BRCA1 gene

Associated Syndrome Name: *BRCA1*-associated hereditary breast and ovarian cancer syndrome

BRCA1 Summary Cancer Risk Table

CANCER	GENETIC CANCER RISK
Breast	High Risk
Ovarian	High Risk
Male Breast	Elevated Risk
Pancreatic	Elevated Risk
Prostate	Elevated Risk

BRCA1 gene Overview

BRCA1-associated hereditary breast and ovarian cancer syndrome [1, 2, 3, 4]

- Individuals with mutations in *BRCA1* have *BRCA1*-associated hereditary breast and ovarian cancer syndrome.
- Women with *BRCA1* mutations have a risk for breast cancer that is greatly increased over the 12.5% lifetime risk for women in the general population of the United States. Most breast cancers in women with *BRCA1* mutations are Triple Negative Breast Cancer (TNBC), a type of breast cancer lacking estrogen and progesterone receptors, and not expressing Her2.
- Women with *BRCA1* mutations also have high risks for ovarian, fallopian tube, and primary peritoneal cancer.
- Men with *BRCA1* mutations have an elevated risk for breast and prostate cancer. The increased risk for prostate cancer may be most significant at younger ages. Additionally, men with a *BRCA1* mutation have a higher risk for an aggressive prostate cancer.
- Male and female patients with *BRCA1* mutations have an elevated risk for exocrine pancreatic cancer. These are cancers developing in the enzyme-secreting cells of the pancreas.
- There is some evidence that individuals with mutations in *BRCA1* have an increased risk for gastric cancer. However, at this time, the evidence is not conclusive and there are no management recommendations for gastric cancer risk.
- Premenopausal individuals with *BRCA1* mutations who do not have a personal history of breast cancer or other contraindications may consider hormone replacement therapy (HRT) after a bilateral salpingo-oophorectomy (BSO).
- Based on limited data of a slightly increased risk of serous uterine cancer in individuals with *BRCA1* mutations, the risks and benefits of concurrent hysterectomy at the time of risk-reducing salpingo-oophorectomy should be discussed. Individuals who undergo hysterectomy are candidates for hormone replacement therapy (HRT) with estrogen alone, which is associated with a lower risk of breast cancer than HRT with estrogen and progesterone.
- While clinical trials regarding salpingectomy are still ongoing, salpingectomy reduces the risk of ovarian cancer in the general population and may be considered for premenopausal patients with hereditary cancer risk who are not yet ready for oophorectomy. Completion oophorectomy after salpingectomy is still recommended.
- Although there are high cancer risks for patients with *BRCA1* mutations, there are interventions that have been shown to be effective at reducing many of these risks. Guidelines from the National Comprehensive Cancer Network (NCCN) for the medical management of patients with *BRCA1* mutations are listed below. It is recommended that patients with *BRCA1* mutations be managed by a multidisciplinary team with experience in the prevention and treatment of the cancers associated with *BRCA1* mutations.

BRCA1 gene Cancer Risk Table

CANCER TYPE	AGE RANGE	CANCER RISK	RISK FOR GENERAL POPULATION
Female Breast	To age 50 [3, 5, 6, 7, 8, 9]	28%-51%, with a particularly increased risk for triple negative breast cancer (TNBC).	2.2%

CANCER TYPE	AGE RANGE	CANCER RISK	RISK FOR GENERAL POPULATION
	To age 70[3, 6, 7, 8, 9, 10]	46%-87%, with a particularly increased risk for triple negative breast cancer (TNBC).	7.6%
	Second primary within 5 years of first breast cancer diagnosis[11, 12, 13, 14]	9%-13%	1.6%
Ovarian	To age 50[5, 8, 9, 13]	8%-23%	0.2%
	To age 70[5, 7, 8, 9]	39%-63%	0.6%
	Ovarian cancer within 10 years of a breast cancer diagnosis[15, 16]	12.7%	<1.0%
Prostate	To age 70[9, 17, 18]	Up to 16%	6.3%
Male Breast	To age 70[9, 19]	1.2%	<0.1%
Pancreatic	To age 80[9, 20]	Elevated risk	1.1%

BRCA1 Cancer Risk Management Table

The overview of medical management options provided is a summary of professional society guidelines. The most recent version of each guideline should be consulted for more detailed and up-to-date information before developing a treatment plan for a particular patient.

This overview is provided for informational purposes only and does not constitute a recommendation. While the medical society guidelines summarized herein provide important and useful information, medical management decisions for any particular patient should be made in consultation between that patient and his or her healthcare provider and may differ from society guidelines based on a complete understanding of the patient's personal medical history, surgeries and other treatments.

