

Product datasheet

Recombinant Human FGFR3 protein ab158438

1 Image

Overview

Product name	Recombinant Human FGFR3 protein
Protein length	Protein fragment

Description

Nature	Recombinant
Source	Wheat germ
Amino Acid Sequence	
Species	Human
Sequence	TEQRVVGRAAEVPGPEPGQQEQLVFGSGDAVELSCP PPGGGPMGPTVWVK DGTGLVPSERVLVGPQRLQVLNASHEDSGAYSCRQR LTQRVLCHF SVRVT
Amino acids	27 to 126
Tags	GST tag N-Terminus

Specifications

Our [Abpromise guarantee](#) covers the use of **ab158438** 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	Protein concentration is above or equal to 0.05 mg/ml.

Preparation and Storage

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

Function

Receptor for acidic and basic fibroblast growth factors. Preferentially binds FGF1.

Tissue specificity

Expressed in brain, kidney and testis. Very low or no expression in spleen, heart, and muscle. In 20- to 22-week old fetuses it is expressed at high level in kidney, lung, small intestine and brain, and to a lower degree in spleen, liver, and muscle. Isoform 2 is detected in epithelial cells. Isoform 1 is not detected in epithelial cells. Isoform 1 and isoform 2 are detected in fibroblastic cells.

Involvement in disease

Defects in FGFR3 are the cause of achondroplasia (ACH) [MIM:100800]. ACH is an autosomal dominant disease and is the most frequent form of short-limb dwarfism. It is characterized by a long, narrow trunk, short extremities, particularly in the proximal (rhizomelic) segments, a large head with frontal bossing, hypoplasia of the midface and a trident configuration of the hands. Defects in FGFR3 are the cause of Crouzon syndrome with acanthosis nigricans (CAN) [MIM:612247]. Classic Crouzon disease which is caused by mutations in the FGFR2 gene is characterized by craniosynostosis (premature fusion of the skull sutures), and facial hypoplasia. Crouzon syndrome with acanthosis nigricans (a skin disorder characterized by pigmentation anomalies), CAN, is considered to be an independent disorder from classic Crouzon syndrome. CAN is characterized by additional more severe physical manifestation, such as Chiari malformation, hydrocephalus, and atresia or stenosis of the choanas, and is caused by a specific mutation (Ala-391 to Glu) in the transmembrane domain of FGFR3. It is proposed to have an autosomal dominant mode of inheritance.

Defects in FGFR3 are a cause of thanatophoric dysplasia type (TD) [MIM:187600, 187601]; also known as thanatophoric dwarfism or platyspondylic lethal skeletal dysplasia Sand Diego type (PLSD-SD). TD is the most common neonatal lethal skeletal dysplasia. Affected individuals display features similar to those seen in homozygous achondroplasia. It causes severe shortening of the limbs with macrocephaly, narrow thorax and short ribs. In the most common subtype, TD1, femur are curved, while in TD2, straight femurs are associated with cloverleaf skull. Mutations affecting different functional domains of FGFR3 cause different forms of this lethal disorder.

Defects in FGFR3 are a cause of hypochondroplasia (HCH) [MIM:146000]. HCH is an autosomal dominant disease and is characterized by disproportionate short stature. It resembles achondroplasia, but with a less severe phenotype.

Defects in FGFR3 are a cause of susceptibility to bladder cancer (BLC) [MIM:109800]. A malignancy originating in tissues of the urinary bladder. It often presents with multiple tumors appearing at different times and at different sites in the bladder. Most bladder cancers are transitional cell carcinomas. They begin in cells that normally make up the inner lining of the bladder. Other types of bladder cancer include squamous cell carcinoma (cancer that begins in thin, flat cells) and adenocarcinoma (cancer that begins in cells that make and release mucus and other fluids). Bladder cancer is a complex disorder with both genetic and environmental influences. Note=Somatic mutations can constitutively activate FGFR3.

Defects in FGFR3 are a cause of cervical cancer (CERCA) [MIM:603956]. A malignant neoplasm of the cervix, typically originating from a dysplastic or premalignant lesion previously present at the active squamocolumnar junction. The transformation from mild dysplastic to invasive carcinoma generally occurs slowly within several years, although the rate of this process varies widely.

Carcinoma in situ is particularly known to precede invasive cervical cancer in most cases. Cervical cancer is strongly associated with infection by oncogenic types of human papillomavirus.

Defects in FGFR3 are the cause of campodactyly tall stature and hearing loss syndrome (CATSHL syndrome) [MIM:610474]. CATSHL syndrome is an autosomal dominant syndrome characterized by permanent and irreducible flexion of one or more fingers of the hand and/or feet, tall stature, scoliosis and/or a pectus excavatum, and hearing loss. Affected individuals have developmental delay and/or mental retardation, and several of these have microcephaly.

