HYPERHOMOCYSTEINEMIA

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- Independent risk factor for venous thromboembolism (VTE).
- Risk of VTE is 2.9-fold when homocysteine levels above the 95th percentile

MTHFR POLYMORPHISM

- 34-37% of the Caucasian population in the U.S. are heterozygous for MTHFR C667T
- 12% of Caucasians in the U.S. are homozygous for the MTHFR polymorphism
- MTHFR C667T is not associated with a significant increase in homocysteine levels
- Homozygosity for this common MTHFR polymorphism is associated with a **slightly** higher plasma homocysteine level
- Double heterozygosity with MTHFR C667T is also associated with a **slightly** higher plasma homocysteine level
- MTHFR polymorphism is **NOT** itself an independent risk factor for arterial or venous thromboembolism

TESTING FOR HYPERHOMOCYSTEINEMIA

- Assess the level of plasma homocysteine as part of the workup for inherited thrombophilia
- Testing for MTHFR C677T, is not recommended as part of the work up for inherited thrombophilia
- Identifying individuals that have MTHFR polymorphisms does not have clinical relevance and does not provide an opportunity for intervention since we cannot change individuals' DNA

TREATMENT FOR HYPERHOMOCYSTEINEMIA

- Treatment is based on the level of homocysteine in the blood as this is best indicator of hematologic risk
- Elevated homocysteine levels can be altered via folic acid, vitamins B6, and B12 supplementation