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Shared Decision-Making Guide for Thrombophilia Testing

Introduction

To facilitate a shared decision-making approach to thrombophilia testing, the purpose of this guide is two-fold:

1. Empower patients to actively participate in their care.
2. Support healthcare providers in facilitating meaningful discussions with their patients about thrombophilia testing.

This guide highlights the value of working together to navigate the complexities and controversies surrounding thrombophilia testing and its role in the management of venous thromboembolism (VTE). By making informed decisions together, we aim to improve patient outcomes and ensure that testing decisions align with each patient's values and medical needs.

Understanding Thrombophilia

What is Thrombophilia?

Thrombophilia is a group of blood clotting disorders or conditions where certain proteins in the blood cause it to clot more easily than normal, affecting approximately 10% of the population. Some people are born with thrombophilia (inherited or genetic thrombophilia), while other people develop thrombophilia later in life (acquired thrombophilia).

Inherited Thrombophilia

- Inherited gene from one parent (heterozygous) or both parents (homozygous).

- The degree of clotting risk fluctuates based on thrombophilia type, whether inherited from one or both parents, and other risk factors present.
- The inherited thrombophilias include deficiencies of antithrombin, protein C, or protein S, and the mutations factor V Leiden (FVL) and prothrombin G20210A (PGM).

Acquired Thrombophilia

- Acquired as the result of a specific cause, such as injury, illness, surgery, and certain medical conditions such as cancer, paroxysmal nocturnal hemoglobinuria, myeloproliferative neosplasms or antiphospholipid syndrome (APS).

Testing for Thrombophilia

Thrombophilia Testing Guidelines

Healthcare providers refer to thrombophilia testing guidelines from professional organizations such as the American College of Chest Physicians (ACCP) and the American Society of Hematology (ASH) to guide their decision-making.

Indications for Testing

Thrombophilia testing is not generally recommended for routine screening or in situations where the results would not change the management plan.

American Society of Hematology (ASH) generally recommends thrombophilia testing in specific situations where it may impact clinical management or decision-making. These situations include:

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- **Non-surgical blood clots:** Test patients with blood clots caused by non-surgical risk factors, including pregnancy, postpartum, or use of birth control pills.

- **High-risk women with family history:** Individuals with a family history of blood clots and a known high-risk thrombophilia (antithrombin, protein C, or protein S deficiency), to determine if they need medication to prevent blood clots during periods of temporary risk, if hormone treatments – such as birth control pills – should be avoided, or if additional precautions are needed to prevent blood clots during and after pregnancy.

- **Unusual blood clot sites:** Patients with blood clots at unusual sites within the body, such as the brain or abdominal blood vessels.

- **Cancer patients with family history:** Test ambulatory cancer patients with a family history of blood clots to determine if they need preventive treatment for blood clots, even if they are at low or intermediate risk.

Pros of Thrombophilia Testing

- **Informed decision-making:** Helps make more informed decisions about managing risks related to blood clots, particularly in high-risk scenarios such as surgery, pregnancy, or prolonged immobilization.

- **Preventive measures:** Enables implementation of preventive measures such as lifestyle changes, medications, or special precautions during periods of increased risk such as surgery or pregnancy.

- **Family awareness:** Can inform family members of potential risks, leading to proactive health measures.

- **Family planning:** Can inform decisions related to contraception, pregnancy management, and potential preventative treatments to prevent complications like miscarriage or pregnancy-related blood clots.

- **Tailored anticoagulation:** Can help determine whether long-term anticoagulation is necessary for individuals with a history of VTE, particularly those with high-risk thrombophilias, such as antithrombin deficiency and antiphospholipid syndrome (APS).

- **Choice of blood thinner:** Certain conditions, such as APS and cancer, have been specifically studied to see which blood thinner is best for patients with these diseases.

- **Peace of mind:** For some patients, knowing their thrombophilia status can provide peace of mind or empower them to take proactive steps in managing their health.

Cons of Thrombophilia Testing

- **Limited impact on management:** Many times, results of thrombophilia testing will not change management (e.g., anticoagulation therapy is often guided by individual risk factors rather than thrombophilia test results).

- **Incomplete information:** Current tests only identify the few known types of thrombophilia. Many genetic factors remain unknown. Therefore, a negative test should not be interpreted as a patient being free of thrombophilia or provide a false sense of security, leading them to overlook other significant clotting risks.

- **Emotional impact:** Positive results can cause undue anxiety or stress for patients and their families, especially in cases where the risk of clotting is low or where the test result might not change the recommended treatment approach.

