CLINICAL SUMMARY

Testing for Lynch Syndrome (HNPCC)

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Genetic Screening for Lynch Syndrome Predicted to Cost-Effectively Improve Health Outcomes

Individuals who inherit Lynch syndrome have up to an 80% lifetime risk for colorectal cancer and up to a 70% lifetime risk for endometrial cancer. Commonly underdiagnosed, Lynch syndrome is one of the most common hereditary cancer syndromes, occurring in approximately 1 in 440 individuals.

Dinh TA, et al. Health benefits and cost-effectiveness of primary genetic screening for Lynch syndrome in the general population. Cancer Prev Res 2011; 4(1): 9-22.

In current medical practice, genetic testing for Lynch syndrome is often limited to individuals with a diagnosis of cancer or in unaffected individuals with a known mutation in the family. Patients who are found to have mutations are offered increased surveillance and/or preventative surgeries, which have been shown to improve outcomes.

In another hereditary cancer syndrome with similar prevalence, Hereditary Breast and Ovarian Cancer (HBOC), which is caused by mutations in the *BRCA1/BRCA2* genes, genetic testing is regularly performed in both women with a diagnosis of cancer and also in women unaffected by cancer, who have a suspicious family history. The authors concluded that testing practices which include unaffected individuals with a suspicious family history should be applied to Lynch syndrome.

Methods:

Using a well-established Archimedes health care simulation model, two large virtual populations were analyzed using different genetic testing strategies: 1) one population using current clinical practice of primarily testing only individuals diagnosed with cancer; and 2) a second population using an experimental model of universal risk assessment for all patients (those with and without cancer) at different ages, followed by genetic testing of individuals at varying degrees of risk for carrying a mutation.

Results:

The optimal strategy was predicted to be universal risk assessment based on family history starting at age 25-35 and proceeding to genetic testing of individuals with risks of carrying a mutation in excess of 5% via established risk models.[1]

Despite this strategy being more liberal than current medical practice:

- This strategy was cost-effective
  - o $26,000 per Quality Adjusted Life Years (QALY) - well below the accepted healthcare cost effectiveness threshold of $50,000 per QALY and comparable to cost-effectiveness of other common cancer screening protocols
- This strategy predicted improved health outcomes for mutation carriers including
  - o significant decreases in cases of colorectal and endometrial cancers
  - o early detection of existing cancers

Bottom Line:

"This finding supports the concept that genetic screening of unaffected, at-risk individuals, when conducted in association with appropriate risk assessment, and when followed by surveillance for colorectal and endometrial cancer would cost-effectively improve health outcomes."

1. PREMM1,2,6 - http://www.dana-farber.org/pat/cancer/gastrointestinal/crc-calculator/new-calculator.asp
   MMRPRO - http://www4.utsouthwestern.edu/breasthealth/cagene/

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