Radiographic findings included tall vertebral bodies with irregular borders and broad femoral metaphyses with long tubular shafts. On audiological exam, each tested member have bilateral

sensorineural hearing loss and absent otoacoustic emissions. The hearing loss was congenital or developed in early infancy, progressed variably in early childhood, and range from mild to severe. Computed tomography and magnetic resonance imaging reveal that the brain, middle ear, and inner ear are structurally normal.

Defects in *FGFR3* are a cause of multiple myeloma (MM) [MIM:254500]. MM is a malignant tumor of plasma cells usually arising in the bone marrow and characterized by diffuse involvement of the skeletal system, hyperglobulinemia, Bence-Jones proteinuria and anemia. Complications of multiple myeloma are bone pain, hypercalcemia, renal failure and spinal cord compression. The aberrant antibodies that are produced lead to impaired humoral immunity and patients have a high prevalence of infection. Amyloidosis may develop in some patients. Multiple myeloma is part of a spectrum of diseases ranging from monoclonal gammopathy of unknown significance (MGUS) to plasma cell leukemia. Note=A chromosomal aberration involving *FGFR3* is found in multiple myeloma. Translocation t(4;14)(p16.3;q32.3) with the IgH locus.

Defects in *FGFR3* are a cause of lacrimo-auriculo-dento-digital syndrome (LADDS) [MIM:149730]; also known as Levy-Hollister syndrome. LADDS is a form of ectodermal dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. LADDS is an autosomal dominant syndrome characterized by aplastic/hypoplastic lacrimal and salivary glands and ducts, cup-shaped ears, hearing loss, hypodontia and enamel hypoplasia, and distal limb segments anomalies. In addition to these cardinal features, facial dysmorphism, malformations of the kidney and respiratory system and abnormal genitalia have been reported. Craniosynostosis and severe syndactyly are not observed.

Defects in *FGFR3* are a cause of keratinocytic non-epidermolytic nevus (KNEN) [MIM:162900]; also known as pigmented moles. Epidermal nevi of the common, non-organoid and non-epidermolytic type are benign skin lesions and may vary in their extent from a single (usually linear) lesion to widespread and systematized involvement. They may be present at birth or develop early during childhood.

Defects in *FGFR3* are a cause of Muenke syndrome (MNKS) [MIM:602849]; also known as Muenke non-syndromic coronal craniosynostosis. MNKS is a condition characterized by premature closure of coronal suture of skull during development (coronal craniosynostosis), which affects the shape of the head and face. It may be uni- or bilateral. When bilateral, it is characterized by a skull with a small antero-posterior diameter (brachycephaly), often with a decrease in the depth of the orbits and hypoplasia of the maxillae. Unilateral closure of the coronal sutures leads to flattening of the orbit on the involved side (plagiocephaly). The intellect is normal. In addition to coronal craniosynostosis some affected individuals show skeletal abnormalities of hands and feet, sensorineural hearing loss, mental retardation and respiratory insufficiency.

Defects in *FGFR3* are a cause of keratosis seborrheic (KERSEB) [MIM:182000]. A common benign skin tumor. Seborrheic keratoses usually begin with the appearance of one or more sharply defined, light brown, flat macules. The lesions may be sparse or numerous. As they initially grow, they develop a velvety to finely verrucous surface, followed by an uneven warty surface with multiple plugged follicles and a dull or lackluster appearance.

Sequence similarities

Belongs to the protein kinase superfamily. Tyr protein kinase family. Fibroblast growth factor receptor subfamily.

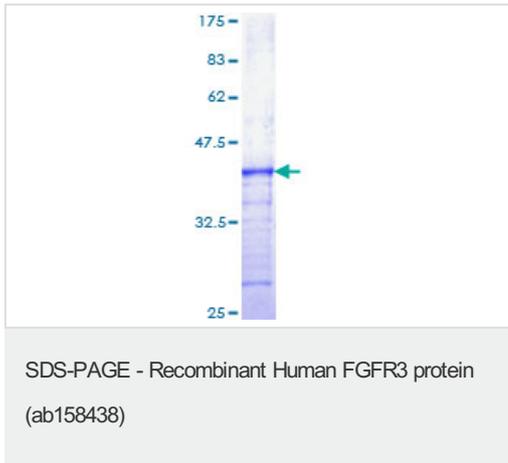
Contains 3 Ig-like C2-type (immunoglobulin-like) domains.

Contains 1 protein kinase domain.

Cellular localization

Membrane.

Images



ab158438 on a 12.5% SDS-PAGE stained with Coomassie Blue.

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