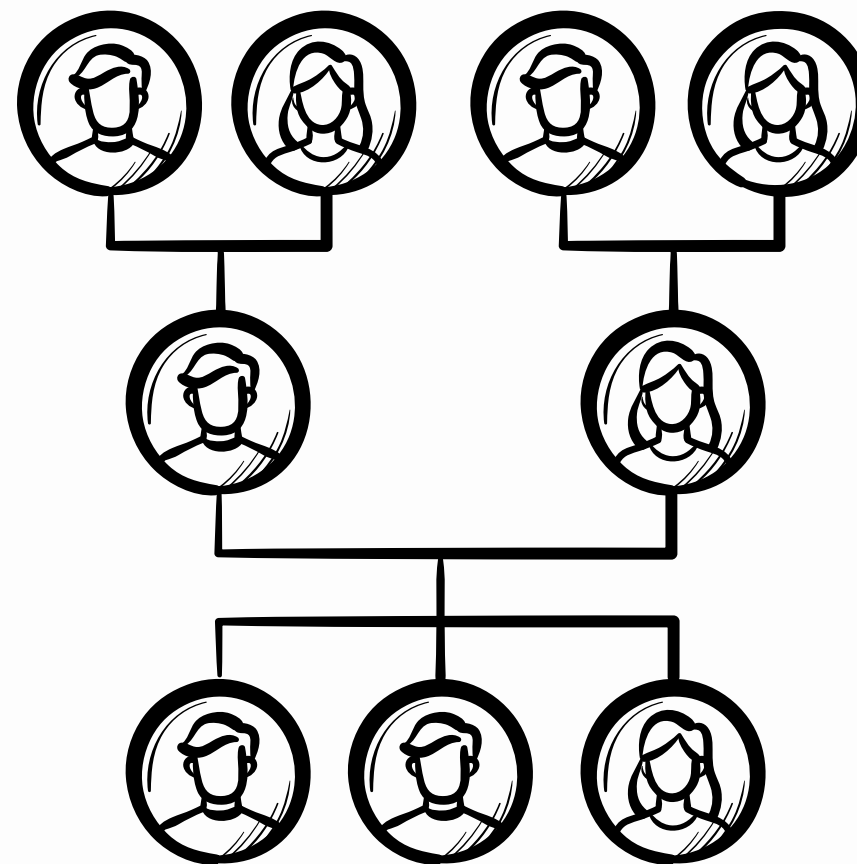


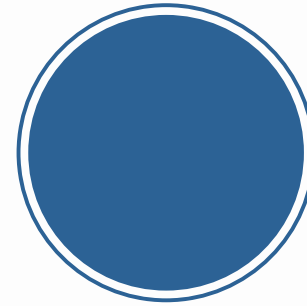
Tracing the Legacy: Understanding Hereditary Cancer and Its Modern Implications

Katia Vine, MS, CGC (she/her/Ella)
Certified Bilingual Genetic Counselor
Genetic Support Foundation

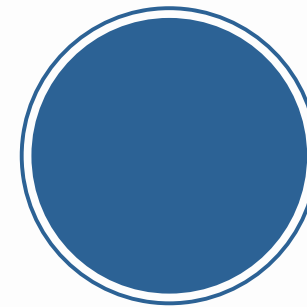
Jenny Rietzler, MS, CGC (she/her)
Certified Genetic Counselor
Genetic Support Foundation



