abcam

Product datasheet

Recombinant Human ALAS2/ASB protein ab79941

1 Image

Description

Product name	Recombinant Human ALAS2/ASB protein
Purity	> 90 % SDS-PAGE.
Expression system	Escherichia coli
Protein length	Protein fragment
Animal free	No
Nature	Recombinant
Species	Human
Amino acids	136 to 553
Tags	His tag N-Terminus

Specifications

Our **Abpromise guarantee** covers the use of **ab79941** in the following tested applications.

The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.

Applications	SDS-PAGE
Form	Liquid
Additional notes	This product was previously labelled as ALAS2

Preparation and Storage

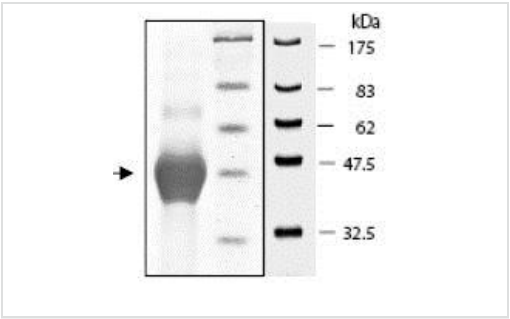
Stability and Storage	Shipped on Dry Ice. Upon delivery aliquot. Store at -80°C. Avoid freeze / thaw cycle. pH: 8.00 Constituents: 0.0462% (R*,R*)-1,4-Dimercaptobutan-2,3-diol, 0.395% Tris HCl, 0.05% Tween, 20% Glycerol (glycerin, glycerine), 0.58% Sodium chloride, 0.00053% PLP

General Info

Tissue specificity	Erythroid specific.

Pathway	Porphyrin metabolism; protoporphyrin-IX biosynthesis; 5-aminolevulinate from glycine: step 1/1.
Involvement in disease	Defects in ALAS2 are a cause of anemia sideroblastic X-linked (XLSA) [MIM:300751]. Sideroblastic anemia is characterized by anemia of varying severity, hypochromic peripheral erythrocytes, systemic iron overload secondary to chronic ineffective erythropoiesis, and the presence of bone marrow ringed sideroblasts. Sideroblasts are characterized by iron-loaded mitochondria clustered around the nucleus. XLSA shows a variable hematologic response to pharmacologic doses of pyridoxine. Defects in ALAS2 are the cause of erythropoietic protoporphyria X-linked dominant (XLDPT) [MIM:300752]. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. XLDPT is a form of porphyria characterized biochemically by a high proportion of zinc-protoporphyrin in erythrocytes, in which a mismatch between protoporphyrin production and the heme requirement of differentiating erythroid cells leads to overproduction of protoporphyrin in amounts sufficient to cause photosensitivity and liver disease. Note=Gain of function mutations in ALS2 are responsible for XLDPT, but they can also be a possible aggravating factor in congenital erythropoietic porphyria and other erythropoietic disorders caused by mutations in other genes (PubMed:21309041).
Sequence similarities	Belongs to the class-II pyridoxal-phosphate-dependent aminotransferase family.
Cellular localization	Mitochondrion matrix.

Images



SDS-PAGE - Recombinant Human ALAS2/ASB protein (ab79941)

105 SDS-PAGE showing ab79941 at approximately 46kDa (8µg).

Please note: All products are "FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC PROCEDURES"

Our Abpromise to you: Quality guaranteed and expert technical support

- Replacement or refund for products not performing as stated on the datasheet
- Valid for 12 months from date of delivery
- Response to your inquiry within 24 hours
- We provide support in Chinese, English, French, German, Japanese and Spanish
- Extensive multi-media technical resources to help you
- We investigate all quality concerns to ensure our products perform to the highest standards

If the product does not perform as described on this datasheet, we will offer a refund or replacement. For full details of the Abpromise, please visit **https://www.abcam.com/abpromise** or contact our technical team.

Terms and conditions

- Guarantee only valid for products bought direct from Abcam or one of our authorized distributors