two breast primaries[c] or breast and ovarian cancers[d] in close relative(s) from the same side of family (maternal or paternal)
- Clustering of breast cancer with male breast cancer, thyroid cancer, sarcoma, adrenocortical carcinoma, endometrial cancer, pancreatic cancer, brain tumors, dermatologic manifestations or leukemia/lymphoma on the same side of family
- Member of a family with a known mutation in a breast cancer susceptibility gene
- Populations at risk[e]
- Any male breast cancer
- Ovarian cancer:[d] One or more on same side of family[f]

→ **Referral to cancer genetics professional recommended** →

ASSESSMENT

Patient needs and concerns:

- Knowledge of genetic testing for cancer risk, including benefits, risks, and limitations
- Goals for cancer family risk/assessment

Detailed family history:

- Expanded pedigree to include first-, second-, and third- degree relatives (parents, children, siblings, aunts, uncles, grandparents, great-grandparents, nieces, nephews, grandchildren, first cousins)
- Types of cancer
- Bilaterality
- Age at diagnosis
- Medical record documentation, particularly pathology reports of primary cancers

Detailed medical and surgical history:

- Any personal cancer history
- Carcinogen exposure: History of radiation therapy
- Reproductive history
- Hormone use
- Previous breast biopsies
- Pathology verification of cancers

Focused physical exam (refer to specific syndrome):

- Breast/ovarian
- Dermatologic
- Head circumference
- Head/neck exam
- Thyroid

→ See Criteria for
[Hereditary Breast/Ovarian Syndrome (HBOC-1)]
[Li-Fraumeni Syndrome (LIFR-1)]
[Cowden Syndrome (COWD-1)]

[a] The maternal and paternal sides of the family should be considered independently for familial patterns of breast/ovarian cancer.

[b] Clinically use age ≤ 50 y because studies define early onset as ranging from ≤ 40 to ≤ 50 y. For the purposes of these guidelines, invasive and ductal carcinoma in situ breast cancers should be included.

[c] Two breast primaries including bilateral disease or cases where there are two or more clearly separate ipsilateral primary tumors.

[d] For the purposes of these guidelines, fallopian tube and primary peritoneal carcinoma should be included.

[e] For populations at risk, requirements for inclusion may be lessened (eg, women of Ashkenazi Jewish descent with breast or ovarian cancer at any age.)

[f] For example, a single ovarian cancer is sufficient if the patient is of Ashkenazi Jewish descent, or has limited family structure.

Note: All recommendations are category 2A unless otherwise indicated.
Clinical Trials: NCCN believes that the best management of any cancer patient is in a clinical trial. Participation in clinical trials is especially encouraged.

Hereditary Breast and/or Ovarian Cancer

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HBOC CRITERIA[a,b]

- **Member of family with a known BRCA1/BRCA2 mutation**

- **Personal history of breast cancer + one or more of the following:**
  - **Diagnosed age ≤ 40 y,[c] with or without family history**
  - **Diagnosed age ≤ 50 y[c] or two breast primaries,[d] with ≥ 1 close blood relative with breast cancer ≤ 50 y or ≥1 close blood relative with ovarian cancer**
  - **Diagnosed at any age, with ≥ 2 close blood relatives with ovarian cancer at any age**
  - **Diagnosed at any age with ≥ 2 close blood relatives with breast cancer, especially if ≥ 1 woman is diagnosed before age 50 y or has two breast primaries[d]**
  - **Close male blood relative with breast cancer**
  - **Personal history of ovarian cancer**
  - **If of certain ethnic descent associated with deleterious mutations (eg, founder populations of Ashkenazi Jewish, Icelandic, Swedish, Hungarian or other) or history of breast and/or ovarian cancer in close blood relative; no additional family history required**

- **Personal history of ovarian cancer + one or more of the following:**
  - **≥ 1 close blood relative with ovarian cancer**
  - **≥ 1 close female blood relative with breast cancer at age ≤ 50 y or two breast primary cancers[d]**
  - **≥ 2 close blood relatives with breast cancer**
  - **≥ 1 close male blood relative with breast cancer**
  - **If of Ashkenazi Jewish descent, no additional family history is required**

- **Personal history of male breast cancer particularly if one or more of the following is also present:[e]**
  - **≥ 1 close male blood relative with breast cancer**
  - **≥ 1 close female blood relative with breast or ovarian cancer**
  - **If of certain ethnic descent associated with deleterious mutations (eg, founder populations of Ashkenazi Jewish, Icelandic, Swedish, Hungarian or other), no additional family history is required**

- **Family history only—Close family member meeting any of the above criteria**

Criteria met → **Follow-up (see HBOC-2)**

Criteria not met → **Refer to NCCN Breast Cancer Screening and Diagnosis Guidelines**

[a] One or more of these criteria is suggestive of hereditary breast/ovarian cancer syndrome that warrants further professional evaluation.

[b] When investigating family histories for HBOC, all close relatives on the same side of the family should be included. Close relatives include first-, second-, and third-degree relatives. Other malignancies reported in some families with HBOC include prostate, pancreatic, and melanoma. The presence of these cancers may increase suspicion of HBOC.

[c] May consider age range between ≤ 40 and ≤ 50 y if clinical situation warrants.

[d] Two breast primaries including bilateral disease or cases where there are two or more clearly separate ipsilateral primary tumors.

[e] Males with limited family history may have an underestimated probability of familial mutation.

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Back to Assessment (see BR/OV-1)