ADSL Lysate – Tissue Lysate

PRODUCT OVERVIEW

Product Name	ADSL, Lysate
Full Product Name	Human ADSL Protein Lysate
Product Gene Name	ADSL lysate
Product Synonym Names	ADSL; AMPS; ASASE; ASL
Host	Human
Form/Format	Lyophilized
Preparation and Storage	Store at -20 degree C Once reconsituted, aliquot and store any remaining material at-20 degree C to avoid repeated freeze-thaw cycles.
Molecular Weight	48,328 Da
OMIM	103050
3D Structure	ModBase 3D Structure for P30566
Related Product Information for ADSL lysate	Human ADSL Protein Lysate 20ug is provided as a Lyophilized powder. This is a Recombinant Protein Lysate produced in HEK293T cells. This protein lysate can be reconsituted using SDS Sample Buffer.
Other Notes	Small volumes of ADSL lysate vial(s) may occasionally become entrapped in the seal of the product vial during shipment and storage. If necessary, briefly centrifuge the vial on a tabletop centrifuge to dislodge any liquid in the container`s cap. Certain products may require to ship with dry ice and additional dry ice fee may apply.
Searchable Terms for ADSL purchase	MBS136961 is a lysate targeting ADSL. Also known as Human ADSL Protein Lysate. Host: Human.

ADDITIONAL DETAILS

NCBI/Uniprot data below describe general gene information for ADSL. It may not necessarily be applicable to this product.

NCBI GI #	4557269
NCBI GeneID	158
NCBI Accession #	NP_000017
UniProt Primary Accession #	P30566
UniProt Secondary Accession #	O75495; Q5TI34; B0QY76
UniProt Related Accession #	P30566
GenBank Nucleotide #	NM_000026
NCBI Official Full Name	adenylosuccinate lyase isoform a
NCBI Official Synonym Full Names	adenylosuccinate lyase
NCBI Official Symbol	ADSL
NCBI Official Synonym Symbols	ASL; AMPS; ASASE
UniProt Protein Name	Adenylosuccinate lyase
UniProt Synonym Protein Names	Adenylosuccinase; ASase

Protein Family	Adenylosuccinate lyase
UniProt Gene Name	ADSL
UniProt Synonym Gene Names	AMPS; ASL; ASase
UniProt Entry Name	PUR8_HUMAN
NCBI Protein Information	adenylosuccinate lyase
NCBI Summary for ADSL	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. [provided by RefSeq, Dec 2015]
UniProt Comments for ADSL	ADSL: Defects in ADSL are the cause of adenylosuccinase deficiency (ADSL deficiency). ADSL deficiency is an autosomal recessive disorder characterized by the accumulation in the body fluids of succinylaminoimidazole-carboxamide riboside (SAICA-riboside) and succinyladenosine (S-Ado). Most children display marked psychomotor delay, often accompanied by epilepsy or autistic features, or both, although some patients may be less profoundly retarded. Occasionally, growth retardation and muscular wasting are also present. Belongs to the lyase 1 family. Adenylosuccinate lyase subfamily. 2 isoforms of the human protein are produced by alternative splicing. Protein type: EC 4.3.2.2; Nucleotide Metabolism - purine; Lyase; Amino Acid Metabolism - alanine, aspartate and glutamate Chromosomal Location of Human Ortholog: 22q13.2 Cellular Component: cytosol Molecular Function: adenylosuccinate lyase activity Biological Process: AMP biosynthetic process; purine ribonucleoside monophosphate biosynthetic process; metabolic process; nucleobase, nucleoside and nucleotide metabolic process; purine nucleotide biosynthetic process; protein tetramerization; purine base metabolic process; 'de novo' IMP biosynthetic process Disease: Adenylosuccinase Deficiency
Research Articles on ADSL	**1.** Case Report: Malaysian patient compound heterozygous for two novel ADSL mutations giving rise to adenylosuccinate lyase deficiency.
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