Time to testing for hereditary cancer: a comparison of age at cancer diagnosis and genetic testing

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BACKGROUND

- Completion of hereditary cancer genetic testing can be influenced by clinician experience, expert guidelines[1], access to care, insurance coverage, and other factors
- Despite existing testing guidelines, many patients who meet criteria experience delays in testing[2] which can impact early intervention, treatment, and cascade testing for family members

AIM: Evaluate testing delays by comparing age at cancer diagnosis and age at testing across cancer types and ethnicities

METHODS

- Difference in age = Age at genetic testing – Age at cancer diagnosis
- Means were compared using unpaired t-tests; $p<0.05$ was considered significant

INCLUSION CRITERIA

Select individuals with a personal history of cancer who underwent multigene panel testing at a single diagnostic laboratory between 2018-August 2025

EXCLUSION CRITERIA

Unknown/unclear cancer history or age at diagnosis, or multiple primary cancers other than non-melanoma skin cancer

RESULTS

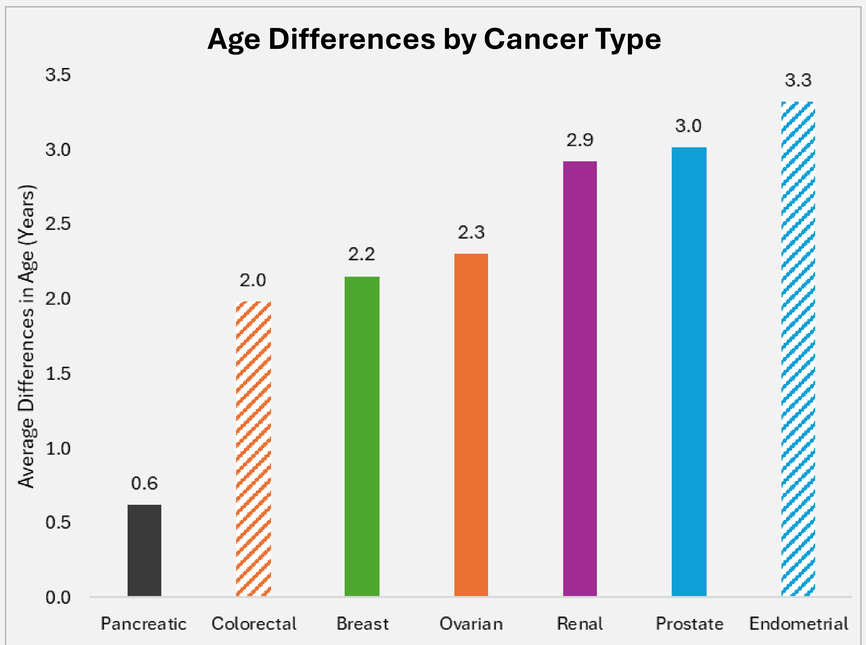

Figure 1. Mean difference in age at cancer diagnosis compared to age at testing by cancer type in this cohort.

Characteristic	N (%)		
Sex Assigned at Birth			
Female	53,989 (82)		
Male	11,818 (18)		
Unknown	7 (<1)		
Genetic Testing Characteristics			
# of Genes Tested, Median (*Range*)	60 (*1-101*)		
(Likely) Pathogenic Variant Identified*	11,632		
Race, Ethnicity & Ancestry (REA)			
African American/Black	6,320 (10)		
Ashkenazi Jewish	904 (1)		
Asian	1,597 (2)		
Hispanic	3,636 (6)		
Caucasian/White	27,437 (42)		
Multiple/Other (Unknown)	2,883 (23,037) (39%)		
Personal History of Cancer			
Breast	43,473 (66)	Pancreatic	3,996 (6)
Colorectal	5,005 (8)	Prostate	6,190 (9)
Endometrial	2,339 (4)	Renal	1,309 (2)
Ovarian	3,502 (5)		
Total # of Individuals	65,814		

Table 1. Summary of demographics. *Excluded autosomal recessive heterozygous variants.

