

Product datasheet

FGFR2 peptide ab76150

概述

产品名称 FGFR2肽

描述

性质 Synthetic

技术指标

Our [Abpromise guarantee](#) covers the use of **ab76150** in the following tested applications.

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

纯度 > 90 % SDS-PAGE.

形式 Lyophilised

制备和贮存

稳定性和存储 Shipped at 4°C. Upon delivery aliquot and store at -20°C. Avoid freeze / thaw cycles.

Lyophilized with 100% acetonitrile

复溶 Reconstitute with 0.1 ml deionized water for a final concentration of 1 mg/ml.

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 gyrate syndrome (BSCGS) [MIM:123790]. BSCGS is an autosomal dominant condition is characterized by the furrowed skin disorder of cutis gyrate, 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.

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