Ordering the Correct Genetic Test: Implications for Oncology and Primary Care Healthcare Professionals

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Genetic testing for hereditary cancer syndromes is becoming increasingly more common. Once a mutation is detected in a family, other family members can undergo single-site mutation testing to determine if they have inherited the increased risk for developing cancer, with the intent of providing tailored and appropriate cancer prevention and early detection measures. Ordering the correct single-site test is critical to providing appropriate recommendations for cancer prevention and early detection.

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Genetic testing for cancer is becoming more and more common. Instead of being ordered by a credentialed genetics provider, genetic testing is now available through primary care providers and directly to the consumer through the Internet. However, significant safety considerations are associated with genetic testing. This article provides a case study that describes a family with a known mutation associated with hereditary nonpolyposis colorectal cancer (HNPCC) who had a potentially dangerous outcome because of incorrect single-site genetic testing. The case study is followed by commentary on issues related to single-site genetic testing and how to prevent such problems.

Case Study

An advanced practice credentialed genetic nurse (APNG) was contacted by J.M., a 26-year-old single mother, about obtaining genetic testing for a known family mutation in the *MSH2* gene, which is one of the genes associated with the HNPCC syndrome also known as Lynch syndrome. Such a mutation is associated with an 80% lifetime risk for developing colorectal cancer and a 70% chance of developing endometrial cancer, as well as other cancers (Lindor, McMaster, Lindor, & Greene, 2008). J.M. was uninsured and travelled more than 140 miles for the services, all of which were to be covered by charitable funding for the uninsured. The patient was carefully instructed prior to the appointment to bring a copy of the test results from any known relative who had tested positive for the mutation. J.M. said that she understood this request and would do so.

The patient arrived at the appointment with a copy of test results from a first cousin who had tested negative for the known mutation. The APNG explained that she would be unable to order the test that day because the policy was to order a test off of a positive result. J.M. was upset and frustrated because she had driven a great distance and said that her other relatives had simply gone to their doctors and had saliva tests ordered without any requirements. A pedigree was constructed, which J.M. perceived as unnecessary because a known mutation in the family had already been established. She was instructed that the pedigree was necessary for identifying other family members that might benefit from testing. Information from the pedigree helped determine that a paternal uncle had been the first to be tested and that J.M.'s father had died of colon cancer at age 57 years. Another living paternal aunt had tested negative, as did two female cousins (the daughters of the previously mentioned paternal uncle). The report that J.M. had brought was from one of the cousins.

The patient received pretest counseling on the implications of testing for this mutation and was informed that, assuming her father was an obligate carrier, she had a 50% chance of having the mutation. J.M. was instructed to return with a copy of the positive test result and the APNG explained that, although simply ordering the test and sending the specimen off of the negative test result J.M. brought to the appointment would be easier, the possibility that the wrong test would be ordered meant that it could not be done.

J.M. sent a copy of the positive test result about a week later. The uncle with the positive test result had a mutation in the *MSH2 1215* gene; however, the negative test result for J.M.'s cousin was determined to be for the *MSH2 1216* gene. The wrong test had been ordered for the cousin at some point, probably as a result of a copying error or that the handwriting on the test request form was illegible. The tests for the other cousin and the aunt were then reviewed and it became apparent that both of the cousins had undergone testing for the wrong mutation. The aunt's test had been correctly ordered. The mistake apparently occurred when one of the cousin's tests was incorrectly ordered from the aunt's report. The other cousin's test was ordered by a different primary care physician, but it was ordered off of her sister's