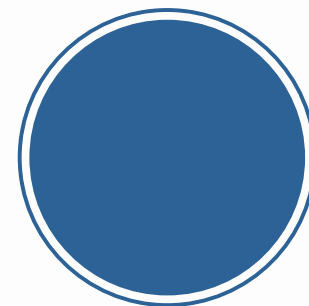
Learning Objectives



Overview of hereditary cancer



Genetic counseling and hereditary cancer



Hereditary cancer genetic testing history



All cancer has a genetic basis

TRUE

FALSE

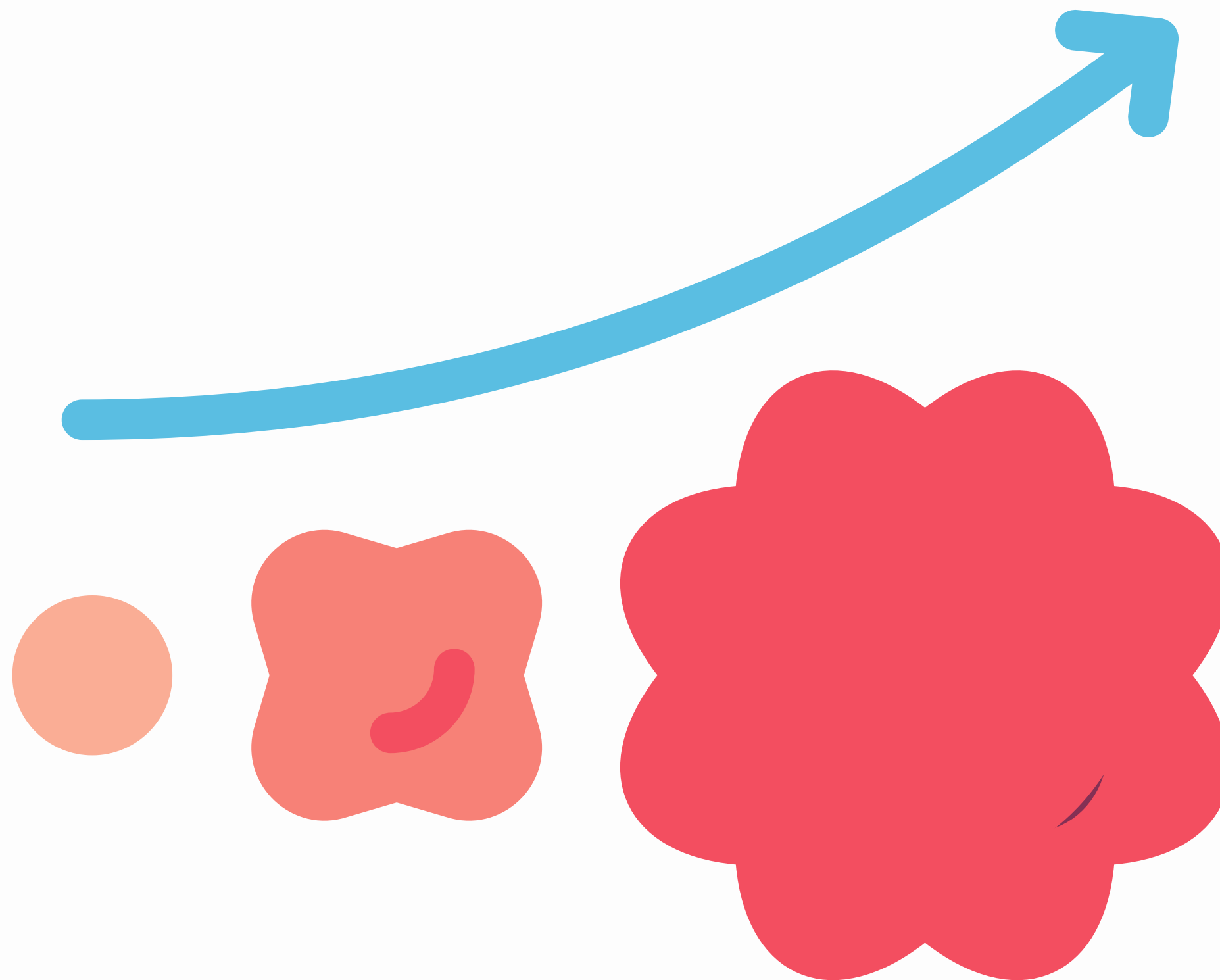


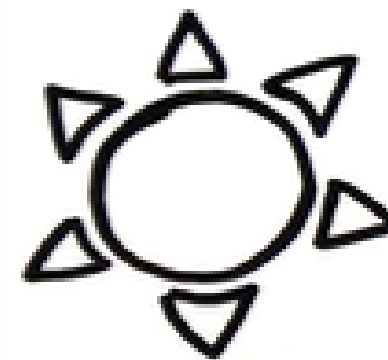
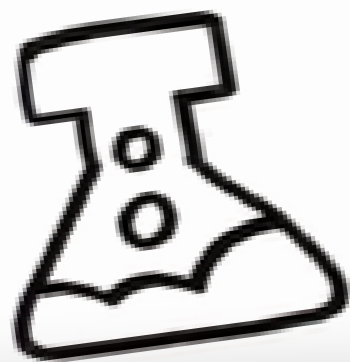
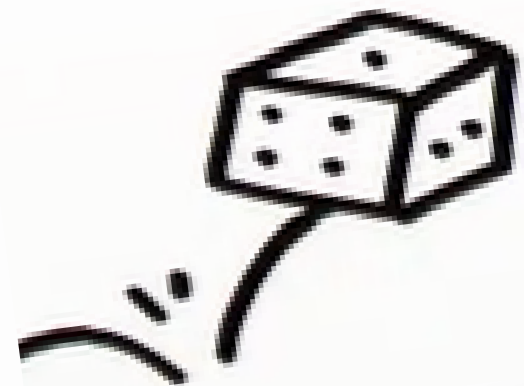
All cancer has a genetic basis

