abcam

Product datasheet

Recombinant human FGFR2 (mutated C491F) protein (Active) ab268563

2 Images

Description

Product name	Recombinant human FGFR2 (mutated C491F) protein (Active)
Biological activity	The specific activity of ab268563 was 253 nmol/min/mg in a kinase assay using Poly (4:1 Glu, Tyr) synthetic peptide as substrate.
Purity	> 90 % SDS-PAGE. Affinity purified.
Expression system	Baculovirus infected Sf9 cells
Accession	P21802
Protein length	Protein fragment
Animal free	No
Nature	Recombinant
Species	Human
Molecular weight information	~72 kDa by SDS-PAGE
Amino acids	285 to 821
Modifications	mutated C491F
Tags	GST tag N-Terminus
Additional sequence information	BC039243

Specifications

Our **Abpromise guarantee** covers the use of **ab268563** in the following tested applications.

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

Applications	Functional Studies SDS-PAGE
Form	Liquid

Preparation and Storage

Stability and Storage	Shipped on Dry Ice. Upon delivery aliquot. Store at -80°C. Avoid freeze / thaw cycle.

pH: 7.50
Constituents: 0.79% Tris HCl, 0.87% Sodium chloride, 0.31% Glutathione, 0.003% EDTA, 0.004% DTT, 0.002% PMSF, 25% Glycerol (glycerin, glycerine)

This product is an active protein and may elicit a biological response in vivo, handle with caution.

General Info

Function

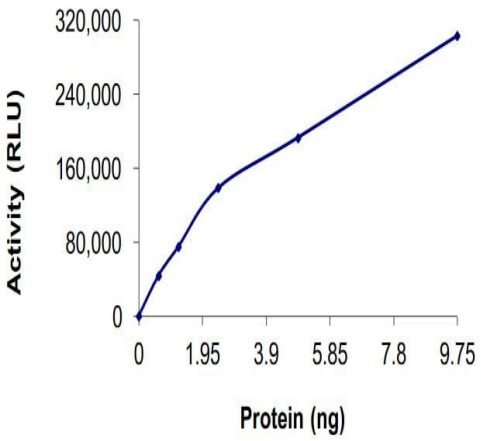
Receptor for acidic and basic fibroblast growth factors.

Involvement in disease

Defects in FGFR2 are the cause of Crouzon syndrome (CS) [MIM:123500]; also called craniofacial dysostosis type I (CFD1). CS is an autosomal dominant syndrome characterized by craniosynostosis (premature fusion of the skull sutures), hypertelorism, exophthalmos and external strabismus, parrot-beaked nose, short upper lip, hypoplastic maxilla, and a relative mandibular prognathism.
Defects in FGFR2 are a cause of Jackson-Weiss syndrome (JWS) [MIM:123150]. JWS is an autosomal dominant craniosynostosis syndrome characterized by craniofacial abnormalities and abnormality of the feet: broad great toes with medial deviation and tarsal-metatarsal coalescence.
Defects in FGFR2 are a cause of Apert syndrome (APRS) [MIM:101200]; also known as acrocephalosyndactyly type 1 (ACS1). APRS is a syndrome characterized by facio-cranio-synostosis, osseous and membranous syndactyly of the four extremities, and midface hypoplasia. The craniosynostosis is bicoronal and results in acrocephaly of brachysphenocephalic type. Syndactyly of the fingers and toes may be total (mitten hands and sock feet) or partial affecting the second, third, and fourth digits. Intellectual deficit is frequent and often severe, usually being associated with cerebral malformations.
Defects in FGFR2 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. Three subtypes of Pfeiffer syndrome have been described: mild autosomal dominant form (type 1); cloverleaf skull, elbow ankylosis, early death, sporadic (type 2); craniosynostosis, early demise, sporadic (type 3).
Defects in FGFR2 are the cause of Beare-Stevenson cutis gyrata syndrome (BSCGS) [MIM:123790]. BSCGS is an autosomal dominant condition is characterized by the furrowed skin disorder of cutis gyrata, acanthosis nigricans, craniosynostosis, craniofacial dysmorphism, digital anomalies, umbilical and anogenital abnormalities and early death.
Defects in FGFR2 are the cause of familial scaphocephaly syndrome (FSPC) [MIM:609579]; also known as scaphocephaly with maxillary retrusion and mental retardation. FSPC is an autosomal dominant craniosynostosis syndrome characterized by scaphocephaly, macrocephaly, hypertelorism, maxillary retrusion, and mild intellectual disability. Scaphocephaly is the most common of the craniosynostosis conditions and is characterized by a long, narrow head. It is due to premature fusion of the sagittal suture or from external deformation.
Defects in FGFR2 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 FGFR2 are the cause of Antley-Bixler syndrome (ABS) [MIM:207410]. ABS is a multiple congenital anomaly syndrome characterized by craniosynostosis, radiohumeral

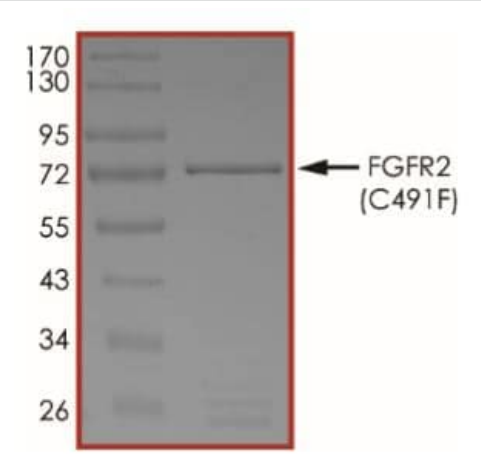
	synostosis, midface hypoplasia, malformed ears, arachnodactyly and multiple joint contractures. ABS is a heterogeneous disorder and occurs with and without abnormal genitalia in both sexes.
Sequence similarities	Belongs to the protein kinase superfamily. Tyr protein kinase family. Fibroblast growth factor receptor subfamily. Contains 3 Ig-like C2-type (immunoglobulin-like) domains. Contains 1 protein kinase domain.
Cellular localization	Secreted and Cell membrane.

Images



Functional Studies - Recombinant human FGFR2 (mutated C491F) protein (Active) (ab268563)

The specific activity of ab268563 was 253 nmol/min/mg in a kinase assay using Poly (4:1 Glu, Tyr) synthetic peptide as substrate.



SDS-PAGE - Recombinant human FGFR2 (mutated C491F) protein (Active) (ab268563)

SDS-PAGE analysis of ab268563.

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