Human FGF14 / SCA27 Protein (isoform 1B)

Catalog Number: **13654-HNAE**

General Information

Gene Name Synonym:

FGF-14; FHF-4; FHF4; SCA27

Protein Construction:

A DNA sequence encoding the human FGF14 isoform 1B (NP_787125.1) (Lys 64-Thr 252) was expressed and purified.

Source: Human

Expression Host: E. coli

QC Testing

Purity: > 97 % as determined by SDS-PAGE

Bio Activity:

Measured by its ability to bind human FGFR4-Fc (Cat:10538-H02H) in a functional ELISA.

Endotoxin:

Please contact us for more information.

Stability:

Samples are stable for up to twelve months from date of receipt at -70 ℃

Predicted N terminal: Met

Molecular Mass:

The recombinant human FGF14 consisting of 190 amino acids and has a calculated molecular mass of 21.1KDa. It migrates as an approximately 18KDa band in SDS-PAGE under reducing conditions.

Formulation:

Lyophilized from sterile PBS, pH 7.5

Normally 5 % - 8 % trehalose, mannitol and 0.01% Tween80 are added as protectants before lyophilization. Specific concentrations are included in the hardcopy of COA. Please contact us for any concerns or special requirements.

Usage Guide

Storage:

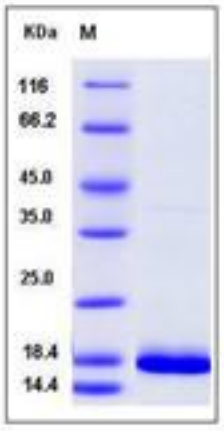
Store it under sterile conditions at -20℃ to -80℃ upon receiving. Recommend to aliquot the protein into smaller quantities for optimal storage.

Avoid repeated freeze-thaw cycles.

Reconstitution:

Detailed reconstitution instructions are sent along with the products.

SDS-PAGE:



Protein Description

FGF14 is a member of the fibroblast growth factor (FGF) family. Members of this family possess broad mitogenic and cell survival activities, and are involved in a variety of biological processes, including embryonic development, cell growth, morphogenesis, tissue repair, tumor growth and invasion. FGF14 is probably involved in nervous system development and function. Defects in FGF14 are the cause of spinocerebellar ataxia type 27 (SCA27). It is a clinically and genetically heterogeneous group of cerebellar disorders. Patients show progressive incoordination of gait and often poor coordination of hands, speech and eye movements, due to degeneration of the cerebellum with variable involvement of the brainstem and spinal cord. SCA27 is an autosomal dominant cerebellar ataxia. It is a slowly progressive disorder, with onset in late-childhood to early adulthood, characterized by ataxia with tremor, orofacial dyskinesia, psychiatric symptoms and cognitive deficits.

References

1.Wang Q, *et al*. (2002) Ataxia and paroxysmal dyskinesia in mice lacking axonally transported FGF14. Neuron. 35 (1): 25-38. 2.Zhao Y, *et al*. (2007) Genetic analysis of SCA 27 in ataxia and childhood onset postural tremor. Am J Med Genet. 144B (3): 395-6. 3.Lou JY, *et al*. (2005) Fibroblast growth factor 14 is an intracellular modulator of voltage-gated sodium channels. J Physiol. 569 (1): 179-93.