TRUE




FALSE






**Cancer happens because of
genetic damage to a cell that
builds up over time**





**Some people are born with
genetic damage (or a mutation)
versus acquiring damage**



Clues for Hereditary Cancer

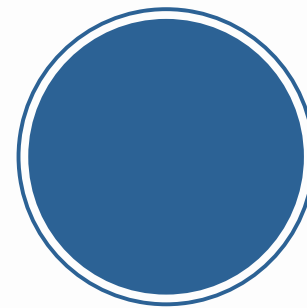
- Personal history of cancer
 - Age of onset before the age of 50
 - Triple negative breast cancer with any age of onset
 - Rare cancer with any age of onset: ovarian cancer, male breast cancer, pancreatic cancer, metastatic or high grade prostate cancer
- High risk ancestry
 - Breast cancer of any age for an individual with Ashkenazi Jewish ancestry

Clues for Hereditary Cancer

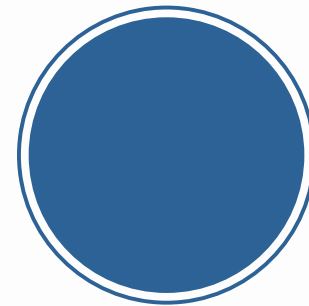
- Family history of cancer
 - Multiple relatives with same type of cancer and/or any relative with onset before the age of 50
 - Any family history of rare cancer with any age of onset: ovarian cancer, male breast cancer, pancreatic cancer, metastatic or high grade prostate cancer



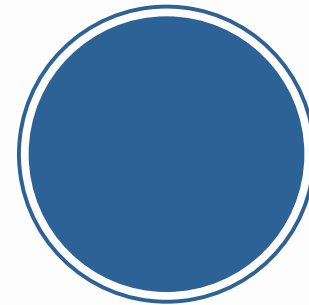
Why refer to genetic counseling?