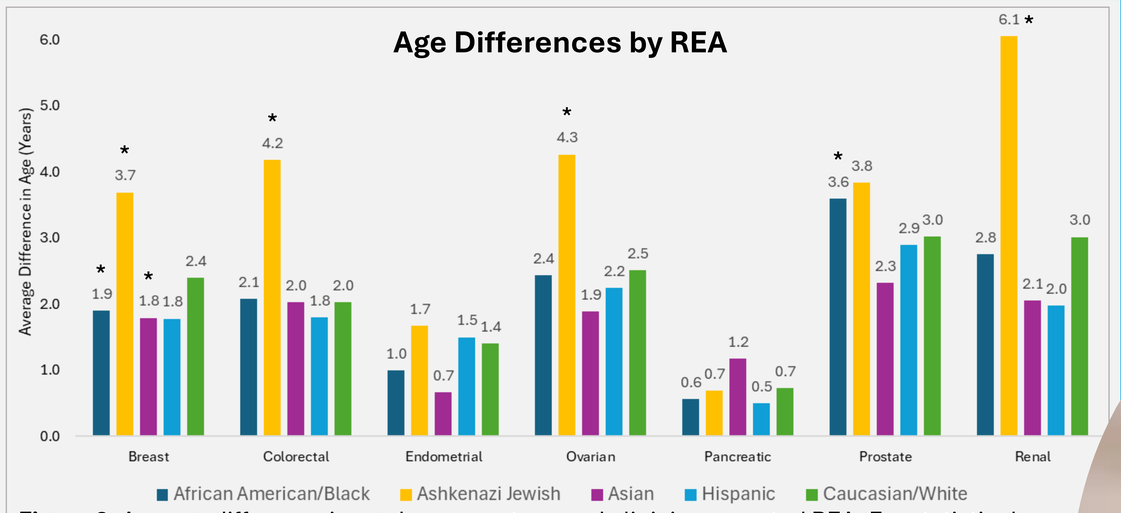

Figure 3. Average difference in age by cancer type and clinician-reported REA. For statistical analyses, Caucasian/White was used as the reference and * indicates p-value <0.05.

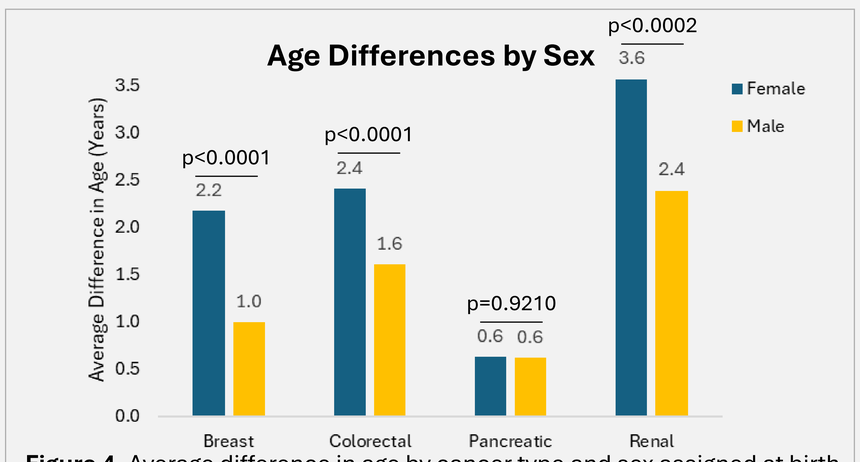

Figure 4. Average difference in age by cancer type and sex assigned at birth.

