Factor V Mutation Symptoms

== Symptoms and signs == 6f4b5241fa Point mutations usually take place during DNA replication. DNA replication occurs when one double-stranded DNA molecule creates two single strands of DNA, each of which is a template for the creation of the complementary strand. A single point mutation can change the whole DNA sequence. Changing one purine or pyrimidine may change the amino acid that the nucleotides code for. 6f4b5241fa The variant causes elevated plasma prothrombin levels (hyperprothrombinemia), possibly due to increased pre-mRNA stability. Prothrombin is the precursor to thrombin, which plays a key role in causing blood to clot (blood coagulation). G20210A can thus contribute to a state of hypercoagulability, but not particularly... 6f4b5241fa High-risk mutations, which disable an important error-free DNA repair process (homology directed... 6f4b5241fa

Prothrombin G20210A It increases the risk of blood clots including from deep vein thrombosis, and of pulmonary embolism. Most people never develop a blood clot in their lifetimes. is also known as factor II, the mutation is also sometimes referred to as the factor II mutation or simply the prothrombin mutation; in either case, the Prothrombin G20210A is a genetic variation associated with increased blood coagulation (thrombophilia). Two copies increases the risk to up to 20 in 1,000 per year. One copy of the mutation increases the risk of a blood clot from 1 in 1,000 per year to 2. 5 in 1,000 6f4b5241fa

Prothrombin G20210A was identified in the 1990s. About 2% of Caucasians carry the variant, while it is less common in other populations. It is estimated to have originated in Caucasians about 24,000 years ago. 6f4b5241fa The condition may be inherited or, more commonly, acquired. 6f4b5241fa Point mutations may arise from spontaneous mutations that occur during DNA replication. The rate of mutation may be increased by mutagens. Mutagens can be physical, such as radiation from UV rays, X-rays or extreme heat, or chemical (molecules that misplace base pairs or disrupt the helical shape of DNA)... 6f4b5241fa While in congenital disease symptoms may be present at birth or show up later, in patients with acquired... 6f4b5241fa == Signs and symptoms == 6f4b5241fa Symptoms may differ greatly, as apparently modifiers control to some degree the amount of FX that is

produced. Some affected individuals have few or no symptoms while others may experience life-threatening bleeding. Typically this bleeding disorder manifests itself as a tendency to easy bruising, nose bleeding, heavy and prolonged menstruation and bleeding during pregnancy and childbirth, and excessive bleeding after dental or surgical interventions. Newborns may bleed in the head, from the umbilicus, or excessively after circumcision. Other bleeding can be encountered in muscles or joints, brain, gut, or urine 6f4b5241fa

TNF receptor associated periodic syndrome Individuals with TRAPS have episodic symptoms such as recurrent high fevers, rash, abdominal pain, joint/muscle aches and puffy eyes. is a periodic fever syndrome associated with mutations in a receptor for the molecule tumor necrosis factor (TNF) that is inheritable in an autosomal dominant TNF receptor associated periodic syndrome (TRAPS) is a periodic fever syndrome associated with mutations in a receptor for the molecule tumor necrosis factor (TNF) that is inheritable in an autosomal dominant manner 6f4b5241fa

BRCA mutation Harmful mutations in these genes may produce a hereditary breast–ovarian cancer syndrome in affected persons. that any cancer that appears was caused by the mutation, rather than some other factor. Women with harmful mutations in either BRCA1 or BRCA2 have a risk of breast cancer that is about five times the normal risk, and a risk of ovarian cancer that is about ten to thirty times normal. High-risk mutations, which disable an important error-free DNA repair A BRCA mutation is a mutation in either of the BRCA1 and BRCA2 genes, which are tumour suppressor genes. Having a high-risk mutation does not guarantee that the woman will develop cancer, nor does it imply that any cancer that appears was caused by the mutation, rather than some other factor. Only 5–10% of breast cancer cases in women are attributed to BRCA1 and BRCA2 mutations (with BRCA1 mutations being slightly more common than BRCA2 mutations), but the impact on women with the gene mutation is more profound. Hundreds of different types of mutations in these genes have been identified, some of which have been determined to be harmful, while others have no proven impact. The risk of breast and ovarian cancer is higher for women with a high-risk BRCA1 mutation than with a BRCA2 mutation 6f4b5241fa

== Signs and symptoms == 6f4b5241fa Frameshift mutations are apparent in severe genetic diseases such as Tay–Sachs disease; they

increase susceptibility to certain cancers and classes of familial hypercholesterolaemia; in... 6f4b5241fa There is no specific treatment for most thrombophilias, but recurrent episodes of thrombosis may be an indication for long-term preventive anticoagulation. The first major form of thrombophilia to be identified by medical science, antithrombin deficiency, was identified in 1965, while the most common abnormalities (including factor V Leiden) were described in the 1990s. 6f4b5241fa == Causes == 6f4b5241fa The gene for Factor I in humans is located on chromosome 4. Factor I is synthesized mostly in the liver, but also in monocytes, fibroblasts, keratinocytes, and endothelial cells. When synthesized, it is a 66kDa polypeptide chain with N-linked glycans at 6 positions. Then, factor I is cleaved by furin to yield the mature factor I protein, which is a disulfide-linked dimer of heavy chain (residues 19-335, 51 kDalton) and light chain (residues 340-583, 37 kDalton). Only the mature protein is active. 6f4b5241fa It is due to a specific gene mutation in which a guanine is changed to an adenine at position 20210 of the DNA of the prothrombin gene. Other blood clotting pathway mutations that increase the risk of clots include factor V Leiden. 6f4b5241fa == Signs and symptoms == 6f4b5241fa