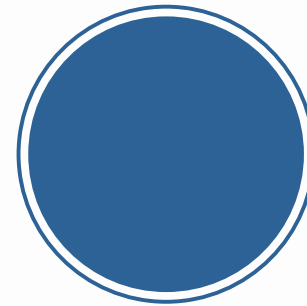
Family history



Informed consent

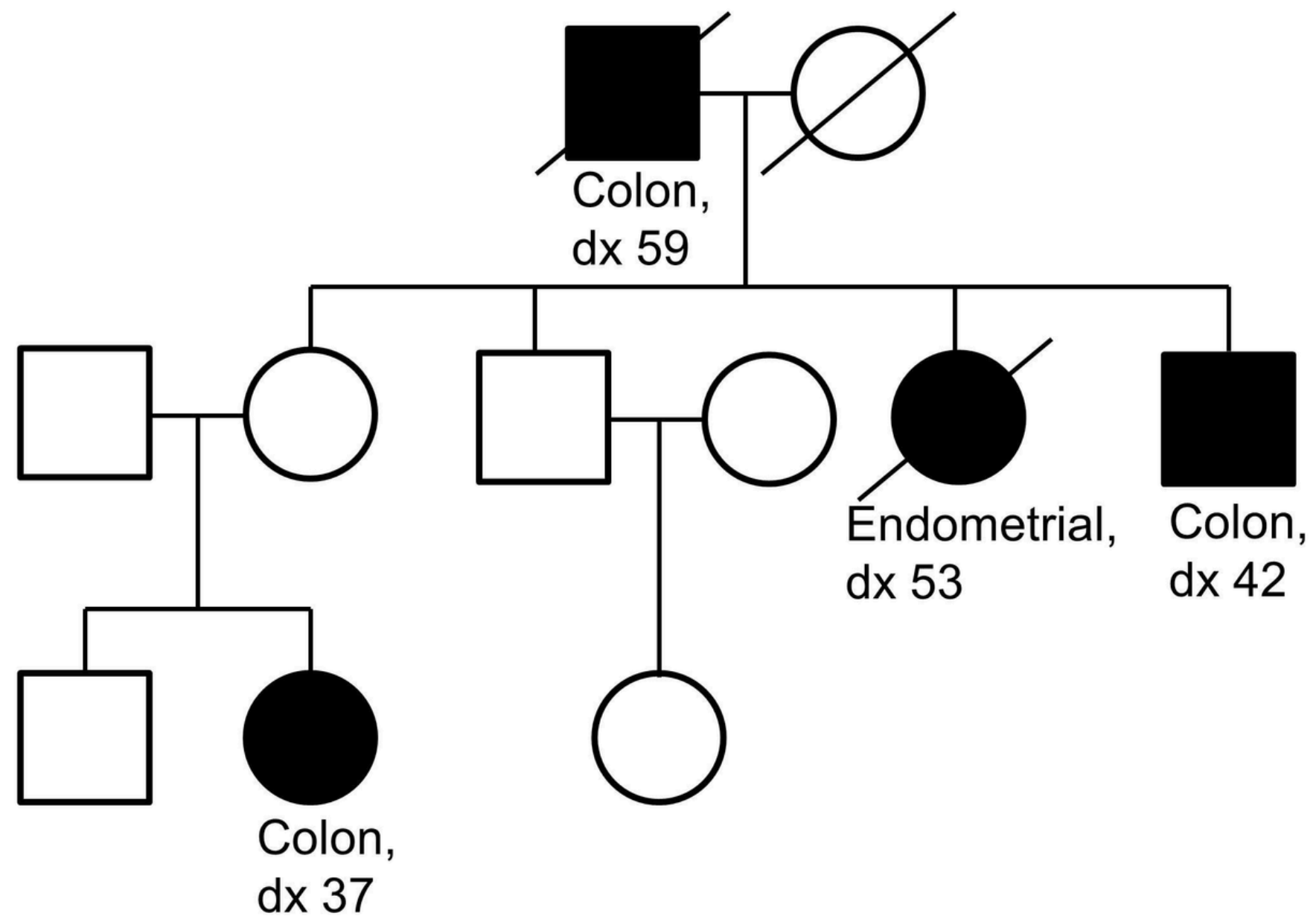


Anticipatory guidance



Personalized recommendations
for management

Family History



Informed Consent



Anticipatory Guidance



Benefits



Limitations



Risks

Personalized recommendations for management



Cost of genetic counseling appointments



- The cost of genetic counseling appointments varies across clinics and depends on insurance coverage
 - Some clinics offer genetic counseling appointments for \$0
 - Genetic counseling telehealth services may offer options such as reduced self-pay fees and financial assistance
