

Hereditary and familial breast cancer

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- Hereditary breast cancer (HBC) runs in families, affecting several close relatives, and is associated with an identifiable genetic mutation. Familial breast cancer (FBC) affects several members of a family but is not attributable to any known genetic mutation. HBC accounts for 5–10% and FBC for 20–30% of all breast cancers. HBCs are more aggressive, present at an earlier age and are more often multicentric and bilateral. High-penetrance mutations are found in BRCA1, - BRCA2, Li-Fraumeni syndrome, Cowden syndrome, Peutz-Jeghers syndrome and hereditary gastric cancer syndrome. BRCA1 (17q21) is associated with a 50–85% lifetime risk of developing breast cancer and up to a 40% risk of ovarian cancer. The breast cancers in BRCA1 are mostly TNBC. BRCA2 (13q12.3) is associated with an up to 50–60% lifetime risk of breast cancer and a 20% risk of ovarian cancer. It is also associated with cancer of the prostate, colon, gallbladder, bile duct, stomach and pancreas. BRCA mutation is more common in males with breast cancer. Genetic risk evaluation should be considered in high-risk individuals (Summary box 58.5). Women with a BRCA mutation may be offered a bilateral risk-reducing mastectomy with immediate breast reconstruction. This reduces the risk of breast cancer by 90%. Chemoprophylaxis with tamoxifen or anastrozole may reduce the risk to 50%. Premenopausal women may be offered bilateral salpingo-oophorectomy after they have completed their family at around 35–40 years of age.

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