FORCE response to the USPSTF Draft Recommendation Statement: Risk Assessment, Genetic Counseling, and Genetic Testing for BRCA-Related Cancer

http://www.uspreventiveservicestaskforce.org/draftrec.htm

Facing Our Risk of Cancer Empowered (FORCE) is a national nonprofit organization that advocates for and improves the lives of individuals and families affected by Hereditary Breast and Ovarian Cancer, including families with a BRCA mutation.

FORCE recognizes that the USPSTF recommendations impact two important areas of patient preventive care:

1. The panel provides evidence-based guidelines for primary care clinicians and health systems on topics of preventive health.
2. The panel's guidelines are cited in the recently-enacted Patient Protection and Affordable Care Act (PPACA); thereby impacting patient access to care and insurance coverage for members of our constituency. The PPACA states that:

   "Coverage of Preventive Services
   PHS Act section 2713 and the interim final regulations require non-grandfathered group health plans and health insurance coverage offered in the individual or group market to provide benefits for and prohibit the imposition of cost-sharing requirements with respect to, the following:

   - *Evidenced-based items or services that have in effect a rating of "A" or "B" in the current recommendations of the United States Preventive Services Task Force (USPSTF) with respect to the individual involved;"*[1]

The following is FORCE's response to the USPSTF draft guidelines for Genetic Risk Assessment and BRCA Mutation Testing for Breast and Ovarian Cancer Susceptibility as it is currently written. Each concern relates to one or both of the areas of impact mentioned above.

1. **Defining the patient population under consideration**

The USPSTF recommendation states: *"These recommendations apply to women who have not received a diagnosis of breast or ovarian cancer but who have family members with breast or ovarian cancer whose BRCA status is unknown. Women presenting to their primary care providers who have a relative with a known potentially harmful mutation in the BRCA1 or BRCA2 genes should receive genetic counseling and consideration for testing. These recommendations do not apply to men, although male family members may be identified for testing during the course of the evaluation."*

FORCE recommends:

- Extend the evaluation and letter grade to women with a known mutation in the family. As written, the USPSTF acknowledges that women with a relative with a known mutation in BRCA1 or BRCA2 should receive genetic counseling and consideration for testing. But by excluding this population in the "Patient Population Under Consideration" section, it implies that the Grade B letter grade for genetic counseling and testing does not extend to the population. Thus the PPACA provisions would not extend to this population.
    - The data on genetic counseling and testing is even stronger for someone who has a known BRCA mutation in the family for whom testing is more likely to be informative than someone without a known mutation in the family.
    - The USPSTF could alter the guidelines to clearly state that women with a relative with a known mutation should also receive genetic counseling and testing services.
- Extend the evaluation and letter grade to women who have been diagnosed with breast cancer and who meet criteria based on personal and family history of cancer for genetic counseling and testing.
    - Research indicates that women with a BRCA mutation who have already been diagnosed with breast cancer are at very high risk for a second primary breast cancer and for ovarian cancer.[2,3,4]
    - Cancer screening and prevention options for a new cancer diagnosis in this cohort of breast cancer survivors are similar to those for women with a mutation who have not had cancer.
    - The prospective PROSE study showed decreased ovarian cancer-associated mortality in BRCA mutation carriers who chose risk-reducing salpingo-oophorectomy. This study included breast cancer survivors with BRCA mutations.[5]
    - Testing within a family is more cost-effective and most likely to yield a conclusive result if it begins with someone who has had a cancer diagnosis consistent with a hereditary cancer syndrome. Omission of this population under "Patient

Populations Under Consideration" implies that testing unaffected women in a family where there has been no identified mutation is a more appropriate approach than first testing cancer survivors in the family who have a greater likelihood of an informative test. Further, under the PPACA, omission of survivors from these guidelines will impact access to care and coverage for preventive services in this population. This may inadvertently increase inappropriate and more costly initial BRCA tests of unaffected family members, rather than first testing the candidate in a family who is most likely to test positive and then testing unaffected individuals for the identified mutation.

