

Date: February 8, 2019

Test Update: MTHFR testing is now a sendout test

Dear Regional Pathology Services Clients,

MTHFR polymorphisms test is now a sendout test. It has been recommended that MTHFR polymorphism testing no longer be performed clinically, with the exception of rare populations, as there is no demonstrable clinical utility for thrombophilia risk (1).

The MTHFR mutations do not accurately predict elevated serum homocysteine levels or risk of elevated homocysteine. When elevated homocysteine is present with other thrombophilia risk factors the thrombosis risk is increased. In the absence of other thrombophilia risk factors, elevated homocysteine is primarily predictive of atherosclerotic disease.

Factor V Leiden (G1691A; R506Q) and Prothrombin (G20210A) mutations will now be performed with an FDA-approved assay. Factor V Leiden and Prothrombin Mutation testing can still be ordered as individual tests or as a combined order. The Thrombophilia Mutation Panel (THMP) will be discontinued.

Specimen requirements will remain the same (5.0 mL EDTA, lavender, 0.5 mL minimum). Turnaround time will be 3-5 working days.

Interfaced Clients. Please add these codes if not already built:

Test Name	LIS Code
Prothrombin/Leiden mutation	PRLEY
Prothrombin gene mutation only	PROTH
Factor 5 (V) Leiden mutation only	LEYDN
MTHFR New Sendout code	MTHR

Please Inactivate codes MTHF and THMP (Thrombophilia mutation panel). In addition, The HYPCGP (Hpercoagulable Panel) needs to be updated by removing code THMP and adding PROTH and LEYDN. If you have any questions about the interface build please contact RPSinterfacesupport@unmc.edu or call Andi Hudson at 402-559-7670.

1)Hickey, S.E *et al.* ACMG Practice Guideline: lack of evidence for *MTHFR* polymorphism testing. *Genetics in Medicine* volume 15, pages 153–156 (2013).

If you would like to discuss this further with your client representative please contact client services at 402-559-6420 and ask to speak with one of the client coordinators.

24/7 Client Services 402-559-6420