Ambry Genetics® | LifeStrands

Germline Testing for Hereditary Cancer

Reference Guide

Guidelines Recommend Genetic Testing For Hereditary Cancer

The American Society of Clinical Oncology (ASCO) recommends that genetic testing be offered to individuals with suspected inherited (genetic) cancer risk in situations where test results can be interpreted, and when they affect medical management of the patient. It is sufficient for cancer risk assessment to evaluate genes of established clinical utility that are suggested by the patient's personal and/or family history.

Adapted from J Clin Oncology, 2015

Patients with a Personal and/or Family History Suggestive of Hereditary Cancer May Benefit From CancerNext-*Expanded*

If your patient has a personal or family history of **ANY** of the following signs* of hereditary cancer, genetic testing should be considered:

CANCER TYPE	MULTIPLE CANCERS OR OTHER CLINICAL RISK FACTORS	EARLY-ONSET CANCERS	ANCESTRY
MALE BREAST OVARIAN PANCREATIC METASTATIC PROSTATE CANCER	2 OR MORE primary cancers in the same person 3 OR MORE cancers on the same side of the family 10 OR MORE colorectal polyps in a person's lifetime	ANY OF THE FOLLOWING CANCERS DIAGNOSED BEFORE 46 YEARS of AGE: Breast, colorectal, uterine	ASHKENAZI JEWISH WITH BREAST CANCER

** This is a suggested list adapted from published genetic testing guidelines*

CancerNext-*Expanded* Genes and Associated Cancers

Turnaround time: 14-21 days

77 gene hereditary cancer test

Management guidelines available for many included genes

GENE(S)	ASSOCIATED CANCERS											
	Breast	Ovarian	Colorectal	Uterine	Pancreatic	Prostate	Gastric	Kidney	Endocrine**	Central Nervous System	Melanoma	Other
AIP										✓		
ALK										✓		✓
*APC**			✓		✓		✓		✓	✓		✓
*ATM**	✓				✓	✓						
*AXIN2**			✓									
BAP1								✓			✓	✓
BARD1	✓											
BLM	✓		✓									
*BRCA1**	✓	✓			✓	✓						
*BRCA2**	✓	✓			✓	✓					✓	
*BMPR1A, SMAD4**			✓				✓					
*BRIP1**	✓	✓										
CDC73								✓				✓
*CDH1**	✓						✓		✓			
CDK4											✓	
CDKN1B									✓	✓		✓
CDKN2A					✓					✓	✓	
*CHEK2**	✓		✓			✓						✓
CTNNA1							✓					
DICER1		✓								✓		✓
EGFR												✓
EPCAM		✓	✓	✓	✓	✓	✓	✓		✓		✓
EGLN1									✓			
FANCC	✓											
FH								✓	✓			✓
FLCN								✓				
GALNT12			✓									
*GREM1**			✓									
HOXB13						✓						
KIF1B									✓			
KIT												✓
LZTR1										✓		
MAX									✓			
MEN1									✓	✓		✓
MET								✓				
MITF								✓			✓	

CancerNext-*Expanded* Genes and Associated Cancers

GENE(S)	ASSOCIATED CANCERS											
	Breast	Ovarian	Colorectal	Uterine	Pancreatic	Prostate	Gastric	Kidney	Endocrine**	Central Nervous System	Melanoma	Other
MLH1, MSH2*, MSH6*, PMS2**		✓	✓	✓	✓	✓	✓	✓		✓		✓
*MSH3**			✓									
*MUTYH**	✓		✓									
*NBN**												✓
*NF1**	✓								✓	✓		✓
NF2									✓	✓		✓
*NTHL1**			✓									
*PALB2**	✓	✓			✓	✓						
PDGFRA												✓
PHOX2B										✓		✓
*POLD1, POLE**			✓									
POT1										✓	✓	
PRKAR1A									✓	✓		✓
PTCH1										✓		✓
*PTEN**	✓		✓	✓				✓	✓	✓	✓	✓
*RAD51C**	✓	✓										
*RAD51D**	✓	✓				✓						
RB1											✓	✓
RECQL	✓											
RET									✓			
SDHA, SDHAF2, SDHB, SDHC, SDHD								✓	✓			
SMARCA4		✓								✓		✓
SMARCB1								✓		✓		✓
SMARCE1										✓		
*STK11**	✓	✓	✓		✓							✓
SUFU										✓		✓
TMEM127									✓			
*TP53**	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓
TSC1, TSC2								✓		✓		✓
VHL								✓	✓	✓		✓
XRCC2	✓											

*Management guidelines available

**Endocrine indicates at least one of the following: paraganglioma, pheochromocytoma, thyroid cancer, parathyroid cancer, carcinoid tumors, pancreatic neuroendocrine tumors, and/or adrenal tumors

Results of Genetic Testing May Inform Personalized Medical Management

The potential benefits of genetic testing for hereditary cancer include:

Inform treatment recommendations[1-9]

Consideration of risk-reducing measures, as appropriate

Determine eligibility for clinical trials

Identify at-risk family members to inform cancer risks and medical management

Supporting Access to Patient Testing

Testing for Family Members

Testing for all blood relatives is available within 90 days of the original report if the patient was tested at Ambry Genetics. Family testing is done via specific site analysis for pathogenic or likely pathogenic variants. (Excludes SNP array and applies to single gene, panel, or exome testing).

About Ambry Genetics®

Ambry Genetics, a subsidiary of REALM IDx, Inc., excels at translating scientific research into clinically actionable test results based on a deep understanding of the human genome and the biology behind genetic disease. Ambry has an unparalleled track record of discoveries over 20 years and a database that continually expands through collaboration with academic, corporate and pharmaceutical partners. Being first to market with innovative products and comprehensive analysis, Ambry enables clinicians to confidently inform patient health decisions. For more information, please visit ambrygen.com.

References

1. Banerjee S & Kaye S. Curr Oncol Rep. 2011 Dec;13(6):442-9
2. Burgess M & Puhalla S. Front Oncol. 2014 Feb 27;4:19
3. Yamamoto KN *et al.* PLoS One. 2014 Aug 26;9(8): e105724
4. Moore *et al.* NEJM 2018 Oct 21 (Epub ahead of print)
5. Ledermann, *et al.* Lancet Oncol. 2014;15(8):852-861
6. Pujade-Lauraine, *et al.* Lancet Oncol. 2017;18:1274-1284
7. Mirza, *et al.* N Engl J Med. 2016;375:2154-2164
8. Coleman RL, *et al.* Lancet. 2017 Oct 28;390(10106):1949-1961
9. Swisher, *et al.* Lancet Oncology 2017;18: 75-87

CONTACT INFORMATION

LifeStrands Genomics

61 Science Park Road The Galen #03-13/14 SG 117525 | T: (65) 6422 6322 | E: enquiry@lifestrandsgx.com | W: www.lifestrandsgx.com

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