Title **Molecular Diagnosis for Hereditary Cancer Predisposing Syndromes: Genetic Testing and Clinical Impact**

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Aim

- To assess the evidence regarding the availability, cost, and analytical and clinical validity of genetic tests for screening and diagnosis of hereditary cancer predisposing syndromes
- To document the impact of genetic testing (GT) on the clinical management of patients with specific hereditary cancer predisposing syndromes

Conclusions and results

The cost, availability, and analytical and clinical sensitivity of 20 hereditary cancer predisposing syndromes were listed in chart form in three distinct groupings:

- Where GT is generally part of clinical management of affected families
- Where the benefit of GT has been demonstrated, but GT is not part of standard management
- Where the clinical benefit of GT is unclear

A list of GT services and laboratories in Canada according to province and city was also compiled.

Although new molecular techniques are being developed rapidly, the implementation of GT for many disorders into standard clinical management has not been justified. High cost, variable analytical and clinical validity, limited availability, and legal/social/ethical issues affect integration of GT into the healthcare system.

Methods

To obtain published and unpublished literature, investigators searched electronic databases, contacted genetic test laboratories, and hand searched bibliographies of selected papers. Two reviewers independently selected 457 relevant articles that met the predetermined selection criteria.

Further research/reviews required

Additional data about cost and the impact of GT on health outcomes are needed to help determine appropriate integration and funding of these tests.

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