Growth of *BRCA1/2* Genetic Testing in Michigan, 2008-2011

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Background

The general population likelihood of *BRCA1/2* breast and ovarian cancer susceptibility gene mutation is estimated between 1/300 and 1/1,000.[1] In the Ashkenazi Jewish population, it is as high as 1/40.[2] Immediate relatives of any individual with a BRCA mutation have a 50% likelihood of mutation.

In 2005, the United States Preventive Services Task Force (USPSTF) issued a Grade B recommendation for genetic counseling and testing based on family history of breast and ovarian cancer and a Grade D recommendation against routine referral for those whose history is not associated with increased risk[3]. In addition, *Healthy People 2020* added a related goal[4]:

Healthy People 2020 Goal G-1
Increase the proportion of women with a family history of breast and/or ovarian cancer who receive genetic counseling

Genetic counseling facilitates accurate risk assessment and appropriate genetic testing with applicable follow-up care and surveillance. This process evaluates patients for testing and determines which tests are most relevant. However, *BRCA1/2* testing can be ordered by non-genetics providers; the frequency of testing by non-genetics providers was previously unknown.

In the United States, clinical testing for the *BRCA1/2* breast and ovarian cancer susceptibility genes is performed by Myriad Genetics, Inc. In 2012 Myriad disclosed data on the number of *BRCA* tests performed yearly in Michigan by type (comprehensive sequencing, rearrangement, three site and single site testing).

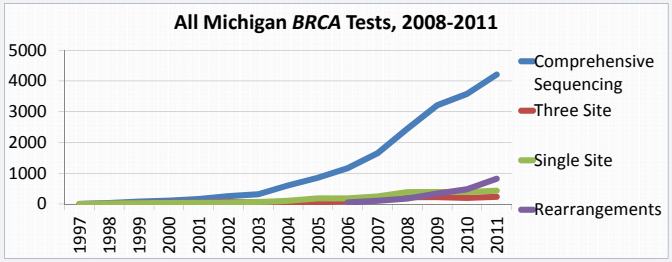
BRCA Test Type	Clinical Indication[3,5]	Cost	Description of Test
Comprehensive Sequencing	Significant family and/or personal history	$3,340	Full sequencing of BRCA1/2 and five specific large genomic rearrangements analyzed
Rearrangements	No mutation detected with Comprehensive, with significant family and/or personal history	$700	Large genomic rearrangements of BRCA1/2
Three Site	Ashkenazi Jewish ancestry with family and/or personal history	$575	Three specific mutations examined (accounts for >90% of BRCA mutations in Ashkenazi Jewish population)
Single Site	Known deleterious mutation in the family	$475	Specific single known mutation analyzed

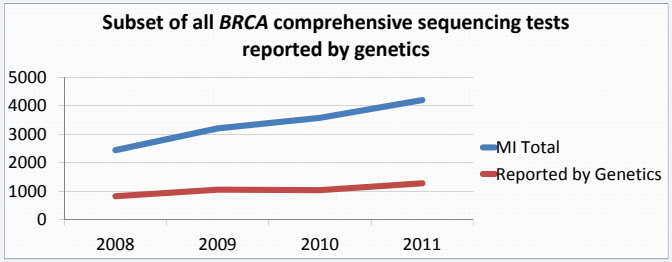
Methods

As part of a cooperative agreement with the Centers for Disease Control and Prevention, the Michigan Department of Community Health Cancer Genomics Program maintains a surveillance database with visit and testing information on all patients seen for *BRCA* genetic counseling by a board-certified genetics professional in Michigan, beginning with visits on October 1, 2007.

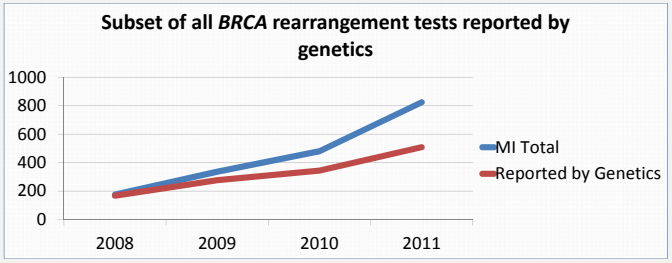
The data on *BRCA* tests ordered by board-certified genetics providers was compared to Myriad's total test numbers in Michigan annually from 2008 through 2011. For each of the four test types, percent growth from 2008 through 2011 and the annual proportion of tests ordered by genetics providers were calculated.

Results

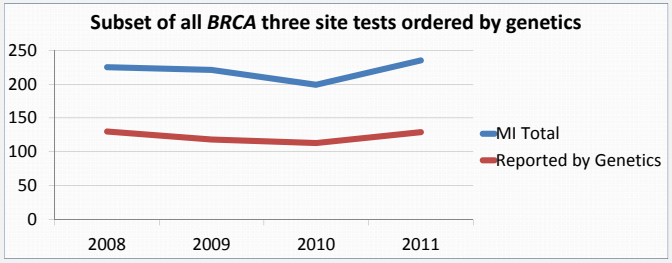




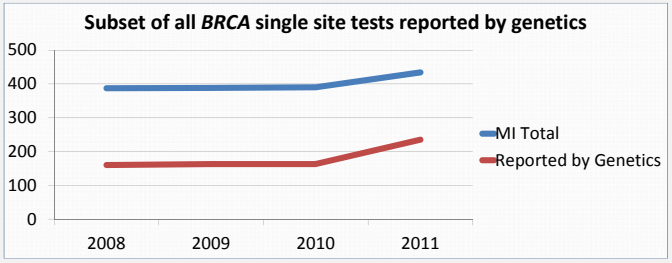
From 2008-2011, comprehensive testing in MI increased by 72.2%. In 2008, tests reported by genetics providers comprised 33.7% of the total; in 2011 they were 30.4%.



From 2008-2011, rearrangement testing in MI increased by 370.9%. In 2008, tests reported by genetics providers comprised 95.4% of the total, and this dropped to 61.7% in 2011.



From 2008-2011, three site testing in MI increased by 4.4%. In 2008, tests reported by genetics providers comprised 57.8% of the total; in 2011 they were 54.9%.



From 2008-2011, single site testing in MI increased by 12.1%. In 2008, tests reported by genetics providers comprised 41.3% of the total, and in 2011 they increased to 54.1%.

Conclusion

BRCA testing by both genetics and other ordering providers is increasing in Michigan. This is primarily due to increases in comprehensive and rearrangement testing; single site and three site tests constitute a diminishing proportion of all *BRCA* tests and display little growth in recent periods. Because single site and three site tests may both used for family members of individuals with identified mutations, we would expect to see these tests increase when high risk patients are correctly identified and tested.

While comprehensive and rearrangement testing are appropriate for those at increased risk of hereditary breast and ovarian cancer, single site and three site tests represent both appropriate clinical care and cost-savings in testing because they:

1) Apply to patients at the highest risk of *BRCA* mutation
2) Are lower cost compared to comprehensive and rearrangement testing
3) Are often used to identify known family mutations, which can impact recommended follow-up care.

Genetics providers disproportionately order single site and three site tests, and these providers represent a declining measure of all *BRCA* testing.

Policy and education efforts are needed to:

* **Promote genetics referral and improve provider knowledge of available genetics services**
* **Educate providers about risk identification and appropriate testing options**

Acknowledgements

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