Symposium 19: Integrated medical care for hereditary cancer syndromes

Genetic counseling and genetic testing for hereditary cancer syndromes

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Breast cancer can be seen as a feature in several hereditary cancer predisposition syndromes; however, sequential, single-syndrome testing has been the absolute of hereditary cancer assessment for decades. After careful evaluation of personal medical and family history, testing has traditionally begun with BRCA1/2 testing and then, if warranted based on the presence of characteristics of other hereditary cancer syndromes, followed with additional single gene testing. But now, the next-generation sequencing has made it possible to test for mutations in all known or suspected breast cancer predisposition genes in one panel, at one time.

Practice guidelines for genetic counselors have been well articulated by the National Society of Genetic Counselors. This guideline presents a current and comprehensive set of practice recommendations for effective genetic cancer risk assessment, counseling, and testing. Also U.S. Preventive Services Task Force Recommendation presents that “Genetic counseling reduces distress, improves risk perception, and reduces intention for testing.” However, now, interpretation of genetic test results has become more complex, and some results are simply uninterpretable given the current state of knowledge. The clients must understand that the result may not be helpful at all if it returns a mutation in a gene we know little about, or, more commonly, if it returns a variant that has not yet been classified as benign or deleterious. Now, we must create new genetic counseling strategy for prior to test counseling and post-test results interpretation.