

# Recombinant Human ADSL

Catalog No: #GP10968



Package Size: #GP10968-1 100ug

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## Description

Product Name	Recombinant Human ADSL
Brief Description	Recombinant Protein
Immunogen Description	Fusion protein corresponding to a region derived from 235-484 amino acids of human ADSL
Target Name	adenylosuccinate lyase
Other Names	ASL; AMPS; ASASE
Accession No.	Swissprot:P30566Gene Accession:BC000253
Uniprot	P30566
GeneID	158;
Storage	-20~-80°C, pH 7.6 PBS

## Background

The protein encoded by this gene belongs to the lyase 1 family. It is an essential enzyme involved in purine metabolism, and catalyzes two non-sequential reactions in the de novo purine biosynthetic pathway: the conversion of succinylaminoimidazole carboxamide ribotide (SAICAR) to aminoimidazole carboxamide ribotide (AICAR) and the conversion of adenylosuccinate (S-AMP) to adenosine monophosphate (AMP). Mutations in this gene are associated with adenylosuccinase deficiency (ADSLD), a disorder marked with psychomotor retardation, epilepsy or autistic features. Alternatively spliced transcript variants have been found for this gene.

## References

Note: For in vitro research use only, not for diagnostic or therapeutic use. This product is not a medical device.

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