



Hereditary Cancer

Identifying hereditary cancer risk with genetic testing for better screening and earlier treatment and prevention

Using the Hereditary Cancer Screening Test

The AccessDx Hereditary Cancer (CGx) screening assay is aimed to detect the causative variants involved in the most common hereditary cancers. These genomic targets were chosen to cover the presence of the genetic variants base on levels of clinical evidence linked to the hereditary cancers with relatively high occurrence in the United States population. The ADX NGS-based Hereditary Cancer test is a custom designed panel that includes 33 genes known to harbor variants linked to the hereditary cancer syndrome.

Healthcare providers might consider this type of testing for individuals with any of the following characteristics:⁹

- Several first-degree family members with cancer
- Many relatives on one side of the family who had the same type of cancer
- A cluster of cancers in an individual's family which have been linked to a single gene mutation (such as some types of breast, ovarian, colorectal, and pancreatic cancers)
- Family member who had more than 1 type of cancer
- Family members who had cancer at a younger age
- Close family members who had cancers that are linked to hereditary cancer syndromes
- A family member who has a rare cancer, such as breast cancer in a man or retinoblastoma
- Certain ethnicity (for example, Ashkenazi Jewish ancestry is linked to ovarian and breast cancers)
- Physical finding which is linked to an inherited cancer (such as having many colon polyps)
- One or more family members who have already had genetic testing that found a mutation

Sequenced Genes

APC	CHEK2	NF1
ATM	COL1A1	PALB2
BAP1	EPCAM	PMS2
BARD1	FBN1	POLD1
BMPR1A	GREM1	POLE
BRCA1	MITF	PTEN
BRCA2	MLH1	RAD51C
BRIP1	MSH2	RAD51D
CDH1	MSH6	SMAD4
CDK4	MUTYH	STK11
CDKN2A	NBN	TP53

The potentially identified pathogenic variants in the genes analyzed by this panel cause variable phenotypes and cancer risks.

Hereditary Cancer Markers Greatly Increases Lifetime Cancer Risk

Hereditary Cancer	Lifetime Risk in General Population	Increased Risk Due to Genetic Mutation (as high as)
Breast	12.3% (females)	87% ¹
Ovarian	1.4%	44% ¹
Endometrial (uterine)	2.7%	71% ²
Colorectal	4.8%	99% ³
Gastric (stomach)	.9%	80% ⁴
Prostate	15.3% (males)	44% ⁵
Pancreatic	1.5%	36% ¹
Melanoma (skin)	4.8%	76% ⁶

A History of Excellence

AccessDx is CAP and CLIA certified, showcasing our commitment to accuracy, speed, and excellence. With over 2,000 clients across the healthcare landscape, our partners recognize our commitment to excellence. AccessDx's dedicated lab operations and client success teams are ready to help serve the needs of your group.

¹Cancer risk estimates for a positive result are typically based on individuals with a family or personal history of cancer. NCCN Clinical Practice Guidelines in Oncology Genetic/Familial High Risk Assessment: Breast, Ovarian and Pancreatic v1.2020

²Genetic/Familial High-Risk Assessment: Breast, Ovarian, and Pancreatic. NCCN Guidelines Version 1.2020. Available at: www.nccn.org/professionals/physician_gls/pdf/genetics_screening.pdf. Accessed January 3, 2020.

³Patel SG, Ahnen DJ, Kinney AY, et al. Am J Gastroenterol. Knowledge and uptake of genetic counseling and colonoscopic screening among individuals at increased risk for lynch syndrome and their endoscopists from the family health promotion project. 2016 Feb;111(2):285-93.

⁴Shenoy S, CDH1 (E-Cadherin) Mutation and Gastric Cancer: Genetics, Molecular Mechanisms and Guidelines for Management. Cancer Manag Res. 2019 Dec 13;11:10477-10486.

⁵NCCN Clinical Practice Guidelines in Oncology. Prostate Cancer Version 2.2021. Available at NCCN.org

⁶Tucker MA, Elder DE, Curry M, Fraser MC, Pichler V, Zamatkin D, Yang XR, Goldstein AM. Risks of Melanoma and Other Cancers in Melanoma-Prone Families over 4 Decades. J Invest Dermatol. 2018 Jul;138(7):1620-1626.