

Recombinant Human L2HGDH protein ab127239

製品の詳細

製品名	Recombinant Human L2HGDH protein
精製度	> 90 % SDS-PAGE. Purified via His tag
発現系	Escherichia coli
アクセッション番号	<u>Q9H9P8</u>
タンパク質長	Protein fragment
Animal free	No
由来	Recombinant
生物種	Human
予測される分子量	17 kDa
領域	65 to 216
タグ	His tag N-Terminus

特性

Our **Abpromise guarantee** covers the use of **ab127239** 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	Lyophilized

前処理および保存

保存方法および安定性	Shipped at 4°C. Store at -20°C. Constituents: 0.32% Tris HCl, 0.58% Sodium chloride
再構成	Reconstitute with water to desired concentration.

関連情報

組織特異性	Widely expressed. Highly expressed in brain, testis and muscle. Expressed to a lower extent in lymphocytes, fibroblasts, keratinocytes, placenta, bladder, small intestine, liver and bone marrow.
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関連疾患	Defects in L2HGDH are the cause of L-2-hydroxyglutaric aciduria (L2HGA) [MIM:236792]. L2HGA is a rare autosomal recessive disorder clinically characterized by mild psychomotor delay in the first years of life, followed by progressive cerebellar ataxia, dysarthria and moderate to severe mental retardation. Diagnosis is based on the presence of an excess of L-2-hydroxyglutaric acid in urine, blood and cerebrospinal fluid.
配列類似性	Belongs to the L2HGDH family.
細胞内局在	Mitochondrion.

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

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