



Test Definition: AATHI

Thrombophilia Profile Interpretation

Overview

Useful For

Interpretation of testing performed as part of a profile to evaluate patients with thrombosis or hypercoagulability states

Method Name

Only orderable as part of a profile. For more information see AATHR / Thrombophilia Profile, Plasma and Whole Blood.

Medical Interpretation

NY State Available

Yes

Specimen

Specimen Type

Plasma Na Cit

Reject Due To

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Plasma Na Cit	Frozen	14 days	

Clinical & Interpretive

Clinical Information

Thrombophilia is defined as an acquired or familial disorder associated with thrombosis. The clinical presentation of an underlying thrombophilia predominantly includes venous thromboembolism (deep vein thrombosis, pulmonary embolism, superficial vein thrombosis). Other manifestations that have been linked to thrombophilia include recurrent miscarriage and complications of pregnancy (eg, severe preeclampsia, abruptio placentae, intrauterine growth restriction, stillbirth). Thrombophilia does not predict arterial thrombosis. Demographic or environmental exposures that compound the risk of venous thromboembolism among persons with a thrombophilia include increasing age, male gender, obesity, surgery, trauma, hospitalization for medical illness, malignant neoplasm, prolonged immobility during travel (eg, prolonged airplane travel), oral contraceptive use, estrogen therapy (both oral and transdermal), tamoxifen and raloxifene therapy, and infertility drugs. Central venous catheters and transvenous pacemaker wires increase the risk for upper extremity deep vein thrombosis; this risk is unrelated to thrombophilia.

Inherited thrombophilias include:

-Deficiency due to reduced plasma protein level or dysfunctional protein of:

-Antithrombin

-Protein C

-Protein S

-Dysfibrinogenemias (rare)

-Activated protein C resistance due to the factor V Leiden variant (F5 c.1601G>A; p.Arg534Gln, previously R506Q)

-Prothrombin F2 c.*97Gly>Ala variant (previously G20210A)

Acquired thrombophilias include a lupus-like anticoagulant (antiphospholipid antibodies) and disseminated intravascular coagulation/intravascular coagulation and fibrinolysis (DIC/ICF). DIC/ICF may cause thrombotic as well as hemorrhagic events. Positive tests for DIC/ICF can also occur as consequences of thrombosis.

Acquired deficiencies of fibrinogen, protein C, protein S, and antithrombin may be found in conjunction with liver disease (they are produced by the liver) or DIC/ICF and are of uncertain significance with respect to thrombosis risk.

Acquired deficiencies of protein C and protein S are also found in patients with liver disease who are being treated with oral anticoagulants (eg, warfarin), since both of these proteins are dependent upon the action of vitamin K for normal function.

Acquired protein S deficiency also occurs in thrombotic thrombocytopenic purpura, pregnancy or estrogen therapy, nephrotic syndrome, and sickle cell anemia. In acute illness, the level of acute-phase reactants rise (including C4b binding protein, which binds and inactivates protein S in the plasma) and the portion of bound protein S also rises leaving a lower proportion of free protein S. The significance of acquired protein S deficiency with respect to thrombosis risk is unknown.

Reference Values

Only orderable as part of a profile. For more information see AATHR / Thrombophilia Profile, Plasma and Whole Blood.

An interpretive report will be provided.

Interpretation

An interpretive report will be provided when testing is completed, noting the presence or absence of thrombophilia.

Cautions

To obtain the most useful information, this testing is best performed in medically stable patients who are not receiving oral vitamin K inhibitor (eg, warfarin,), heparin, low-molecular-weight heparin, hirudin (Refludan), argatroban, fibrinolytic agents (eg, streptokinase, tissue plasminogen activator), or platelet GPIIb/IIIa (alpha IIb beta3) inhibitors (abx cimab [ReoPro], tirofiban [Aggrastat]). However, useful information can be obtained in patients receiving anticoagulation therapy.

Clinical Reference

1. Pengo V, Tripodi A, Reber G, et al. Update of the guidelines for lupus anticoagulant detection. Subcommittee on Lupus Anticoagulant/Antiphospholipid Antibody of the Scientific and Standardisation Committee of the International Society on Thrombosis and Haemostasis. *J Thromb Haemost.* 2009;7(10):1737-1740

- Keeling D, Mackie I, Moore GW, et al. Guidelines on the investigation and management of antiphospholipid syndrome. Br J Haematol. 2012;157(1):47-58
- Clinical and Laboratory Standards Institute (CLSI). Laboratory Testing for the Lupus Anticoagulant; Approved Guideline. CLSI document H60-A., CLSI; 2014
- Favaloro EJ, Lippi G, eds. Hemostasis and Thrombosis, Methods and Protocols. Humana Press; 2017

Performance

Method Description

A coagulation expert (clinician or hematopathologist) reviews the laboratory data and an interpretive report is issued.

PDF Report

No

Day(s) Performed

Monday through Friday

Report Available

4 to 7 days

Performing Laboratory Location

Mayo Clinic Laboratories - Rochester Main Campus

Fees & Codes

Fees

- Authorized users can sign in to [Test Prices](#) for detailed fee information.
- Clients without access to Test Prices can contact [Customer Service](#) 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact [Customer Service](#).

Test Classification

Not Applicable

CPT Code Information

85390-26 Special Coagulation Interpretation

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
AATHI	Thrombophilia Interpretation	69049-5

Result ID	Test Result Name	Result LOINC® Value
603184	Thrombophilia Interpretation	69049-5

603325	Reviewed by	18771-6
--------	-------------	---------