

MLPH gene

melanophilin

Normal Function

The *MLPH* gene provides instructions for making a protein called melanophilin. This protein is found in pigment-producing cells called melanocytes, where it helps transport structures called melanosomes. These structures produce a pigment called melanin, which is the substance that gives skin, hair, and eyes their color (pigmentation). Melanophilin interacts with proteins produced from the *MYO5A* and *RAB27A* genes to form a complex that transports melanosomes to the outer edges of melanocytes. From there, the melanosomes are transferred to other types of cells, where they provide the pigment needed for normal hair, skin, and eye coloring.

Health Conditions Related to Genetic Changes

Griscelli syndrome

At least one mutation in the *MLPH* gene has been found to cause Griscelli syndrome. This genetic change causes a form of the condition designated type 3, which is characterized by unusually light (hypopigmented) skin and silvery-gray hair. Unlike the other forms of Griscelli syndrome, type 3 does not involve abnormalities of the brain or immune system.

The known *MLPH* gene mutation changes a single protein building block (amino acid) in the melanophilin protein. Specifically, it replaces the amino acid arginine with the amino acid tryptophan at protein position 35 (written as Arg35Trp or R35W). This mutation reduces the amount of melanophilin produced; the small amount of protein that is produced is unable to form a complex with the proteins made from the *MYO5A* and *RAB27A* genes. Without this complex, melanosomes cannot be transported to the edges of melanocytes. Instead, they clump near the center of melanocytes, trapping melanin within these cells and preventing normal pigmentation of skin and hair.

Other Names for This Gene

- exophilin-3
- I(1)-3Rk
- I1Rk3

- MELPH_HUMAN
- Slac-2a
- SLAC2-A
- slp homolog lacking C2 domains a
- synaptotagmin-like protein 2a

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of MLPH ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=79083\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=79083[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28MLPH%5BTIAB%5D%29+OR+%28melanophilin%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>)

Catalog of Genes and Diseases from OMIM

- MELANOPHILIN; MLPH (<https://omim.org/entry/606526>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/79083>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=MLPH\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=MLPH[gene]))

References

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Genomic Location

The *MLPH* gene is found on chromosome 2 (<https://medlineplus.gov/genetics/chromosome/2/>).

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