- **Missed diagnosis and over-diagnosis:** Like all medical tests, thrombophilia testing is not perfect, and false positives or negatives can occur, leading to the risk of false negatives (missed diagnosis) and false positives (over-diagnosis).

- **Coverage and cost:** Testing can be expensive, and insurance may not cover it unless it is considered medically necessary. Out-of-pocket costs can be substantial, which might deter some patients.

- **Insurance discrimination:** There are federal laws to help protect people from health insurance and job discrimination. However, these laws have limitations, and it is possible that a person with hereditary thrombophilia may have to pay higher premiums for health or disability insurance and/or get denied life or disability insurance.

Timing of Testing

Thrombophilia testing should not be performed at the time of a blood clot diagnosis or while a patient is currently receiving anticoagulation, as results may be inaccurate and difficult to interpret.

Perform testing (when indicated) after completion of initial therapy and if it might change management strategies. Wait two weeks after discontinuing warfarin, or two days for direct oral anticoagulants and heparin.

Pregnancy affects many clotting proteins and it is not ideal to test for thrombophilia during pregnancy or in the first few weeks/months after giving birth.

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Shared-Decision Making Approach

This guide utilizes a shared decision making (SDM) approach, which is a patient-centered care model where a patient and their healthcare provider work together to make decisions about their care. The goal is for patients to understand their options and have their preferences and goals used to help guide decisions. In the SDM model, tests and treatments should be selected based on evidence, the provider's knowledge and experience, and the patient's values and beliefs.

Approach to Conversation: Healthcare Providers to Patients

1. Introduction to Thrombophilia and Its Implications

- **Educate about thrombophilia:** Using patient-friendly language and visual aids, explain thrombophilia, distinguishing between hereditary and acquired types, and its significance in relation to blood clots.

- **Discuss implications:** Highlight the potential implications of identifying thrombophilia for patient care, including management and preventive measures.

2. Reviewing the ASH Guidelines and Recommendations

- **Summarize guidelines:** Provide an overview of the ASH guidelines, emphasizing the context in which thrombophilia testing is recommended or not.

- **Clarify evidence:** Provide transparency regarding basis of evidence for guideline recommendations (e.g. nearly all recommendations in the guidelines are based on very low evidence).

3. Personalizing the Decision-Making Process

- **Assess risk factors:** Evaluate the patient's risk factors for VTE, including family history of thrombophilia or blood clots, and any current or future situations that might increase clotting risk (e.g., surgery, pregnancy).

- **Consider patient values:** Take into account the patient's values and preferences, including their comfort with uncertainty, preference for knowing their genetic status, their feelings about prolonged anticoagulation therapy, and consider any cultural, racial, and ethnic differences in patient attitudes toward testing.

4. Discussing the Benefits and Risks of Testing

- **Explain potential outcomes:** Describe the potential outcomes of testing, including scenarios where testing might or might not change management.

- **Discuss psychological impact:** Address the potential psychological impact of knowing one's thrombophilia status and the implications for family members.

5. Exploring Patient Preferences and Concerns

- **Invite questions:** Encourage questions and express concerns about thrombophilia testing, blood clot risk, and the implications of the test results.

- **Explore readiness:** Discuss how much the patient wants to know about their genetic information and their readiness to deal with that knowledge.

6. Empowered Decision Making and Next Steps

- **Review of information:** Provide a summary of the complex information that has been discussed thus far regarding thrombophilia testing.
- **Collaborate on decision:** Work together to make a decision about thrombophilia testing that aligns with the patient's values, preferences, and clinical context.
- **Provide resources:** Offer additional information and support, including patient education materials, support groups, and genetic counseling if appropriate.
- **Give time:** Allow the patient time to think through their choices and offer support by answering additional questions. Then provide space for decision-making.
- **Plan follow-up:** Arrange follow-up discussions to review test results, if performed, and to discuss the next steps in management based on those results.

Conclusion

Engaging in a shared decision-making process for thrombophilia testing involves clear communication, understanding patient concerns, and discussing the benefits and limitations of testing. Before thrombophilia testing is ordered, the goals of the testing and the potential psychological effects should be discussed and understood. This approach ensures that patients are well-informed and can actively participate in their healthcare decisions.

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Resources

- American Society of Hematology VTE Guidelines: Thrombophilia Testing
- Thrombophilia Patient Resources - National Blood Clot Alliance
- Understanding Thrombophilia – National Blood Clot Alliance
- Online Blood Clot Patient Support Group
- ASH Teaching Slides for Thrombophilia Testing, PowerPoint | PDF
- Thrombophilia Testing | ASH Clinical Practice Guidelines on Venous Thromboembolism (VTE)
- Family Testing for Clotting Disorders – National Blood Clot Alliance

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GRIFOLS

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