

SH2D1A gene

SH2 domain containing 1A

Normal Function

The *SH2D1A* gene provides instructions for making a protein called signaling lymphocyte activation molecule (SLAM) associated protein (SAP). SAP interacts with other proteins called SLAM family receptors to activate signaling pathways that are involved in the control of immune cells (lymphocytes). In particular, it helps regulate lymphocytes that destroy other cells (cytotoxic lymphocytes) and is necessary for the development of specialized lymphocytes called natural killer T cells. SAP also helps control immune reactions by triggering self-destruction (apoptosis) of lymphocytes when they are no longer needed.

Health Conditions Related to Genetic Changes

X-linked lymphoproliferative disease

More than 70 *SH2D1A* gene mutations have been identified in people with X-linked lymphoproliferative disease (XLP). Some *SH2D1A* gene mutations impair SAP function. Others result in an abnormally short protein that is unstable or nonfunctional, or prevent any SAP from being produced. The loss of functional SAP disrupts proper control of the immune system and may result in the life-threatening immune reaction to Epstein-Barr virus infection that occurs in this disorder. In addition, cancers of immune system cells (lymphomas) may develop in affected individuals when defective lymphocytes are not properly destroyed by apoptosis.

Other Names for This Gene

- DSHP
- Duncan disease SH2-protein
- EBVS
- MTCP1
- SAP
- SH2 domain-containing protein 1A
- SH21A_HUMAN
- signaling lymphocyte activation molecule-associated protein

- SLAM-associated protein
- XLP
- XLPD

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SH2D1A ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4068\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4068[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28SH2D1A%5BTIAB%5D%29+OR+%28%28LYP%5BTIAB%5D%29+OR+%28SAP%5BTIAB%5D%29+OR+%28XLP%5BTIAB%5D%29+OR+%28DSHP%5BTIAB%5D%29+OR+%28EBVS%5BTIAB%5D%29+OR+%28XLPD%5BTIAB%5D%29+OR+%28MTCP1%5BTIAB%5D%29+OR+%28SLAM-associated+protein%5BTIAB%5D%29+OR+%28signaling+lymphocyte+activation+molecule-associated+protein%5BTIAB%5D%29+OR+%28signaling+lymphocytic+activation+molecule-associated+protein%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+AND+%22last+360+days%22%5Bdp%5D>)

Catalog of Genes and Diseases from OMIM

- SH2 DOMAIN PROTEIN 1A; SH2D1A (<https://omim.org/entry/300490>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/4068>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=SH2D1A\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=SH2D1A[gene]))

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Genomic Location

The *SH2D1A* gene is found on the X chromosome (<https://medlineplus.gov/genetics/chromosome/x/>).

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