

SLURP1 gene

secreted LY6/PLAUR domain containing 1

Normal Function

The *SLURP1* gene provides instructions for making a protein called secreted Ly6/uPAR-related protein-1 (SLURP-1). This protein is found in skin cells and other cells that line the surfaces and cavities of the body. Like other Ly6/uPAR-related proteins, SLURP-1 folds into a particular shape and is thought to attach (bind) to other proteins called receptors to carry out signaling within cells. However, SLURP-1's role in the skin and the rest of the body is not completely understood.

Laboratory studies show that SLURP-1 can bind to nicotinic acetylcholine receptors (nAChRs). SLURP-1 specifically interacts with the alpha7 ($\alpha 7$) subunit, which is a piece of some nAChRs. Nicotinic acetylcholine receptors are best known for their role in chemical signaling between nerve cells, but they are also found in other tissues. In the skin, nAChRs regulate the activity of genes involved in the growth and division (proliferation), maturation (differentiation), and survival of cells. Through its interaction with these receptors, SLURP-1 may be involved in skin growth and development.

Health Conditions Related to Genetic Changes

Mal de Meleda

At least 15 mutations in the *SLURP1* gene have been found to cause mal de Meleda, a rare disorder characterized by tough, thickened skin on the hands and feet. On the palms and soles, the thickening is known as palmoplantar keratoderma; the thickened skin also extends to the backs of the hands and feet and up to the wrists and ankles. The *SLURP1* gene mutations involved in this condition lead to production of an altered SLURP-1 protein that is unstable and quickly broken down, if any protein is produced at all. As a result, affected individuals have little or no SLURP-1 protein. It is unclear how a lack of this protein leads to the skin problems that occur in mal de Meleda. Researchers speculate that without SLURP-1, the activity of genes controlled by nAChR signaling is altered, leading to overgrowth of skin cells or survival of cells that normally would have died. The excess of cells can result in skin thickening. It is unclear why skin on the hands and feet is particularly affected.

Other Names for This Gene

- anti-neoplastic urinary protein
- ANUP
- ARS
- ARS(component B)-81/S
- ArsB
- LY6LS
- lymphocyte antigen 6-like secreted
- MDM
- secreted Ly-6/uPAR-related protein 1
- secreted Ly-6/uPAR-related protein 1 precursor
- secreted Ly6/uPAR related protein 1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SLURP1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=57152\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=57152[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28SLURP1%5BTIAB%5D%29+OR+%28Ly6/uPAR-related+protein+1%29+OR+%28ARS+B%29+OR+%28SLURP-1%29+OR+%28ARS+component+B%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+3600+days%22%5Bdp%5D>)

Catalog of Genes and Diseases from OMIM

- SECRETED LY6/PLAUR DOMAIN-CONTAINING PROTEIN 1; SLURP1 (<https://omim.org/entry/606119>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/57152>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=SLURP1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=SLURP1[gene]))

References

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Genomic Location

The *SLURP1* gene is found on chromosome 8 (<https://medlineplus.gov/genetics/chromosome/8/>).

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