

12. Genetische Beratung

Swiss guidelines for referral individuals for risk assessment, genetic counselling and testing for breast/ovarian cancer predisposition syndrome

I. Individuals with a close relative¹ with a known pathogenic variant in *BRCA1* or *BRCA2*, or in another gene conferring high risk for breast and ovarian cancer.

II. WOMEN with a personal history of BREAST CANCER and one of the following:

- Age at diagnosis <40 years;
- Triple negative (oestrogen receptor, progesterone receptor and HER2 negative) breast cancer ≤60 years;
- Age at diagnosis ≤50 years, with ≥2 close relatives¹ with breast cancer at any age or with only 1 close relative¹ with breast cancer ≤50 years;
- Bilateral breast cancer, if the first cancer was diagnosed ≤50 years;
- Bilateral breast cancer at any age, with ≥1 close relative¹ with breast cancer [if only one relative affected, then age at diagnosis ≤50 years];
- Diagnosed at any age, with ≥1 close relative¹ with ovarian² cancer at any age;
- Diagnosed at any age, with ≥2 close relatives¹ with breast cancer [particularly if ≥1 breast cancer diagnosed ≤50 years or if bilateral breast cancer];
- A close male relative¹ with breast cancer (any age);
- A personal history of ovarian² cancer;

III. Women with a personal history of OVARIAN² CANCER and one of the following:

- Non-mucinous epithelial subtypes, particularly high grade serous histology, at any age;
- A personal history of breast cancer;
- One or more close relatives¹ with ovarian² cancer (any age);
- One or more close female or male relatives¹ with breast cancer, particularly if breast cancer diagnosed ≤50 years;

IV. MEN with a personal history of BREAST CANCER:

- Particularly, if one or more close male relatives¹ with breast cancer;
- Particularly, if one or more close female relatives¹ with breast or ovarian² cancer;

V. Ashkenazi Jewish heritage:

Search for the 3 founder *BRCA1* and *BRCA2* pathogenic variants³ regardless of personal or family history;

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VI. Family history only (i.e. unaffected individuals):

One or more close relatives¹ with breast or ovarian² cancer fulfilling one of the above criteria (points II–IV).

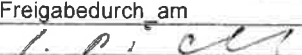
1 Close relative is defined as a first- or a second-degree relative on the same side of the family:

- first-degree relatives: mother/father, sister/brother, daughter/son;
- second-degree relatives: grandparents, aunt/uncle, niece/nephew, grandchildren.

2 Ovarian cancer also includes primary peritoneal cancer and fallopian tube cancer.

3 *BRCA1*: c.68_69delAG, c.5266dupC; *BRCA2*: c.5946delT.

Referenzdokument: Risikoabschätzung, Krebsliga Schweiz

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