PROS1 gene

protein S

Normal Function

The *PROS1* gene provides instructions for making a protein called protein S that is important for the regulation of blood clotting. Although protein S can work on its own to inhibit certain clotting factors, it often works as a cofactor. Cofactors enhance the function of proteins that speed up chemical reactions (enzymes).

Protein S is made by cells in the liver. The protein circulates in the bloodstream in two forms; it is either attached (bound) to a specific protein, or it occurs by itself in a free form. The free form of protein S can act as a cofactor for an enzyme called activated protein C (APC). APC turns off (inactivates) the blood clotting proteins known as factor Va and factor VIIIa. Protein S also helps a protein called tissue factor pathway inhibitor (TFPI) block the activity of another clotting protein, factor Xa. This inactivation slows down the clotting process so that blood flow is not impaired.

Health Conditions Related to Genetic Changes

Protein S deficiency

Hundreds of variants (also called mutations) in the *PROS1* gene have been found to cause protein S deficiency. People with this condition have an increased risk of developing blood clots. Most of these variants change a single DNA building block (base pair) in the *PROS1* gene.

Some variants in the *PROS1* gene have been found to reduce the amount of available protein S. Other variants impair the function of protein S. Individuals with protein S deficiency may not have enough functional protein S to inactivate clotting proteins, which increases the risk of developing blood clots.

Other Names for This Gene

- PROS
- protein S, alpha

- PS21
- PS22
- PS23
- PS24
- PS25
- PSA
- THPH5
- THPH6
- vitamin K-dependent plasma protein S

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of PROS1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5627[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28PROS1%5BTI%5D%29+OR+%28protein+S%5BTI%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- PROTEIN S; PROS1 (https://omim.org/entry/176880)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/5627)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=PROS1[gene])

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Genomic Location

The *PROS1* gene is found on chromosome 3 (https://medlineplus.gov/genetics/chromosome/3/).

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