CADTH Evidence Driven.

IN BRIEF A Summary of the Evidence

Genetic Testing of Patients With a First, Unexplained Blood Clot: A Review

Key Messages

- Factor V Leiden and prothrombin gene mutations are genetic conditions that increase the risk of developing a blood clot. This can include a clot in the deep veins of the leg — called deep vein thrombosis (DVT). DVTs can break away and cause a blockage in another part of the body such as the lung — called a pulmonary embolus (PE), which can be life-threatening.
- Highly accurate, commercial tests are available to identify people who have factor V Leiden and prothrombin gene mutations but whether they are helpful in patients with a first, unexplained blood clot was uncertain.
- After reviewing the clinical and economic evidence, an expert panel recommended that patients with a first, unexplained blood clot should not be routinely tested for factor V Leiden or prothrombin mutations because there was no evidence to show that testing changes the way the clot is treated, improves health outcomes, or helps to predict the risk of having another clot. Testing does result in additional costs.
- Genetic tests for factor V Leiden and prothrombin mutations may be useful for other patient groups.

Context

There are many reasons why a blood clot might form when it shouldn't. These include trauma, surgery, immobilization (e.g., a long car or airplane trip), pregnancy, some medications, cancer, and other medical conditions. But some of us have gene mutations in our DNA that can affect the clotting factors in our blood. These mutations result in thrombophilia — a group of disorders that lead to an increased risk of blood clots. The most common of these mutations, factor V Leiden, affects the blood clotting protein factor V. A second mutation can affect factor II, another important clotting factor, which is also known as prothrombin.

Technology

Commercial tests are available in Canada to detect specific factor V and prothrombin mutations; i.e., factor V G1691A and prothrombin G20210A. These tests have been found to be highly accurate.

Issues

Currently across Canada, testing for factor V Leiden and prothrombin gene mutations in patients with a first, unexplained blood clot varies. It's not clear when the tests should be ordered and how the tests impact the way in which patients are managed or whether testing improves their health outcomes.

Methods

CADTH conducted a systematic review of the clinical evidence and performed a health economic analysis comparing testing with no testing. Based on these assessments and clinical expertise, CADTH's Health Technology Expert Review Panel (HTERP) developed recommendations for factor V Leiden and prothrombin gene mutation testing in patients with a first, unexplained clot.

Results

Findings from the systematic review showed that, even though there is a link between factor V Leiden and prothrombin mutations and first, unexplained blood clots, there was limited evidence to suggest they increase the risk of future clot recurrence. Evidence on whether testing influences patient management or clinical outcomes was sparse and of low quality. Clinical practice guidelines suggest that there is insufficient evidence to warrant changing the way patients are treated based on testing results. Taken together, it appears that routine testing for factor V Leiden and prothrombin gene mutations in patients with a first, unprovoked venous thromboembolism may have limited clinical effectiveness. The results of the cost analysis indicate that, based on the clinical results, not testing patients with a first, unexplained blood clot would result in cost savings without impacting patient care.

Read more about CADTH and its review of factor V Leiden and prothrombin mutation testing at:

www.cadth.ca/reports/factor-v-leiden-and-prothrombin-mutation-testing-patients-unprovoked-thromboembolism.

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