abcam

Product datasheet

Recombinant Human GDF 5 protein ab55329

1 图像

概述

产品名称	重组人GDF 5蛋白
蛋白长度	Full length protein

描述

性质	Recombinant
来源	Escherichia coli
氨基酸序列	
种属	Human
分子量	13 kDa

技术指标

Our Abpromise guarantee covers the use of **ab55329** in the following tested applications.

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

生物活性	Induction of alkaline phosphatase activity in ATDC5 cells: EC50 = 40 nM
应用	Functional Studies SDS-PAGE
纯度	> 95 % SDS-PAGE. Purified by proprietary chromatographic techniques
形式	Lyophilised
补充说明	For long term storage post reconstitiution, it is recommended to add a carrier protein (0.1% HSA or BSA).

制备和贮存

稳定性和存储	Shipped at 4°C. Store at +4°C short term (1-2 weeks). Upon delivery aliquot. Store at -20°C or -80°C. Avoid freeze / thaw cycle. Preservative: None This product is an active protein and may elicit a biological response in vivo, handle with caution.
复溶	Reconstitute the lyophilized GDF5 in sterile 20mM AcOH to not less than 100µg/ml, which can

then be further diluted to other aqueous solutions. No additives.

常规信息

功能

Could be involved in bone and cartilage formation. Chondrogenic signaling is mediated by the high-affinity receptor BMPR1B.

组织特异性

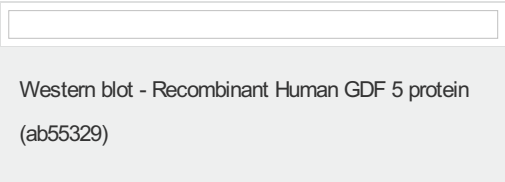
Predominantly expressed in long bones during embryonic development.

疾病相关

Defects in GDF5 are the cause of acromesomelic chondrodysplasia Grebe type (AMDG) [MIM:200700]. Acromesomelic chondrodysplasias are rare hereditary skeletal disorders characterized by short stature, very short limbs, and hand/foot malformations. The severity of limb abnormalities increases from proximal to distal with profoundly affected hands and feet showing brachydactyly and/or rudimentary fingers (knob-like fingers). AMDG is an autosomal recessive form characterized by normal axial skeletons and missing or fused skeletal elements within the hands and feet.
Defects in GDF5 are the cause of acromesomelic chondrodysplasia Hunter-Thompson type (AMDH) [MIM:201250]. AMDH is an autosomal recessive form of dwarfism. Patients have limb abnormalities, with the middle and distal segments being most affected and the lower limbs more affected than the upper. AMDH is characterized by normal axial skeletons and missing or fused skeletal elements within the hands and feet.
Defects in GDF5 are the cause of brachydactyly type C (BDC) [MIM:113100]. BDC is an autosomal dominant disorder characterized by an abnormal shortness of the fingers and toes.
Defects in GDF5 are the cause of Du Pan syndrome (DPS) [MIM:228900]; also known as fibular hypoplasia and complex brachydactyly. Du Pan syndrome is a rare autosomal recessive condition characterized by absence of the fibulae and severe acromesomelic limb shortening with small, non-functional toes. Although milder, the phenotype resembles the autosomal recessive Hunter-Thompson [MIM:201250] and Grebe types [MIM:200700] of acromesomelic chondrodysplasia.
Defects in GDF5 are a cause of symphalangism proximal syndrome (SYM1) [MIM:185800]. SYM1 is characterized by the hereditary absence of the proximal interphalangeal (PIP) joints (Cushing symphalangism). Severity of PIP joint involvement diminishes towards the radial side. Distal interphalangeal joints are less frequently involved and metacarpophalangeal joints are rarely affected whereas carpal bone malformation and fusion are common. In the lower extremities, tarsal bone coalition is common. Conducive hearing loss is seen and is due to fusion of the stapes to the petrous part of the temporal bone.
Defects in GDF5 are the cause of multiple synostoses syndrome type 2 (SYNS2) [MIM:610017]. Multiple synostoses syndrome is an autosomal dominant condition characterized by progressive joint fusions of the fingers, wrists, ankles and cervical spine, characteristic facies and progressive conductive deafness.
Defects in GDF5 are a cause of brachydactyly type A2 (BDA2) [MIM:112600]. Brachydactylies (BDs) are a group of inherited malformations characterized by shortening of the digits due to abnormal development of the phalanges and/or the metacarpals. They have been classified on an anatomic and genetic basis into five groups, A to E, including three subgroups (A1 to A3) that usually manifest as autosomal dominant traits.
Genetic variations in GDF5 are associated with susceptibility to osteoarthritis type 5 (OS5) [MIM:612400]. Osteoarthritis is a degenerative disease of the joints characterized by degradation of the hyaline articular cartilage and remodeling of the subchondral bone with sclerosis. Clinical symptoms include pain and joint stiffness often leading to significant disability and joint replacement.
Defects in GDF5 may be a cause of brachydactyly type A1 (BDA1) [MIM:112500]. Brachydactylies (BDs) are a group of inherited malformations characterized by shortening of the digits due to abnormal development of the phalanges and/or the metacarpals. They have been

	classified on an anatomic and genetic basis into five groups, A to E, including three subgroups (A1 to A3) that usually manifest as autosomal dominant traits.
序列相似性	Belongs to the TGF-beta family.
细胞定位	Secreted.

图片



Western blot - Recombinant Human GDF 5 protein (ab55329)

This antibody was tested against recombinant full length protein. Human GDF5 is a homodimer consisting of two 117 residue polypeptide chains.

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