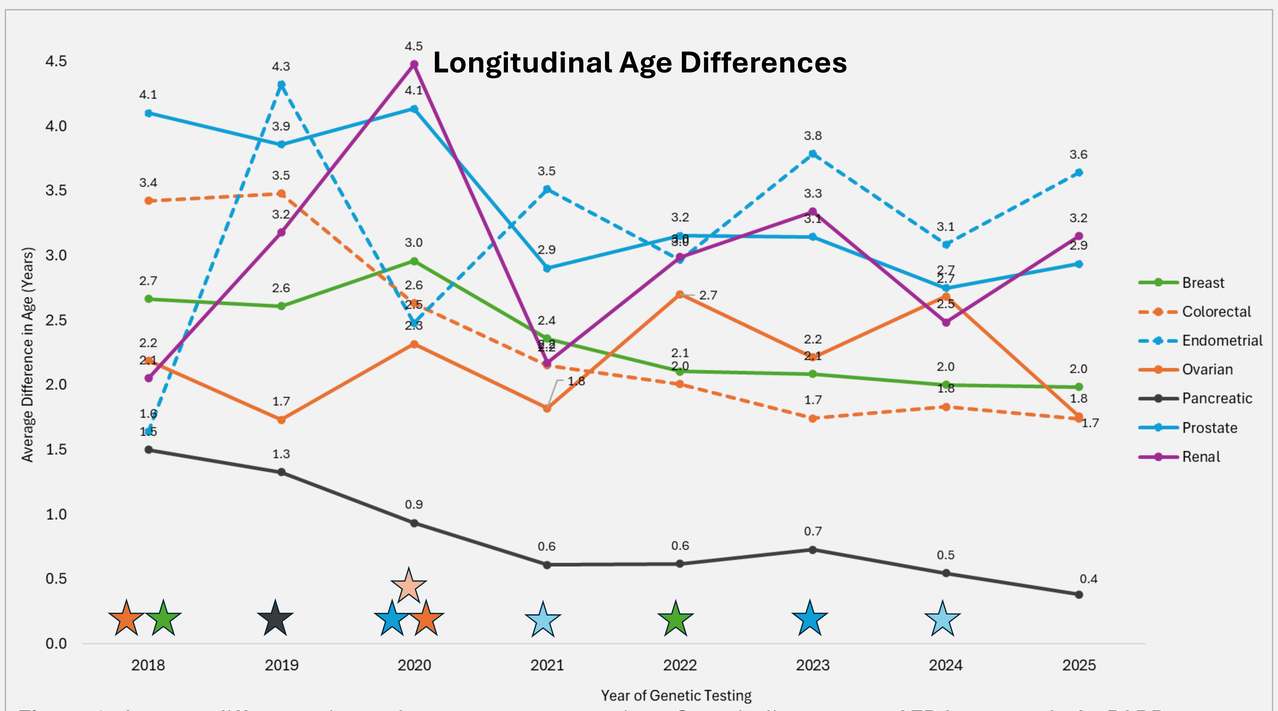

Figure 2. Average difference in age by cancer type over time. Stars indicate year of FDA approval of a PARP inhibitor or immunotherapy for cancer-specific treatment, with color corresponding to the impacted cancer type.

TAKE HOME POINTS

- Individuals with pancreatic cancer had the shortest delay in genetic testing and individuals with endometrial cancer had the longest.
- Ovarian cancer was associated with a 2.3-year delay which is of note given the treatment implications and morbidity with this disease.
- Testing for cancers tended to increase with FDA approval of relevant therapies.
- Historically, fewer males complete genetic testing. However, when they do, testing occurs significantly closer to diagnosis for breast, colorectal, and renal cancer compared to females with the same diagnoses.
- Ashkenazi Jewish ethnicity had the largest delay in genetic testing compared to other ethnicities; individuals reported as Asian with pancreatic cancer or African American with prostate cancer may also have delays in testing.
- Future directions include expanding analyses to additional cancer types, evaluating specific cancer pathologies, assessing whether social determinants of health impact testing delays, and controlling for year of cancer diagnosis.

REFERENCES

1. Hampel H, Bennett RL, Buchanan A, *et al*. A practice guideline from the American College of Medical Genetics and Genomics and the National Society of Genetic Counselors: referral indications for cancer predisposition assessment. Genet Med. 2015. PMID: 25394175.
2. Byrne M, Sia TY, Fong C, *et al*. Mainstreaming in parallel with ovarian cancer tumor testing to improve genetic testing uptake. Gynecol Oncol. 2024. PMID: 38493020.