abcam

Product datasheet

Recombinant human FGFR1 protein ab60853

2 Images

Overview

Product name	Recombinant human FGFR1 protein
Protein length	Protein fragment

Description

Nature	Recombinant
Source	Insect cells

Amino Acid Sequence

Species	Human
Amino acids	399 to 822
Tags	GST tag N-Terminus

Specifications

Our Abpromise guarantee covers the use of **ab60853** in the following tested applications.

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

Applications	SDS-PAGE Functional Studies
Form	Liquid
Additional notes	ab204877 (Poly (4:1 Glu, Tyr) peptide) can be utilized as a substrate for assessing kinase activity

Preparation and Storage

Stability and Storage	Shipped on dry ice. Upon delivery aliquot and store at -80ºC. Avoid freeze / thaw cycles. Preservative: None Constituents: 25% Glycerol, 50mM Tris HCl, 150mM Sodium chloride, 0.25mM DTT, 0.1mM EGTA, 0.1mM EDTA, 0.1mM PMSF, pH 7.5 This product is an active protein and may elicit a biological response in vivo, handle with caution.

General Info

Function

Receptor for basic fibroblast growth factor. Receptor for FGF23 in the presence of KL (By similarity). A shorter form of the receptor could be a receptor for FGF1 (aFGF).

Tissue specificity

Detected in astrocytoma, neuroblastoma and adrenal cortex cell lines. Some isoforms are detected in foreskin fibroblast cell lines, however isoform 17, isoform 18 and isoform 19 are not detected in these cells.

Involvement in disease

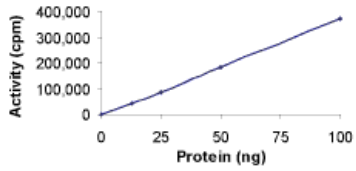
Defects in FGFR1 are a cause of Pfeiffer syndrome (PS) [MIM:101600]; also known as acrocephalosyndactyly type V (ACS5). PS is characterized by craniosynostosis (premature fusion of the skull sutures) with deviation and enlargement of the thumbs and great toes, brachymesophalangy, with phalangeal ankylosis and a varying degree of soft tissue syndactyly.
Defects in FGFR1 are a cause of idiopathic hypogonadotropic hypogonadism (IHH) [MIM:146110]. IHH is defined as a deficiency of the pituitary secretion of follicle-stimulating hormone and luteinizing hormone, which results in the impairment of pubertal maturation and of reproductive function.
Defects in FGFR1 are the cause of Kallmann syndrome type 2 (KAL2) [MIM:147950]; also known as hypogonadotropic hypogonadism and anosmia. Anosmia or hyposmia is related to the absence or hypoplasia of the olfactory bulbs and tracts. Hypogonadism is due to deficiency in gonadotropin-releasing hormone and probably results from a failure of embryonic migration of gonadotropin-releasing hormone-synthesizing neurons. In some cases, midline cranial anomalies (cleft lip/palate and imperfect fusion) are present and anosmia may be absent or inconspicuous.
Defects in FGFR1 are the cause of osteoglophonic dysplasia (OGD) [MIM:166250]; also known as osteoglophonic dwarfism. OGD is characterized by craniosynostosis, prominent supraorbital ridge, and depressed nasal bridge, as well as by rhizomelic dwarfism and nonossifying bone lesions. Inheritance is autosomal dominant.
Defects in FGFR1 are the cause of trigonocephaly non-syndromic (TRICEPH) [MIM:190440]; also known as metopic craniosynostosis. The term trigonocephaly describes the typical keel-shaped deformation of the forehead resulting from premature fusion of the frontal suture. Trigonocephaly may occur also as a part of a syndrome.
Note=A chromosomal aberration involving FGFR1 may be a cause of stem cell leukemia lymphoma syndrome (SCLL). Translocation t(8;13)(p11;q12) with ZMYM2. SCLL usually presents as lymphoblastic lymphoma in association with a myeloproliferative disorder, often accompanied by pronounced peripheral eosinophilia and/or prominent eosinophilic infiltrates in the affected bone marrow.
Note=A chromosomal aberration involving FGFR1 may be a cause of stem cell myeloproliferative disorder (MPD). Translocation t(6;8)(q27;p11) with FGFR1OP. Insertion ins(12;8)(p11;p11p22) with FGFR1OP2. MPD is characterized by myeloid hyperplasia, eosinophilia and T-cell or B-cell lymphoblastic lymphoma. In general it progresses to acute myeloid leukemia. The fusion proteins FGFR1OP2-FGFR1, FGFR1OP-FGFR1 or FGFR1-FGFR1OP may exhibit constitutive kinase activity and be responsible for the transforming activity.
Note=A chromosomal aberration involving FGFR1 may be a cause of stem cell myeloproliferative disorder (MPD). Translocation t(8;9)(p12;q33) with CEP110. MPD is characterized by myeloid hyperplasia, eosinophilia and T-cell or B-cell lymphoblastic lymphoma. In general it progresses to acute myeloid leukemia. The fusion protein CEP110-FGFR1 is found in the cytoplasm, exhibits constitutive kinase activity and may be responsible for the transforming activity.

Sequence similarities

Belongs to the protein kinase superfamily. Tyr protein kinase family. Fibroblast growth factor receptor subfamily.
Contains 3 Ig-like C2-type (immunoglobulin-like) domains.

	Contains 1 protein kinase domain.
Post-translational modifications	Binding of FGF1 and heparin promotes autophosphorylation on tyrosine residues and activation of the receptor.
Cellular localization	Membrane. Nucleus. Cytoplasm. Cytoplasmic vesicle

Images



Functional Studies - FGFR1 protein (Cytoplasmic domain) (ab60853)

Sample Kinase Activity Plot.

SDS-PAGE - FGFR1 protein (Cytoplasmic domain) (ab60853)

ab60853 on SDS-PAGE, MW ~73 kDa.

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