10320 Camino Santa Fe, Suite G
San Diego, CA 92121
Tel: 858.875.1900 Fax: 858.875.1999

Fibrillin 1 Recombinant Rabbit mAb

Catalog # AP100073

Product Information

Application	WB, IHC-P, IHC-F, IF, E
Primary Accession	P35555
Other Accession	P35555
Reactivity	Human
Host	Rabbit
Clonality	Recombinant
Calculated MW	312298
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human Fibrillin 1
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SIMILARITY	Belongs to the fibrillin family.Contains 47 EGF-like domains.Contains 9 TB (TGF-beta binding) domains.
SUBUNIT	Interacts with VEGFC and VEGFD. Monomer in the absence of bound VEGFC or VEGFD. Homodimer in the presence of bound VEGFC or VEGFD. Can also form a heterodimer with KDR. Interacts with PTPN14; the interaction is enhanced by stimulation with VEGFC. Interacts with CRK, GRB2, PTK2/FAK1, SHC1, PIK3R1 and PTPN11/SHP-2. Identified in a complex with SRC and ITGB1.
Post-translational modifications	Forms intermolecular disulfide bonds either with other fibrillin-1 molecules or with other components of the microfibrils.
DISEASE	Defects in FBN1 are a cause of Marfan syndrome (MFS) [MIM:154700]. MFS is an autosomal dominant disorder that affects the skeletal, ocular, and cardiovascular systems. A wide variety of skeletal abnormalities occurs with MFS, including scoliosis, chest wall deformity, tall stature, abnormal joint mobility. Ectopia lentis occurs in up to about 80% of MFS patients and is almost always bilateral. The leading cause of premature death in MFS patients is progressive dilation of the aortic root and ascending aorta, causing aortic incompetence and dissection. Note=The majority of the more than 600 mutations in FBN1 currently known are point mutations, the rest are frameshifts and splice site mutations. Marfan syndrome has been suggested in at least 2 historical figures, Abraham Lincoln and Paganini. Defects in FBN1 are a cause of isolated ectopia lentis (EL) [MIM:129600]. The symptoms of this autosomal dominant fibrillinopathy overlap with those of Marfan syndrome, with the exclusion of the skeletal and cardiovascular manifestations. Defects in FBN1 are the cause of Weill-Marchesani syndrome autosomal dominant (ADWMS) [MIM:608328]. A rare connective tissue disorder characterized by short stature, brachydactyly, joint stiffness, and eye abnormalities including microspherophakia, ectopia lentis, severe myopia and glaucoma. Defects in FBN1 are a cause of Shprintzen-Goldberg craniosynostosis syndrome (SGS) [MIM:182212]. SGS is a very rare syndrome characterized by a marfanoid habitus, craniosynostosis, characteristic dysmorphic facial features, skeletal and cardiovascular abnormalities, mental retardation, developmental delay and learning disabilities. Defects in FBN1 are a cause of overlap connective tissue disease (OCTD) [MIM:604308]. A heritable disorder of connective tissue

	characterized by involvement of the mitral valve, aorta, skeleton, and skin. MASS syndrome is closely resembling both the Marfan syndrome and the Barlow syndrome. However, no dislocation of the lenses or aneurysmal changes occur in the aorta, and the mitral valve prolapse is by no means invariable. Defects in FBN1 are a cause of stiff skin syndrome (SSKS) [MIM:184900]. It is a syndrome characterized by hard, thick skin, usually over the entire body, which limits joint mobility and causes flexion contractures. Other occasional findings include lipodystrophy and muscle weakness.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	This gene encodes a member of the fibrillin family. The encoded protein is a large, extracellular matrix glycoprotein that serve as a structural component of 10-12 nm calcium-binding microfibrils. These microfibrils provide force bearing structural support in elastic and nonelastic connective tissue throughout the body. Mutations in this gene are associated with Marfan syndrome, isolated ectopia lentis, autosomal dominant Weill-Marchesani syndrome, MASS syndrome, and Shprintzen-Goldberg craniosynostosis syndrome. [provided by RefSeq, Jul 2008]

Additional Information

Gene ID	2200
Other Names	FBN1_HUMAN; FBN1; FBN; FBN1_MOUSE; Fbn-1; fibrillin 1; fibrillin 1 (Marfan syndrome); Marfan syndrome; asprosin
Dilution	WB=1:500-2000,IHC-P=1:200-800,IHC-F=1:200-800,IF=1:200-800,ELISA=1:5000-10000
Storage	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. When reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.

Protein Information

Name	FBN1 (HGNC:3603)
Synonyms	FBN
Function	[Fibrillin-1]: Structural component of the 10-12 nm diameter microfibrils of the extracellular matrix, which conveys both structural and regulatory properties to load-bearing connective tissues (PubMed:15062093, PubMed:1860873). Fibrillin-1-containing microfibrils provide long-term force bearing structural support (PubMed:27026396). In tissues such as the lung, blood vessels and skin, microfibrils form the periphery of the elastic fiber, acting as a scaffold for the deposition of elastin (PubMed:27026396). In addition, microfibrils can occur as elastin-independent networks in tissues such as the ciliary zonule, tendon, cornea and glomerulus where they provide tensile strength and have anchoring roles (PubMed:27026396). Fibrillin-1 also plays a key role in tissue homeostasis through specific interactions with growth factors, such as the bone morphogenetic proteins (BMPs), growth and differentiation factors (GDFs) and latent transforming growth factor-beta-binding proteins (LTBPs), cell-surface integrins and other extracellular matrix protein and proteoglycan components (PubMed:27026396). Regulates osteoblast maturation by controlling TGF- beta bioavailability and calibrating TGF-beta and BMP levels, respectively (By

similarity). Negatively regulates osteoclastogenesis by binding and sequestering an osteoclast differentiation and activation factor TNFSF11 (PubMed:24039232). This leads to disruption of TNFSF11-induced Ca(2+) signaling and impairment of TNFSF11-mediated nuclear translocation and activation of transcription factor NFATC1 which regulates genes important for osteoclast differentiation and function (PubMed:24039232). Mediates cell adhesion via its binding to cell surface receptors integrins ITGAV:ITGB3 and ITGA5:ITGB1 (PubMed:12807887, PubMed:17158881). Binds heparin and this interaction has an important role in the assembly of microfibrils (PubMed:11461921).

Cellular Location

Secreted. Note=Fibrillin-1 and Asprosin chains are still linked together during the secretion from cells, but are subsequently separated by furin (PubMed:24982166) [Asprosin]: Secreted. Note=Secreted by white adipose tissue and circulates in the plasma.

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