- Primary CPT code used for billing is 96041. Patients are encouraged to contact insurance directly to assess coverage.

Cost of genetic testing for cancer susceptibility genes



Test	Timeframe for results	Cash Price
Panel testing	2-4 weeks	\$249 - >\$3000
Familial variant testing*	2-4 weeks	\$0 - >\$2000
STAT testing	1-2 weeks	\$249 - >\$3000

*Free testing window for a familial variant can vary between laboratories

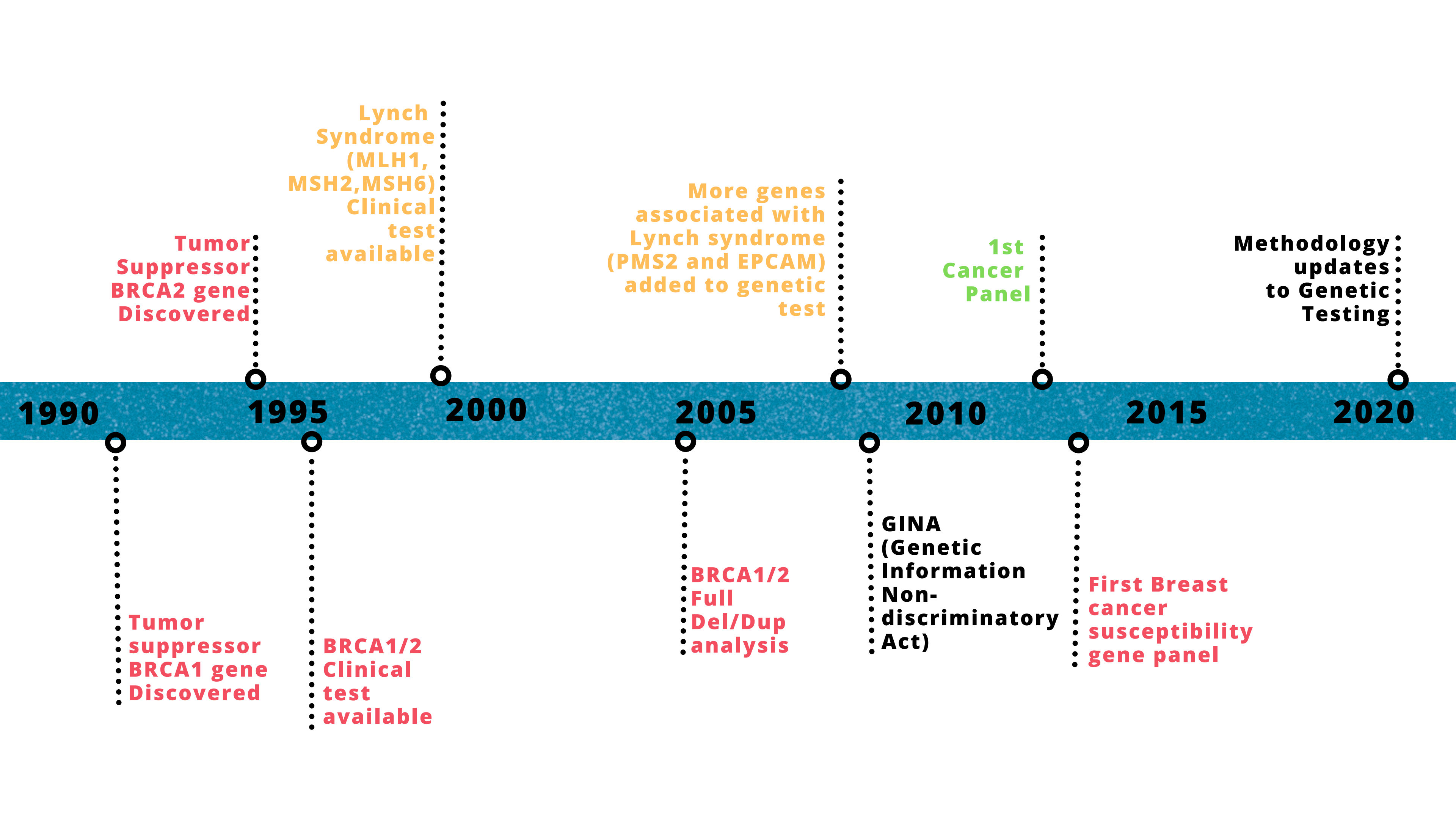
- The price after medical insurance payment varies significantly and is difficult to estimate accurately