2. Defining health care providers who provide genetic counseling

The USPSTF recommendation states: *"Genetic counseling regarding BRCA mutation testing may be performed by trained health professionals, including trained primary care providers."*

Genetics is a rapidly evolving area of medicine and most primary care providers do not have formal training in genetics and receive their information from the laboratories performing the testing.[6,7]

The American College of Surgeons, Commission On Cancer (CoC) is a consortium of professional organizations dedicated to improving survival and quality of life for cancer patients through standard-setting, prevention, research, education, and the monitoring of comprehensive quality care. With regards to which experts should provide genetic counseling services the CoC states: *"Please note, specialized training in cancer genetics should be ongoing; educational seminars offered by commercial laboratories about how to perform genetic testing are not considered adequate training for cancer risk assessment and genetic counseling."*[8]

FORCE recommends:

- Define health care providers who provide genetic counseling to specify providers who have training and clinical experience in cancer genetics. Adopt the Commission On Cancer's guidelines. Specify that training by commercial laboratories does not constitute advanced training in genetics.

3. Assigning a letter grade to risk-management options

The task force mentions risk-management interventions but does not assign letter grades to the specific prevention and screening options available for risk-management. Awarding a grade to the counseling and testing process acknowledges that genetic counseling and testing have clinical utility as preventive services; the clinical utility of genetic counseling and testing for BRCA, however, lies in the high-risk individual accessing interventions that will lower their risk for breast or ovarian cancer or detect these cancers at an earlier stage. Without a letter grade assigned to the interventions, these preventive services are not covered under the PPACA.

FORCE recommends:

- Review research to assign a letter grade to the following risk-management options.
  - Research shows that increased breast screening with mammogram and breast MRI leads to earlier detection of breast cancer in this cohort.[9,10,11]
  - Prospective data shows that bilateral risk-reducing mastectomy lowers the risk for breast cancer in this cohort.[12]
  - Prospective data demonstrates that risk-reducing bilateral salpingo-oophorectomy lowers cancer-specific and overall mortality in mutation carriers in this cohort.[5]
  - Research shows that use of oral contraceptives is associated with a lower risk for ovarian cancer in women with mutations.[13]

We support the USPSTF retaining parenthetical statements on the need for more research for risk-management interventions where evidence is lacking or inconclusive.

4. Overall guidelines

Regarding Screening, Risk Assessment, and Testing, the task force states: *"In the course of standard elicitation of family history information from patients, primary care providers should ask about specific types of cancer, which family members were affected, and the age and sex of affected family members."*

A family history of certain cancers associated with BRCA may also be indicative of other hereditary syndromes. Lynch Syndrome, for example, can be associated with ovarian, colon, and endometrial cancers. By providing guidelines only on identification of individuals who may carry a BRCA mutation, the USPSTF is missing the opportunity to guide practitioners to identify individuals who may carry other types of hereditary syndromes for which preventive care options are available.

Based on systematic evaluation of the extensive research evidence, the CDC's Office of Public Health Genomics has designated family history evaluation and genetic testing for Lynch Syndrome and BRCA in people with a personal or family history consistent with these cancers as *Tier 1 Public Health Interventions*.[14]

The USPSTF would better serve health care professionals and the public by developing a single set of evidence-based guidelines that address the collection and evaluation of personal and family medical history to identify individuals appropriate for genetic counseling and testing for hereditary disease syndromes. These guidelines should include hereditary disease syndromes that have associated genetics tests, have clinical utility, and have been evaluated by the task force.

FORCE recommends:

- Review of evidence and development of guidelines for Risk Assessment, Genetic Counseling, and Genetic Testing for Lynch Syndrome Related Cancers and other relevant hereditary cancer syndromes. These should be integrated with guidelines on Risk Assessment, Genetic Counseling, and Genetic Testing for BRCA-Related Cancer into one set of practice guidelines for collecting family history and referral of appropriate individuals for genetic counseling, testing, and related preventive services.

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[1] United States. US Department of Health & Human Services; Centers for Medicare & Medicaid Services. The Center for Consumer Information & Insurance Oversight**.** Affordable Care Act Implementation FAQs - Set 12. (http://cciio.cms.gov/resources/factsheets/aca_implementation_faqs12.html#_ftn5)

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