

Product datasheet

Recombinant Human ALAS2/ASB protein ab79941

画像数 1

製品の詳細

製品名	Recombinant Human ALAS2/ASB protein
精製度	> 90 % SDS-PAGE.
発現系	Escherichia coli
タンパク質長	Protein fragment
Animal free	No
由来	Recombinant
生物種	Human
領域	136 to 553
タグ	His tag N-Terminus

特性

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.

アプリケーション	SDS-PAGE
製品の状態	Liquid
備考	This product was previously labelled as ALAS2

前処理および保存

保存方法および安定性	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
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関連情報

組織特異性	Erythroid specific.
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パスウェイ

Porphyrin metabolism; protoporphyrin-IX biosynthesis; 5-aminolevulinate from glycine: step 1/1.

関連疾患

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]. Porphyrins 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).

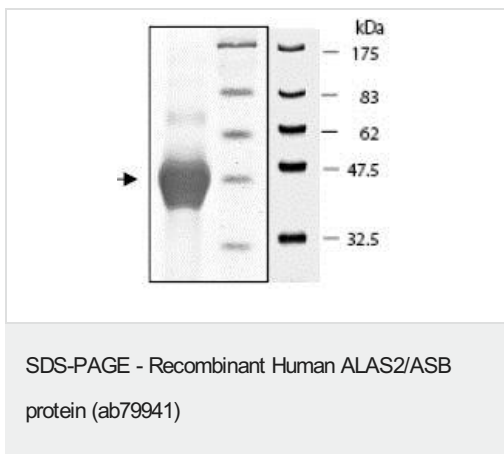
配列類似性

Belongs to the class-II pyridoxal-phosphate-dependent aminotransferase family.

細胞内局在

Mitochondrion matrix.

画像



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

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

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