Cost of genetic testing for cancer susceptibility genes



- Medicaid may limit testing company options, though it makes it illegal to charge a patient for testing (unless you consent to cash-pay ahead of time)
- Many testing companies have cost-reduction programs – always inquire ahead of testing and ask for a “benefits investigation”
- If deductible has not been met, costs may be 100s - 1,000s, even with insurance

Hereditary cancer: Genetic testing history



1990

1995

2000

2005

2010

2015

2020

Tumor
Suppressor
BRCA2 gene
Discovered

Lynch
Syndrome
(MLH1,
MSH2,MSH6)
Clinical
test
available

More genes
associated with
Lynch syndrome
(PMS2 and EPCAM)
added to genetic
test

1st
Cancer
Panel

Methodology
updates
to Genetic
Testing

Tumor
suppressor
BRCA1 gene
Discovered

BRCA1/2
Clinical
test
available

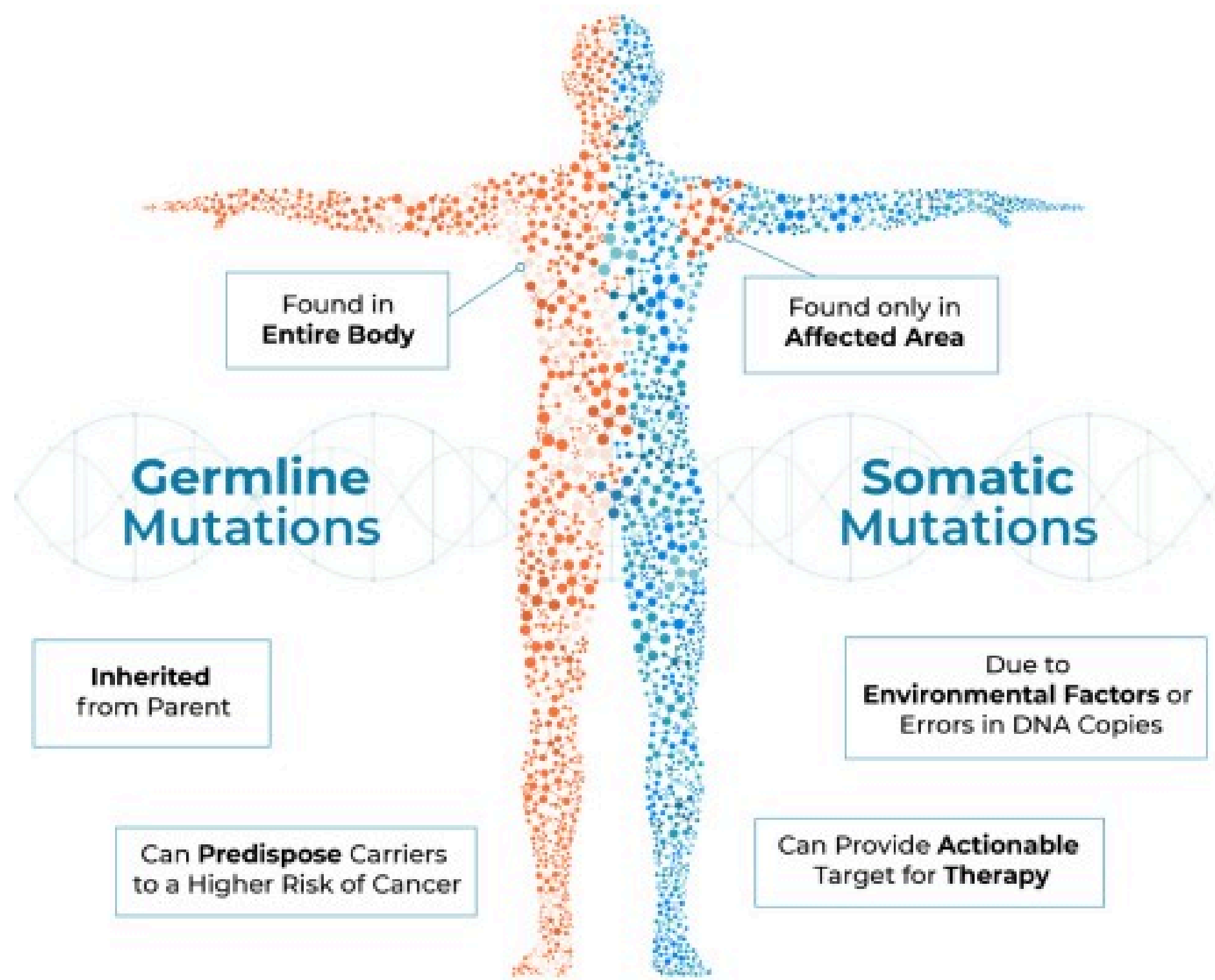
BRCA1/2
Full
Del/Dup
analysis

GINA
(Genetic
Information
Non-
discriminatory
Act)

First Breast
cancer
susceptibility
gene panel

Future role of genetic testing in cancer

Integrated tumor and germline testing



Integrated germline and somatic cancer testing provides opportunity to identify cancer risk and resolve variant origins.

Authors: [Kingshuk Das](#), [Amber Carter](#), [Brandie Heald](#), [Scott T. Michalski](#), [Sarah M. Nielsen](#), [Nhu Ngo](#), [Sara Elrefai](#), [Kelly Warsinske](#), [Stacy Lenarcic](#), [Heather Hampel](#), [Robert Luke Nussbaum](#), and [Edward D. Esplin](#) | [AUTHORS INFO & AFFILIATIONS](#)

Publication: Journal of Clinical Oncology • Volume 40, Number 16 suppl • https://doi.org/10.1200/JCO.2022.40.16_suppl.10589

Comment | Published: 27 May 2025

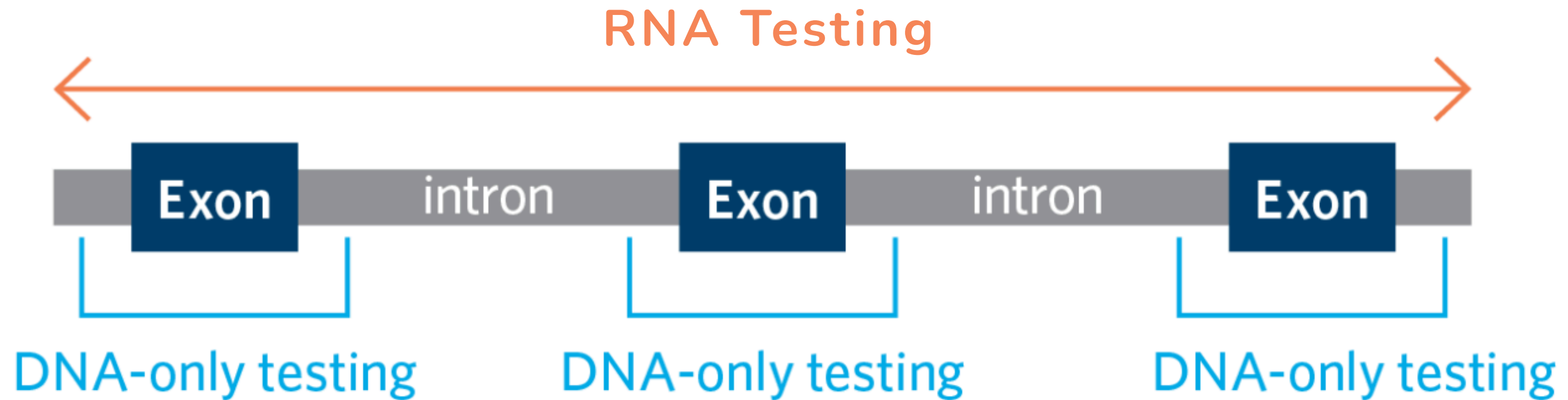
Patient-centered integration of tumor and germline genetic results can improve cancer care

