USB1 gene

U6 snRNA biogenesis phosphodiesterase 1

Normal Function

The *USB1* gene provides instructions for making an enzyme that functions as an RNA exonuclease. RNA exonucleases cut off (cleave) building blocks called nucleotides one at a time from molecules of RNA (a chemical cousin of DNA). This process helps stabilize the RNA and protects it from damage.

Specifically, the USB1 enzyme protects a small RNA molecule called U6, which is an essential component of a complex called a spliceosome. The USB1 enzyme also helps transport (chaperones) U6 to the spliceosome and helps it attach (bind) to the proteins in the complex. Spliceosomes process RNA molecules called messenger RNAs (mRNAs) by recognizing and removing regions known as introns and splicing the mRNA molecules back together to provide the blueprint for making proteins.

Different versions (isoforms) of the USB1 enzyme are produced in different tissues, where they play various roles. In blood-forming tissues, the USB1 enzyme is thought to be important for the maturation of neutrophils. Neutrophils are a type of white blood cell involved in the immune system. In the skin, the USB1 enzyme is found in pigment-producing cells (melanocytes), cells in the outer layer of the skin called keratinocytes, and structural cells called fibroblasts. Its role in the function of these cells is unknown.

Health Conditions Related to Genetic Changes

Poikiloderma with neutropenia

At least 24 mutations in the *USB1* gene have been identified in people with poikiloderma with neutropenia (PN). This condition involves a group of skin abnormalities called poikiloderma and a persistent shortage (deficiency) of neutrophils (chronic neutropenia).

The *USB1* gene mutations that cause PN are thought to lead to an enzyme whose function is impaired. As a result of the dysfunctional USB1 exonuclease, the U6 RNA is not protected from damage and not correctly chaperoned to the spliceosomes, leading to impairment of key biological functions. The specific connection between *USB1* gene mutations and the signs and symptoms of PN is unknown. However, the existence of tissue-specific isoforms of the enzyme could help explain why this disorder mainly

affects the skin and immune system.

Other Names for This Gene

- C16orf57
- chromosome 16 open reading frame 57
- hMPN1: mutated in PN1
- U six biogenesis 1
- U6 small nuclear RNA biogenesis phosphodiesterase 1
- U6 snRNA biogenesis 1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of USB1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=79650[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28USB1%5BTIAB%5D%29+OR+%28U6+snRNA+biogenesis+phosphodiesterase+1%5BTIAB%5D%29%29+OR+%28%28C16orf57%5BTIAB%5D%29+OR+%28HVSL+motif+containing+1%5BTIAB%5D%29+OR+%28Mpn1%5BTIAB%5D%29+OR+%28U+six+biogenesis+1%5BTIAB%5D%29+OR+%28U6+snRNA+biogenesis+1%5BTIAB%5D%29+OR+%28mutated+in+poikiloderma+with+neutropenia+protein+1%5BTIAB%5D%29+OR+%28putative+U6+snRNA+phosphodiesterase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D)

Catalog of Genes and Diseases from OMIM

- U6 SMALL NUCLEAR RNA BIOGENESIS PHOSPHODIESTERASE 1; USB1 (https://omim.org/entry/613276)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/79650)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=USB1[gene])

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Genomic Location

The *USB1* gene is found on chromosome 16 (https://medlineplus.gov/genetics/chromosome/16/).

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