abcam

## Product datasheet

# Recombinant Human GDF6 protein ab50230

1 Image

### Description

| | |
|---|---|
| **Product name** | Recombinant Human GDF6 protein |
| **Purity** | > 95 % SDS-PAGE. |
| **Endotoxin level** | < 0.100 Eu/µg |
| **Expression system** | Escherichia coli |
| **Protein length** | Full length protein |
| **Animal free** | No |
| **Nature** | Recombinant |
| **Species** | Human |
| **Sequence** | TAFASRHGKR HGKKSRLRCS KKPLHVNFKE LGWDDWIIAP LEYEAYHCEG VCDFPLRSHL EPTNHAIIQT LMNSMDPGST PPSCCVPTKL TPISILYIDA GNNVVYKQYE DMVVESCGCR |

### Specifications

Our **Abpromise guarantee** covers the use of **ab50230** in the following tested applications.

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

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|---|---|
| **Applications** | Western blot<br>Functional Studies<br>SDS-PAGE |
| **Form** | Lyophilized |

### Preparation and Storage

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|---|---|
| **Stability and Storage** | Shipped at 4°C. Upon delivery aliquot and store at -20°C. Avoid freeze / thaw cycles. |
| **Reconstitution** | Centrifuge the vial prior to opening. Reconstitute in water to a concentration of 0.1-1.0 mg/ml. This solution can then be diluted into other aqueous buffers and stored at 4oC for 1 week or -20oC for future use. |

## General Info

**Function**

Growth factor that controls proliferation and cellular differentiation in the retina and bone formation. Plays a key role in regulating apoptosis during retinal development. Establishes dorsal-ventral positional information in the retina and controls the formation of the retinotectal map (PubMed:23307924). Required for normal formation of bones and joints in the limbs, skull, digits and axial skeleton. Plays a key role in establishing boundaries between skeletal elements during development. Regulation of GDF6 expression seems to be a mechanism for evolving species-specific changes in skeletal strucutres. Seems to positively regulates differentiation of chondrogenic tissue through the growth factor receptors subunits BMPR1A, BMPR1B, BMPR2 and ACVR2A, leading to the activation of SMAD1-SMAD5-SMAD8 complex. The regulation of chondrogenic differentiation is inhibited by NOG (PubMed:26643732). Also involved in the induction of adipogenesis from mesenchymal stem cells. This mechanism acts through the growth factor receptors subunits BMPR1A, BMPR2 and ACVR2A and the activation of SMAD1-SMAD5-SMAD8 complex and MAPK14/p38.

**Involvement in disease**

Klippel-Feil syndrome 1, autosomal dominant
A chromosomal aberration involving GDF6 has been found in a patient with Klippel-Feil syndrome (KFS). Paracentric inv(8)(q22;2q23.3).
Microphthalmia, isolated, 4
Leber congenital amaurosis 17
Defects in POP1 may be the cause of multiple synostoses syndrome (SYNS). SYNS is a bone disease characterized by multiple progressive joint fusions that commonly involve proximal interphalangeal, tarsal-carpal joints. Additional features can include progressive conductive deafness.

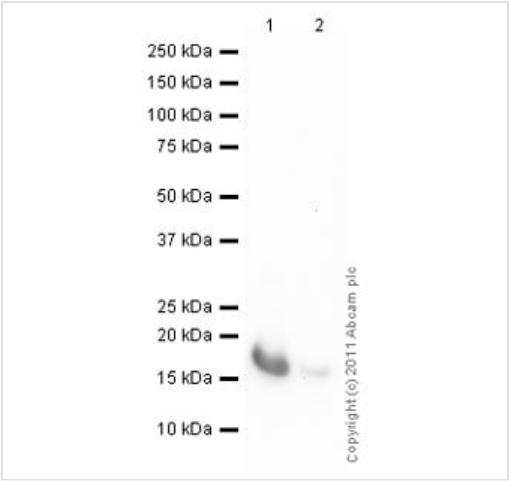
**Sequence similarities**

Belongs to the TGF-beta family.

**Cellular localization**

Secreted.

## Images




Western blot - Recombinant Human GDF6 protein (ab50230)

ab50230 is a homodimer consisting of two 120aa monomers. The homodimer format is expected to run at 27kDa, so the observed band is thought to be the monomer form.

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