[Alanna J. Church](#) ✉, [Oindrila Bhattacharyya](#), [Julie O. Culver](#), [Yonatan Amzaleg](#), [Erin Linnenbringer](#), [Maeve Smart](#), [Christina Ip-Toma](#), [Adan Reinos Rivera](#), [Bethany Davis](#), [Charité Ricker](#), [Heinz-Josef Lenz](#), [Stacy W. Gray](#), [Heather Hampel](#) & [David W. Craig](#)

[Nature Medicine](#) (2025) | [Cite this article](#)

883 Accesses | 8 Altmetric | [Metrics](#)

RNA Genetic Testing for Hereditary Cancer



<https://www.ambrygen.com/providers/genetic-testing/rna>

Original Investigation

Diagnostic Outcomes of Concurrent DNA and RNA Sequencing in Individuals Undergoing Hereditary Cancer Testing

Carolyn Horton, MS¹; Lily Hoang, BS¹; Heather Zimmermann, PhD¹; [et al](#)

» [Author Affiliations](#) | [Article Information](#)

» [RELATED ARTICLES](#) [FIGURES](#) [SUPPLEMENTAL CONTENT](#)

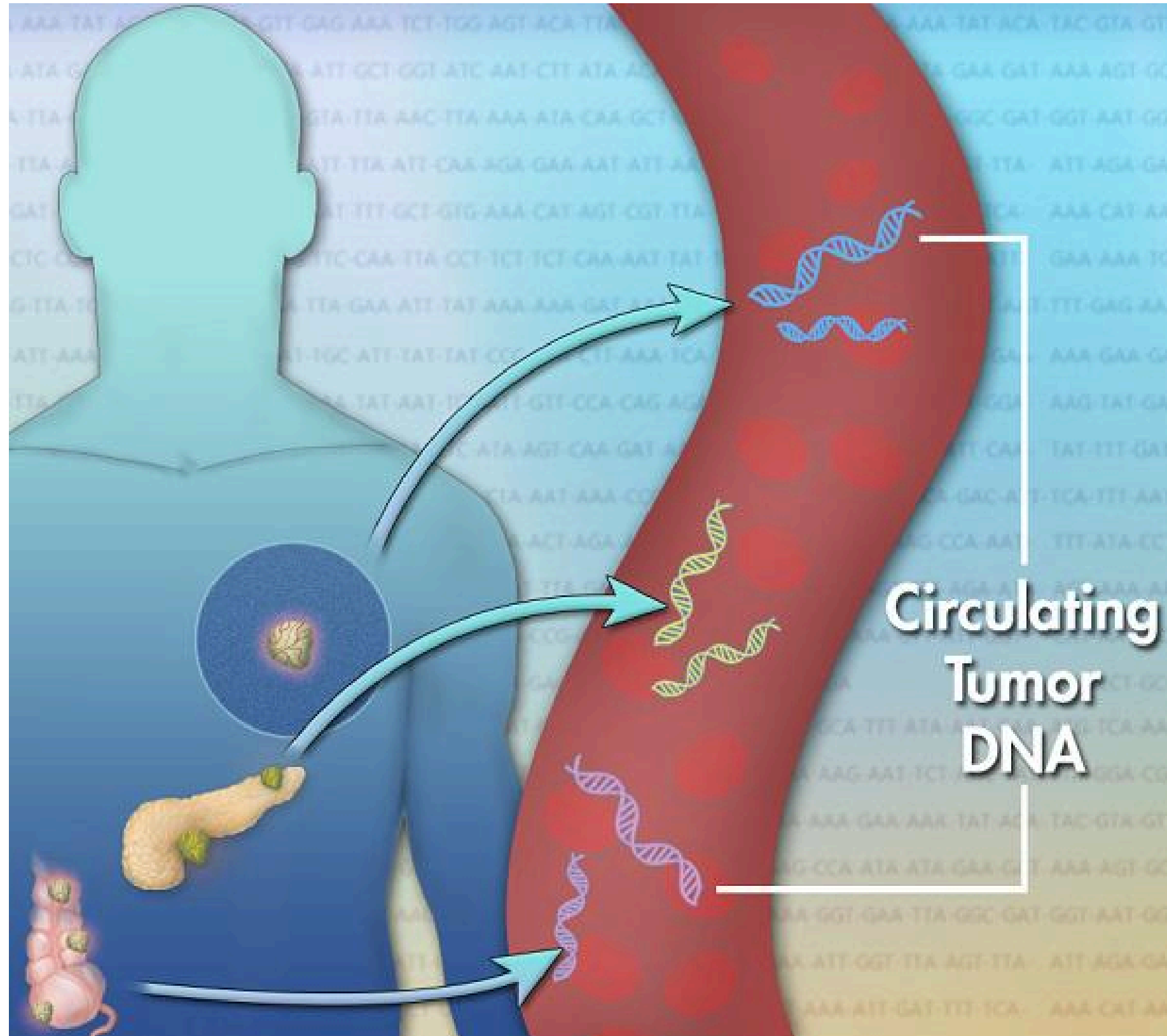
Assessment of Diagnostic Outcomes of RNA Genetic Testing for Hereditary Cancer

