Condition: Hereditary breast and ovarian cancer

Inheritance:

Autosomal Dominant**.**

Genetic etiology:

Mutation in either *BRCA1* or *BRCA2*.

Frequency:

The frequency of a *BRCA* gene mutation is about 1:400 overall; mutation frequency is increased in some specific populations due to founder effect, with limited repertoire of common mutations in these populations.

Clinical features:

Hereditary breast and ovarian cancer is associated with a markedly increased risk of breast and ovarian cancer. Male breast cancer may occur with either *BRCA1* or *BRCA2* mutation, but is much more common with the latter. Males with *BRCA1* or *BRCA2* mutation have an increased risk of prostate cancer, and BRCA2 mutation is also associated with many other tumor types, including gastrointestinal tumors and melanoma.

Management:

Screening for breast, ovarian, and prostate cancer; prophylactic surgery, including mastectomy and/or oophorectomy can reduce risk of breast and ovarian cancer in women. Chemoprevention with tamoxifen may be effective at reducing the risk of breast cancer.

Genetic counseling:

Computer programs are available to calculate the risk based on personal and family history and to guide decision-making regarding testing. *BRCA1* and *BRCA2* mutation testing is clinically available. Not all mutations can be detected, making it necessary to interpret a negative result in the absence of family history of a known mutation cautiously.

Human Genetics and Genomics, Fourth Edition. Bruce R. Korf and Mira B. Irons.