abcam

Product datasheet

Recombinant human FGFR2 (mutated V564F) protein ab204104

4 图像

描述

产品名称	重组人FGFR2 (mutated V564F)蛋白
生物活性	The specific activity of ab204104 was determined to be 260 nmol/min/mg
纯度	> 85 % Densitometry. Affinity purified.
表达系统	Baculovirus infected Sf9 cells
Accession	P21802-20
蛋白长度	Protein fragment
无动物成分	No
性质	Recombinant
种属	Human
序列	RMKNTTKKPDFSSQPAVHKLTKRIPLRRQVSAESSSSMNSNT PLVRITTR LSSTADTPMLAGVSEYELPEDPKWEFPRDKLTLGKPLGEGCF GQVVMAEA VGIDKDKPKEAVTVAVKMLKDDATEKDLSDLVSEMEMMKMIG KHKNIINL LGACTQDGPLYVIFEYASKGNLREYLRARRPPGMEYSYDINR VPEEQMTF KDLVSCTYQLARGMEYLASQKCIHRDLAARNVLVTENNVMKI ADFGLARD INNIDYYKKTTNGRLPVKWMAPEALFDRVYTHQSDVWSFGVL MWEIFTLG GSPYPGIPVEELFKLLKEGHRMDKPANCTNELYMMMRDCWHA VPSQRPTF KQLVEDLDRILTLTTNEEYLDLSQPLEQYSPSYPDTRSSCSS GDDSVFSP DPMPYEPCLPQYPHINGSVKT
预测分子量	74 kDa including tags
氨基酸	285 to 704
修饰	mutated V564F
标签	GST tag N-Terminus
额外的序列信息	BC039243.

技术指标

Our **Abpromise guarantee** covers the use of **ab204104** in the following tested applications.

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

应用	SDS-PAGE Functional Studies Western blot
形式	Liquid

制备和贮存

稳定性和存储	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.

常规信息

功能	Receptor for acidic and basic fibroblast growth factors.
疾病相关	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.

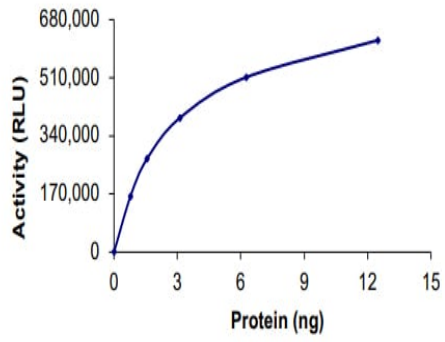
序列相似性

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.

细胞定位

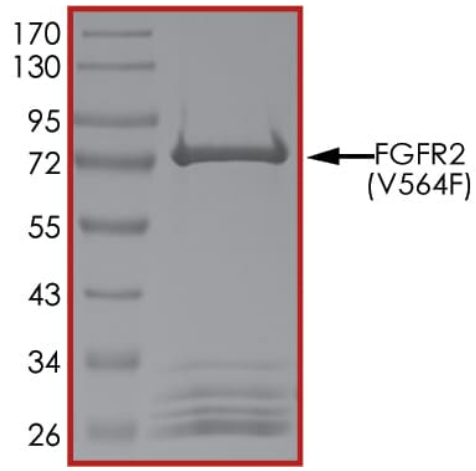
Secreted and Cell membrane.

图片



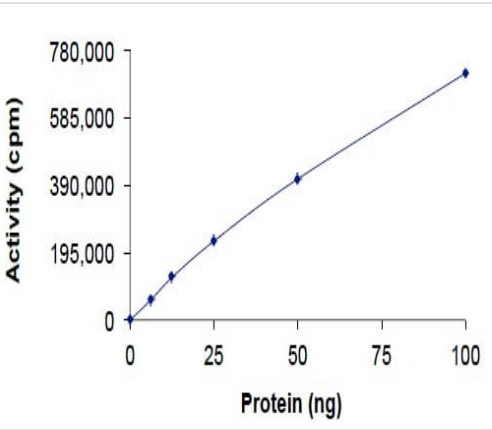
Functional Studies - Recombinant human FGFR2 (mutated V564F) protein (ab204104)

The specific activity of FGFR2 (ab204104) was determined to be 481 nmol/min/mg as per activity assay protocol and was equivalent to 280 nmol/min/mg as per radiometric assay



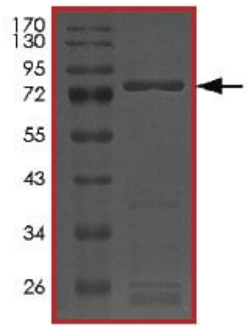
SDS-PAGE - Recombinant human FGFR2 (mutated V564F) protein (ab204104)

SDS PAGE analysis of ab204104



Functional Studies - Recombinant human FGFR2 (mutated V564F) protein (ab204104)

Kinase assay using ab204104 showing the specific activity to be 260 nmol/min/mg.



SDS-PAGE - Recombinant human FGFR2 (mutated V564F) protein (ab204104)

SDS-PAGE analysis of ab204104.

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