abcam

Product datasheet

Recombinant Human PAH protein ab159048

1 Abreviews 1 Image

Description

Product name	Recombinant Human PAH protein
Expression system	Wheat germ
Protein length	Full length protein
Animal free	No
Nature	Recombinant
Species	Human
Sequence	MSTAVLENPGLGRKLSDFGQETSYIEDNCNQNGAISLIFSL KEEVGALAK VLRLFEENDVNLTHIESRPSRLKKDEYEFFTHLDKRSLPAL TNIIKILRH DIGATVHELSRDKKKDTVPWFPRTIQELDRFANQILSYGAE LDADHPGFK DPVYRARRKQFADIAYNYRHGQPIPRVEYMEEGKKTWGTV FKTLKSLYKT HACYEYNHIFPLLEKYCGFHEDNIPQLEDVSQFLQTCTGFR LRPVAGLLS SRDFLGGLAFRVFHCTQYIRHGSKPMYTPEPDICHELLGH VPLFSDRSFA QFSQEIGLASLGAPDEYIEKLATIYWFTVEFGLCKQGDSIKA YGAGLLSS FGELQYCLSEKPKLLPLELEKTAIQNYTVTEFQPLYYVAES FNDAKEKVR NFAATIPRPFSVRYDPYTQRIEVLDNTQQLKILADSINSEIGIL CSALQK IK
Amino acids	1 to 452
Tags	GST tag N-Terminus

Specifications

Our **Abpromise guarantee** covers the use of **ab159048** in the following tested applications.

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

Applications	Western blot
	ELISA

Form	Liquid
Additional notes	

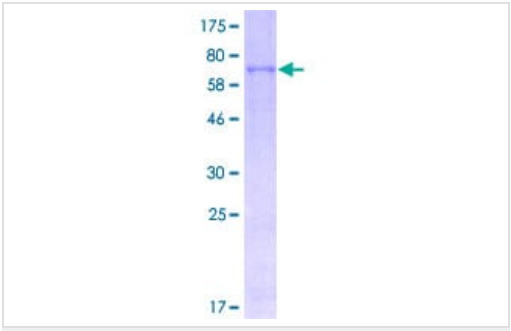
Preparation and Storage

Stability and Storage	Shipped on dry ice. Upon delivery aliquot and store at -80ºC. Avoid freeze / thaw cycles. pH: 8.00 Constituents: 0.31% Glutathione, 0.79% Tris HCl

General Info

Pathway	Amino-acid degradation; L-phenylalanine degradation; acetoacetate and fumarate from L-phenylalanine: step 1/6.
Involvement in disease	Defects in PAH are the cause of phenylketonuria (PKU) [MIM:261600]. PKU is an autosomal recessive inborn error of phenylalanine metabolism, due to severe phenylalanine hydroxylase deficiency. It is characterized by blood concentrations of phenylalanine persistently above 1200 mumol (normal concentration 100 mumol) which usually causes mental retardation (unless low phenylalanine diet is introduced early in life). They tend to have light pigmentation, rashes similar to eczema, epilepsy, extreme hyperactivity, psychotic states and an unpleasant 'mousy' odor. Defects in PAH are the cause of non-phenylketonuria hyperphenylalaninemia (Non-PKU HPA) [MIM:261600]. Non-PKU HPA is a mild form of phenylalanine hydroxylase deficiency characterized by phenylalanine levels persistently below 600 mumol, which allows normal intellectual and behavioral development without treatment. Non-PKU HPA is usually caused by the combined effect of a mild hyperphenylalaninemia mutation and a severe one. Defects in PAH are the cause of hyperphenylalaninemia (HPA) [MIM:261600]. HPA is the mildest form of phenylalanine hydroxylase deficiency.
Sequence similarities	Belongs to the biopterin-dependent aromatic amino acid hydroxylase family. Contains 1 ACT domain.

Images



SDS-PAGE - Recombinant Human PAH protein (ab159048)

ab159048 on a 12.5% SDS-PAGE stained with Coomassie Blue.

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