Glycogen storage disease type V Its incidence is reported as one in 100,000, roughly the same as glycogen storage disease type I. of life, frequently due to misdiagnosis and dismissal of symptoms. The median age of symptom onset is 3 years, with the median diagnostic delay being Glycogen storage disease type V (GSD5, GSD-V), also known as McArdle's disease, is a metabolic disorder, one of the metabolic myopathies, more specifically a muscle glycogen storage disease, caused by a deficiency of myophosphorylase 6f4b5241fa

Point mutation A point mutation is a genetic mutation where a single nucleotide base is changed, inserted or deleted from a DNA or RNA sequence of an organism's genome A point mutation is a genetic mutation where a single nucleotide base is changed, inserted or deleted from a DNA or RNA sequence of an organism's genome. Point mutations have a variety of effects on the downstream protein product—consequences that are moderately predictable based upon the specifics of the mutation. g. synonymous mutations) to deleterious effects (e. frameshift mutations), with regard to protein production, composition, and function. These consequences can range from no effect (e. g 6f4b5241fa

De novo mutation A de novo mutation is a newly present mutation in an individual organism. These may occur in gametogenesis due to a germline mutation in a parent, or A de novo mutation is a newly present mutation in an individual organism. These may occur in gametogenesis due to a germline mutation in a parent, or as a postzygotic mutation 6f4b5241fa

Complement factor I It is a soluble glycoprotein that circulates in human blood at an average concentration of 35 µg/mL. Dysregulated factor I activity has clinical implications. Loss of function mutations in the Complement Complement factor I, also known as C3b/C4b inactivator, is a protein that in humans is encoded by the CFI gene. structure of human Factor I has been deposited as PDB: 2XRC. Complement factor I (factor I) is a protein of the complement system, first isolated in 1966 in guinea pig serum, that regulates complement activation by cleaving cell-bound or fluid phase C3b and C4b 6f4b5241fa

== Types == 6f4b5241fa

Factor X deficiency Produced in the liver, FX, when activated, cleaves prothrombin to generate thrombin in the intrinsic pathway of coagulation. This process is vitamin K dependent and enhanced by activated factor V. enhanced by activated factor V. [citation needed] The condition may be inherited or, more commonly, acquired. [citation needed] Symptoms may differ greatly Factor X deficiency (X as Roman numeral ten) is a bleeding disorder characterized by a lack in the production of factor X (FX), an enzyme protein that causes blood to clot in the coagulation cascade 6f4b5241fa

Frameshift mutation The earlier in the sequence the deletion or insertion occurs, the more altered the protein. Due to the triplet nature of gene expression by codons, the insertion or deletion can change the reading frame (the grouping of the codons), resulting in a completely different translation from the original. A frameshift mutation will in general cause the reading of the codons after the mutation to code for different amino acids. The frameshift mutation will also alter the first stop codon ("UAA", "UGA" or "UAG") encountered in the sequence. The polypeptide being created could be abnormally short or abnormally long, and will most likely not be functional. A frameshift mutation is not the same as a single-nucleotide polymorphism in which a nucleotide is replaced, rather than inserted or deleted. A frameshift mutation (also called a framing error or a reading frame shift) is a genetic mutation caused by indels

(insertions or deletions) of a number A frameshift mutation (also called a framing error or a reading frame shift) is a genetic mutation caused by indels (insertions or deletions) of a number of nucleotides in a DNA sequence that is not divisible by three 6f4b5241fa

TNF receptor associated periodic syndrome presents with the following signs and symptoms: 6f4b5241fa The disease was first reported in 1951 by British physician Brian McArdle of Guy's Hospital, London. 6f4b5241fa

Thrombophilia Such abnormalities can be identified in 50% of people who have an episode of thrombosis (such as deep vein thrombosis in the leg) that was not provoked by other causes. A significant proportion of the population has a detectable thrombophilic abnormality, but most of these develop thrombosis only in the presence of an additional risk factor. The most common ones are factor V Leiden (a mutation in the F5 gene at position 1691) and prothrombin G20210A, a mutation in prothrombin (at position Thrombophilia (sometimes called hypercoagulability or a prothrombotic state) is an abnormality of blood coagulation that increases the risk of thrombosis (blood clots in blood vessels) 6f4b5241fa

== Signs and symptoms == 6f4b5241fa == Synthesis == 6f4b5241fa

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