abcam

Product datasheet

Recombinant Human GDF 5 protein ab52315

Description

Product name	Recombinant Human GDF 5 protein
Biological activity	The EC^{50} as determined by its ability to induce alkaline phosphatase production by ATDC-5 chondrogenic cells is 1-10 µg/ml.
Purity	>= 98 % SDS-PAGE.
Expression system	Escherichia coli
Accession	P43026
Protein length	Full length protein
Animal free	No
Nature	Recombinant
Species	Human
Sequence	APLATRQGKRPSKNLKARCSRKALHVNFKDMGWDDWIIA PLEYEAFHCEG LCEFPLRSHLEPTNHAVIQTLMNSMDPESTPPTCCVPTRL SPISILFIDS ANNVVYKQYEDMVVESCGCR

Specifications

Our **Abpromise guarantee** covers the use of **ab52315** in the following tested applications.

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

Applications	Phosphatase Activity SDS-PAGE
Form	Lyophilized
Additional notes	This product is manufactured by BioVision, an Abcam company and was previously called 4580 BMP-14 (GDF-5/CDMP-1), human recombinant. 4580-50 is the same size as the 50 µg size of ab52315.

Preparation and Storage

Stability and Storage	Shipped at 4°C. Store at +4°C short term (1-2 weeks). Upon delivery aliquot. Store at -20°C long term. Avoid freeze / thaw cycle. Constituent: 0.294% Sodium citrate

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

Reconstitution

Reconstitute to a concentration of 0.1-1.0 mg/ml in water containing BSA (50 µg BSA per 1 µg of protein). This solution can then be diluted into other aqueous buffers.

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.

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