Central Zone

Pathology and Laboratory Medicine Memorandum

To: Nova Scotia Healthcare Providers and Health Service Directors

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Date: April 17, 2023

Subject: New heritable thrombophilia (Factor V Leiden gene mutation and Prothrombin gene mutation) genetic testing criteria

New heritable thrombophilia testing criteria will be implemented on **June 1, 2023** and genetic testing for Factor V Leiden (FVL) and Prothrombin gene mutation (PGM) will be restricted to patients seen in **Hematology, Neurology, Medical Genetics and Internal Medicine**. Individuals should receive appropriate pre- and post-test counselling and testing should only be ordered when it will impact clinical management. These new testing criteria were developed in accordance with national and international best practice guidelines, as well as Choosing Wisely recommendations.

In general, heritable thrombophilia (FVL, PGM) genetic testing is **NOT** recommended in the following settings:

- General population screening
- Asymptomatic adults and children including those with a family history of thrombophilia
- Adults with a venous thromboembolism (VTE) in the setting of major transient risk factors (surgery, trauma, etc.)
- Routine testing in adults with arterial thrombosis
- Individuals with unprovoked VTE receiving long-term anticoagulation therapy
- Before use of oral contraceptives, hormone replacement therapy, and/or other estrogen receptor modulators
- Prenatal or newborn testing
- Routine testing of children with a first episode of a VTE
- Women with recurrent pregnancy loss/stillbirths, fetal growth restriction, or preeclampsia

Please submit any questions to Dr. Tanya Gillan (tanya.gillan@nshealth.ca) or Dr. David Conrad (david.conrad@nshealth.ca)

References:

1. Choosing Wisely Canada – Hematology (https://choosingwiselycanada.org/recommendation/hematology/)
2. Choosing Wisely – American Society of Hematology (https://www.choosingwisely.org/societies/american-society-of-hematology/)
3. Arachchillage et al., (2022), Thrombophilia Testing: A British Society for Haematology Guideline. British Journal of Haematology PMID: 35645034).
4. Kovalenko et al., (2022), Confirming Choosing Wisely: Inpatient thrombophilia testing. Journal of Clinical Oncology, 40:28 (supp. pp22).
5. Bergstrom at al., (2020), Heritable thrombophilia test utilization and cost savings following guideline-based restrictions: An interrupted time series analysis. Thrombosis Research (PMID: 32330793).