Recombinant Human PCSK9 (Asp374Tyr) Protein, C-His

Summary

Catalog No.	EHJ24002
Alternative Names	Proprotein convertase subtilisin/kexin type 9, Subtilisin/kexin-like protease PC9, NARC-1, PC9, Proprotein convertase 9, Neural apoptosis-regulated convertase 1, NARC1, PCSK9
Form	Lyophilized
Storage buffer	Lyophilized from a solution in PBS pH 7.4, 1mM EDTA, 4% Trehalose, 1% Mannitol.
Purity	>90% as determined by SDS-PAGE.
Applications	ELISA, Immunogen, SDS-PAGE, WB, Bioactivity testing in progress
Endotoxin level	Please contact with the lab for this information.
Expression system	Mammalian Cells
Accession	Q8NBP7
Protein length	Met1-Gln692 (D374Y)
Nature	Recombinant
Predicted molecular weight	75.30 kDa
Stability and Storage	Use a manual defrost freezer and avoid repeated freeze thaw cycles. Store at 2 to 8°C for frequent use. Store at -20 to -80°C for twelve months from the date of receipt.
Reconstitution	Reconstitute in sterile water for a stock solution.A copy of datasheet will be provided with the products, please refer to it for details.

Species	Homo sapiens (Human)
Shipping	In general, proteins are provided as lyophilized powder/frozen liquid. They are shipped out with dry ice/blue ice unless customers require otherwise.
Note	For research use only.

Description

Genetic variation in PCSK9 has an enormous impact on LDL-C concentration in humans and both gain-of-function (GOF) and loss-of-function (LOF) PCSK9 mutations have been described. While PCSK9 LOF mutations cause hypocholesterolemia, GOF mutations are a rare cause of familial hypercholesterolemia (FH), a monogenic disease characterized by very high levels of LDL-C and premature atherosclerotic cardiovascular disease (ASCVD). PCSK9 GOF mutations are causative of FH, because the enhancement in PCSK9 function leads to increased LDLr degradation and reduced recycling to the cell surface. As a consequence, there is a reduction in LDL uptake and an increase in circulating LDL-C concentration. The best characterized PCSK9 GOF mutation is p.(Asp374Tyr) which produces a ten-fold increase in LDLr degradation by increasing the binding affinity of PCSK9 to the epidermal growth factor-like domain of LDLr. This variant was demonstrated to inhibit LDL uptake still at a concentration 25 times lower than the wild-type PCSK9.

Data Image