# SAP/SH2D1A Knockout Mice: A Model for X-linked Lymphoproliferative Disease

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## Technology description

## Summary

NIH announces the availability for licensing of SAP/SH2D1A knockout mice, which can be used as a model for X-linked lymphoproliferative disease (XLP), and exploited to design therapeutics or gene-therapy for XLP. These knockout mice can be used as well to study other T cell-mediated diseases, such as asthma and hypersensitivity, involving Th2 cells. This model is also useful for researchers interested in T-cell signaling and cytokine production by T-helper cells.
SAP (SLAM-associated protein) is a small lymphocyte-specific signaling molecule that is defective or absent in patients with XLP. SAP has unusually high affinity for SLAM (also called CD150) and has been suggested to function by blocking binding of SHP-2 or other SH2-containing signaling proteins to SLAM receptors. SAP has also been shown to be required for recruitment and activation of the Src-family kinase FynT after SLAM ligation, where the SAP SH2 domain binds to the SH3 domain of FynT and directly couples FynT to SLAM.
Mutations in the SH2D1 A gene on the Xq24-26 chromosome are known to be responsible for many cases of X-linked Lymphoproliferative syndrome.

## Institution

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