abcam

Product datasheet

Recombinant human GDF 5 protein (Active) ab269223

1 Image

Description

Product name	Recombinant human GDF 5 protein (Active)
Biological activity	Alkaline phosphatase activity in ATDC5 cells ≤1.2 ug/mL; ≥ 8.3 x 10^2 units/mg (typical ED_{50} is < 25 ng/mL).
Purity	> 95 % SDS-PAGE. NULL
Endotoxin level	< 1.000 Eu/µg
Expression system	Escherichia coli
Accession	P43026
Protein length	Full length protein
Animal free	No
Nature	Recombinant
Species	Human
Sequence	MAPLATRQGK RPSKNLKARC SRKALHVNFK DMGWDDWIIA PLEYEAFHCE GLCEFPLRSH LEPTNHAVIQ TLMNSMDPES TPPTCCVPTR LSPISILFIDSA NNVVYKQY EDMVVESCGC R
Predicted molecular weight	55 kDa
Amino acids	382 to 501
Additional sequence information	Mature chain

Specifications

Our **Abpromise guarantee** covers the use of **ab269223** 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	Lyophilized

Preparation and Storage

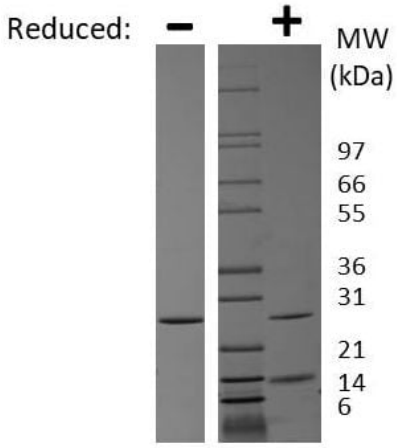
Stability and Storage	Shipped at Room Temperature. Upon delivery aliquot. Store at -20°C or -80°C. Working aliquots stored with a carrier protein are stable for at least 3 months at -20°C to -80°C.. Constituent: 0.1% Trifluoroacetic acid Lyophilized from This product is an active protein and may elicit a biological response in vivo, handle with caution.
Reconstitution	Sterile water at 0.1 mg/mL

General Info

Function	Could be involved in bone and cartilage formation. Chondrogenic signaling is mediated by the high-affinity receptor BMPR1B.
Tissue specificity	Predominantly expressed in long bones during embryonic development.
Involvement in disease	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.
Sequence similarities	Belongs to the TGF-beta family.
Cellular localization	Secreted.

Images



SDS-PAGE - Recombinant human GDF 5 protein (Active) (ab269223)

SDS-PAGE analysis of ab269223 at 1ug/lane under (-) non-reducing and (+) reducing conditions. 4-20% Tris glycine gel. Stained with coomassie blue.

Our Abpromise to you: Quality guaranteed and expert technical support

- Replacement or refund for products not performing as stated on the datasheet
- Valid for 12 months from date of delivery
- Response to your inquiry within 24 hours
- We provide support in Chinese, English, French, German, Japanese and Spanish
- Extensive multi-media technical resources to help you
- We investigate all quality concerns to ensure our products perform to the highest standards

If the product does not perform as described on this datasheet, we will offer a refund or replacement. For full details of the Abpromise, please visit **https://www.abcam.com/abpromise** or contact our technical team.

Terms and conditions

- Guarantee only valid for products bought direct from Abcam or one of our authorized distributors