Medical Policy

Genetic Testing for Inherited Thrombophilia

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<tr>
<th>Type:</th>
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<td>Medical Necessity and Investigational / Experimental</td>
<td>Laboratory/Pathology</td>
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<th>Original Policy Date:</th>
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<tr>
<td>January 11, 2013</td>
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Definitions of Decision Determinations

Medically Necessary: A treatment, procedure or drug is medically necessary only when it has been established as safe and effective for the particular symptoms or diagnosis, is not investigational or experimental, is not being provided primarily for the convenience of the patient or the provider, and is provided at the most appropriate level to treat the condition.

Investigational/Experimental: A treatment, procedure or drug is investigational when it has not been recognized as safe and effective for use in treating the particular condition in accordance with generally accepted professional medical standards. This includes services where approval by the federal or state governmental is required prior to use, but has not yet been granted.

Split Evaluation: Blue Shield of California / Blue Shield of California Life & Health Insurance Company (Blue Shield) policy review can result in a Split Evaluation, where a treatment, procedure or drug will be considered to be investigational for certain indications or conditions, but will be deemed safe and effective for other indications or conditions, and therefore potentially medically necessary in those instances.

Description

Inherited thrombophilias are a group of clinical conditions in which there is a genetic variant defect associated with a predisposition for venous thromboembolism (VTE). However, not all patients with a genetic predisposition to thrombosis will develop VTE. The presence of inherited thrombophilia will presumably interact with other VTE risk factors to determine an individual's
overall risk of VTE. Some of the more common risk factors for thromboembolic events include surgery, obesity, immobility, pregnancy, hormone replacement therapy (HRT), along with a variety of hereditary factors.

According to Bauer and Lip (2011) the most common type of inherited thrombophilia is a factor V Leiden mutation, which accounts for up to 50% of the inherited thrombophilia syndromes. Some of the more common conditions associated with inherited thrombophilias include; activated protein C resistance, prothrombin gene mutation, protein C deficiency, protein S deficiency, prothrombin deficiency and hyper-homocysteinemia (MTHFR mutations).

Policy

Genetic testing for inherited thrombophilia may be considered **medically necessary** during pregnancy when **any** of the following conditions are present:

- Prior history of venous thromboembolism during pregnancy or while on birth control pills
- History of idiopathic venous thromboembolism with no identifiable underlying cause
- History of venous thromboembolism at an unusual site (e.g., cerebral, mesenteric, portal, hepatic)
- Family history of Factor V Leiden mutation

Genetic testing for factor V Leiden mutations, prothrombin gene mutations, and mutations in the MTHFR gene, is considered **investigational** for other indications, including, but not limited to, other possible inheritable forms of thrombophilia and medical management of venous thromboembolism.

Policy Guideline

Specific CPT codes for this testing became available in 2012:

- 81240: F2 (prothrombin, coagulation factor II)(e.g., hereditary hypercoagulability) gene analysis, 20210G>A variant
- 81241: F5 (coagulation Factor V)(e.g., hereditary hypercoagulability) gene analysis, Leiden variant
- 81291: MTHFR (5, 10-methylenetetrahydrofolate reductase)(e.g., hereditary hypercoagulability) gene analysis, comm.
- on variants (e.g., 677T, 1298C)

**Documentation Required for Clinical Review**

- History and physical including:
  - Progress notes regarding patient's history with venous thromboembolism (VTE)
• Documentation of family history of Factor V Leiden mutation (if applicable)

The materials provided to you are guidelines used by this plan to authorize, modify, or deny care for persons with similar illness or conditions. Specific care and treatment may vary depending on individual need and the benefits covered under your contract. These Policies are subject to change as new information becomes available.