Inequitable Access to Genetic Testing Leads to Missed Screening and Prevention Opportunities for Individuals at Risk for Hereditary Breast and Ovarian Cancer

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BACKGROUND

- The NCCN[1] recommends offering increased medical management for hereditary breast and ovarian cancer (HBOC) women with a pathogenic/likely pathogenic variant (PV/LPV) in 8 genes:
  - *ATM*, *BRCA1*, *BRCA2*, *CDH1*, *CHEK2*, *PALB2*, *PTEN*, & *TP53*.
- Management options, depending on the gene, include:
  - Biannual clinical breast exams
  - Annual breast MRI screening
  - Risk-reducing mastectomy (RRM)
  - Risk-reducing salpingo-oophorectomy (RRSO).
- Despite this, health insurance policies addressing the medical necessity of genetic testing for these genes vary widely.
- An assessment of the impact of this variability on access to enhanced medical management for women with a PV/LPV in these 8 HBOC genes in a large cohort of patients is needed.

METHODS

- Clinical and family histories were reviewed for patients undergoing multigene panel testing for that included the 8 genes of interest.
- Each patient was assessed for meeting criteria for 4 policies:
  - #1 = **NCCN** (adopted by Anthem, Cigna and others), excluding prostate cancer criteria, which were not assessed
  - #2 = **Aetna**
  - #3 = **Blue Shield California/Federal Blue Cross-Blue Shield** (both have since revised their policies to align with NCCN)
  - #4 = **eviCore** (used by over 30 payors including AmeriHealth, Highmark and Horizon)
- We determined the potential missed management opportunities for those who would not have been tested under each payor's criteria.
- We estimated the maximal clinical impact of these missed screening and risk-reduction options for patients with BRCA PV/LPV (not accounting for patient age or medical history such as prior risk-reducing surgery).

RESULTS

Met NCCN HBOC criteria*[1]: n=107,344

→ Under Policies 1, 2, and 3, 2% to 10.3% of patients (n=2547 – 13,116) would not have been eligible for genetic testing.

HBOC PVs/LPVs: n=10,477 (9.8%)

Under Policies 1, 2, and 3, up to 4% (n=423) would not have been eligible for genetic testing

→ Based on NCCN HBOC gene management:
- Missed breast MRI opportunity: n=423
- Missed RRM opportunity: n=208
- Missed RRSO opportunity: n=163

→ Potential missed benefit to *BRCA* carriers:
- 132 breast cancers detected earlier or prevented**
- 52 ovarian cancers prevented^

* Excluding prostate cancer criteria.
** Assuming an 85% lifetime risk for female breast cancer with BRCA PV/LPV & risk-reduction of 95% with RRM
^ Assuming a 40% lifetime risk for ovarian cancer with BRCA and risk-reduction of 80% with RRSO

Figure 1. Payor Policy* Alignment with NCCN Criteria

	BREAST CANCER, AFFECTED	#1 - AETNA	#2 - BS CS/ BCBS-FED	#3 - EVICORE	#4 - NCCN
BREAST	Breast cancer dx <=45				
	Breast cancer dx 46-50 + unknown or limited family history				
	Breast cancer dx 46-50 + a second breast cancer at any age				
	Breast cancer dx 46-50 + >=1 close blood relative with breast, ovarian, pancreatic, or prostate cancer				
	Breast cancer dx <=60 + triple-negative breast cancer				
	Breast cancer + Ashkenazi Jewish ancestry				
	Breast cancer + >=1 close blood relative with breast <=50, ovarian, pancreatic, or metastatic/high- or very high-risk group prostate cancer				
	Breast cancer + >=3 total diagnoses of breast cancer in patient and/or close blood relatives				
	Male breast cancer				
	FAMILY HISTORY				
BREAST	FDR/SDR with Breast cancer dx <=45				
	FDR/SDR with Breast cancer dx 46-50 + unknown or limited family history				
	FDR/SDR with Breast cancer dx 46-50 + a second breast cancer at any age				
	FDR/SDR with Breast cancer dx 46-50 + >=1 close blood relative with breast, ovarian, pancreatic, or prostate cancer at any age				
	FDR/SDR with Breast cancer dx <=60 + triple-negative breast cancer				
	FDR/SDR with Breast cancer + Ashkenazi Jewish ancestry				
	FDR/SDR with Breast cancer + >=1 close blood relative with breast <=50, ovarian, pancreatic, or metastatic/high- or very high-risk group prostate cancer				
	FDR/SDR with Breast cancer + >=3 total diagnoses of breast cancer in patient and/or close relatives				
	FDR/SDR with Male breast cancer				
OVARIAN	FDR/SDR with Epithelial ovarian cancer				
PANCREATIC	FDR with Exocrine pancreatic cancer at any age				

Payor policy covers more than NCCN criterion
Payor policy is aligned with NCCN
Payor policy covers some of NCCN criterion
Payor policy covers less than NCCN criterion
Payor policy does not cover NCCN criterion

*Policy versions reviewed:
- Aetna (Effective 4/15/2021)
- Blue Shield of CA / Federal BC/BS (Effective 12/1/2020)
- eviCore (Effective 7/1/2020)
- NCCN v2.2021 (Effective 11/20/2020)

CONCLUSIONS

Payor policies differed widely from NCCN criteria (Fig. 1). Under the most limited payor policy:

- **10% of women meeting NCCN guidelines would not have been tested.**
  - Payor-specific policies can miss a substantial proportion of women appropriate for genetic testing.
- **4% of women with PV/LPV would not have been tested.**
  - Varying testing guidelines lead to mutation carriers being missed by payor policies that deviate from NCCN.
- **423 women with PV/LPVs would not have been offered increased screening and/or risk-reducing surgeries because they were untested.**
  - Lack of testing access due to misaligned medical policies represents missed opportunities for offering screening and risk reduction options that could potentially save lives.

Uniform payor criteria could potentially reduce healthcare costs.

- Aligning testing criteria across payors will reduce the time spent by healthcare providers in assessing medical necessity for each patient.
- By eliminating the effort needed to develop their own customized policies, these actions could reduce health insurer costs as well.

These finding support payors adopting uniform criteria for genetic testing, such as are offered by NCCN.

REFERENCES

1. Daly MB, Pal T, Berry MP, et al.; NCCN Clinical Practice Guidelines in Oncology: NCCN Guidelines for Genetic/Familial High-Risk Assessment: Breast, Ovarian and Pancreatic. Version 2.2021.Available at NCCN.org

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