Rachid Karam¹, Blair Conner¹, Holly LaDuca¹, Kelly McGoldrick¹, Kate Krempely¹, Marcy E Richardson¹, Heather Zimmermann¹, Stephanie Gutierrez¹, Patrick Reineke¹, Lily Hoang¹, Kyle Allen¹, Amal Yussuf¹, Suzette Farber-Katz^{1 2}, Huma Q Rana³, Samantha Culver³, John Lee⁴, Sarah Nashed⁵, Deborah Toppmeyer⁵, Debra Collins⁶, Ginger Haynes¹, Tina Pesaran¹, Jill S Dolinsky¹, Brigitte Tippin Davis¹, Aaron Elliott¹, Elizabeth Chao^{1 7}

Affiliations + expand

PMID: 31642931 PMCID: PMC6820040 DOI: 10.1001/jamanetworkopen.2019.13900

Circulating tumor DNA testing



Credit: Jonathan Bailey, NHGRI

Circulating Tumor DNA Analysis in Patients With Cancer: American Society of Clinical Oncology and College of American Pathologists Joint Review

Jason D Merker¹, Geoffrey R Oxnard¹, Carolyn Compton¹, Maximilian Diehn¹, Patricia Hurley¹, Alexander J Lazar¹, Neal Lindeman¹, Christina M Lockwood¹, Alex J Rai¹, Richard L Schilsky¹, Apostolia M Tsimberidou¹, Patricia Vasalos¹, Brooke L Billman¹, Thomas K Oliver¹, Suanna S Bruinooge¹, Daniel F Hayes¹, Nicholas C Turner¹

Affiliations + expand

PMID: 29504847 DOI: 10.1200/JCO.2017.76.8671

The value of cell-free DNA for molecular pathology

Caitlin M Stewart^{1 2}, Prachi D Kothari^{1 3}, Florent Mouliere^{4 5}, Richard Mair^{4 5 6}, Saira Somnay⁷, Ryma Benayed², Ahmet Zehir², Britta Weigelt², Sarah-Jane Dawson^{8 9 10}, Maria E Arcila², Michael F Berger^{1 2}, Dana Wy Tsui^{1 2}

Affiliations + expand

PMID: 29380875 PMCID: PMC6656375 DOI: 10.1002/path.5048

Cancer Vaccines

Personalized Cancer Vaccines Go Viral: Viral Vectors in the Era of Personalized Immunotherapy of Cancer

Laura Secl¹, Guido Leoni¹, Valentino Ruzza¹, Loredana Siani¹, Gabriella Cotugno¹, Elisa Scarselli¹, Anna Morena D'Alise¹

Affiliations + expand

PMID: 38068911 PMCID: PMC10706435 DOI: 10.3390/ijms242316591

Review Article | Published: 23 August 2022

Cancer vaccines: the next immunotherapy frontier

[Matthew J. Lin](#), [Judit Svensson-Arvelund](#), [Gabrielle S. Lubitz](#), [Aurélien Marabelle](#), [Ignacio Melero](#), [Brian D. Brown](#) & [Joshua D. Brody](#) 

[Nature Cancer](#) **3**, 911–926 (2022) | [Cite this article](#)

128k Accesses | **628** Citations | **480** Altmetric | [Metrics](#)



https://news.cancerresearchuk.org/shorthand_story/cancer-vaccines-where-are-we/



Questions?

Thank You!

844-743-6384

geneticsupportfoundation.org

kvine@geneticsupport.org

jrietzler@geneticsupport.org