CANCER TYPE	PROCEDURE	AGE TO BEGIN	FREQUENCY (UNLESS OTHERWISE INDICATED BY FINDINGS)
Female Breast	Breast awareness - Women should be familiar with their breasts and promptly report changes to their healthcare provider. Periodic, consistent breast self-examination (BSE) may facilitate breast awareness.[2]	Individualized	NA
	Clinical breast examination[2]	25 years	Every 6 to 12 months
	Breast MRI with and without contrast[2]	25 years, or individualized to a younger age if a relative has been diagnosed younger than age 30.	Annually
	Mammogram[2]	30 years. If MRI unavailable, start at 25 years, or individualized to a younger age if a relative has been diagnosed younger than age 30.	Annually
	Consider risk-reducing mastectomy.[2]	Individualized	NA
	Consider options for breast cancer risk-reduction agents (i.e. tamoxifen).[2]	Individualized	NA

CANCER TYPE	PROCEDURE	AGE TO BEGIN	FREQUENCY (UNLESS OTHERWISE INDICATED BY FINDINGS)
Ovarian	Bilateral salpingo-oophorectomy (BSO). Discuss the risks and benefits of concurrent hysterectomy at the time of BSO.[2]	35 to 40 years, recognizing that childbearing is a consideration	NA
	Consider options for ovarian cancer risk-reduction agents (i.e. hormonal contraceptives).[2, 21]	Individualized	NA
Prostate	Consider prostate cancer screening with digital rectal examination (DRE) and prostate specific antigen (PSA).[2]	40 years	Annually
	Consider multiparametric MRI (mpMRI) within a clinical trial setting, after discussion about benefits and limitations of such screening.[2]	Baseline at age 50, or 10 years younger than the earliest age of prostate cancer diagnosis in the family	Individualized
	Since mutation carriers are at an increased risk for more aggressive prostate cancer this information may be included as part of the risk and benefit discussion about prostate cancer screening.[22, 23]	NA	NA
	Since mutation carriers are at an increased risk for more aggressive prostate cancer this information may be considered when choosing management options for men with a diagnosis of prostate cancer.[22]	NA	NA
Male Breast	Breast self-examination[2]	35 years	Monthly
	Clinical breast examination[2]	35 years	Annually
	Consider mammogram.[2]	50 years, or 10 years earlier than the youngest male breast cancer diagnosis in the family	Annually
Pancreatic	For patients with a family history of pancreatic cancer, consider endoscopic ultrasound (EUS) and/or contrast-enhanced MRI/magnetic resonance cholangiopancreatography (MRCP). It is recommended that patients who are candidates for pancreatic cancer screening be managed by a multidisciplinary team with experience in screening for pancreatic cancer, preferably within a study setting.[2]	Age 50, or 10 years younger than the earliest age of pancreatic cancer diagnosis in the family	Annually
	Provide education about ways to reduce pancreatic cancer risk, such as not smoking and losing weight.[24]	Individualized	Individualized
For Patients With A Cancer Diagnosis	For patients with a gene mutation and a diagnosis of cancer, targeted therapies may be available as a treatment option for certain tumor types (e.g., platinum chemotherapy, PARP-inhibitors)[22, 24, 25, 26, 27, 28, 29]	NA	NA

Information for Family Members

The following information for Family Members will appear as part of the MMT for a patient found to have a mutation in the BRCA1 gene.

This patient's relatives are at risk for carrying the same mutation(s) and associated cancer risks as this patient. Cancer risks for females and males who have this/these mutation(s) are provided below.

Family members should talk to a healthcare provider about genetic testing. Close relatives such as parents, children, brothers and sisters have the highest chance of having the same mutation(s) as this patient. Other more distant relatives such as cousins, aunts, uncles, and grandparents also have a chance of carrying the same mutation(s). Testing of at-risk relatives can identify those family members with the same mutation(s) who may benefit from surveillance and early intervention.

In rare instances, an individual may inherit mutations in both copies of the *BRCA1* gene, leading to the condition Fanconi anemia, complementation group S (FANCS). This condition is rare and may include physical abnormalities, developmental delay, and a high risk for cancer. The children of this patient are at risk of inheriting FANCS only if the other parent is also a carrier of a *BRCA1* mutation. Screening the other biological parent of any children for *BRCA1* mutations may be appropriate.[2, 30]

Parents who are concerned about the possibility of passing on a *BRCA1* mutation to a future child may want to discuss options for prenatal testing and assisted reproduction techniques, such as pre-implantation genetic diagnosis (PGD).[2]

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