

Product name:	FBN1 Rabbit Polyclonal Antibody
Cat number:	ABN10856
Conjugate:	Unconjugated
Size:	100µL
Clone:	Polyclonal
Concentration:	1mg/ml
Host:	Rabbit
Isotype:	IgG
Immunogen:	The antiserum was produced against synthesized peptide derived from human Fibrillin-1. AA range:2811-2860
Reactivity:	Human,Mouse,Rat
Applications:	WB 1:500-1:2000,IHC 1:100-1:300,ICC/IF 1:50-1:200,ELISA 1:10000-1:20000
Purification:	Affinity purification
Form:	Liquid
Buffer:	Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% New type preservative N.
Storage:	Store at 4°C short term. Aliquot and store at -20°C for 12 months. Avoid freeze/thaw cycles.

Background:

This gene encodes a member of the fibrillin family of proteins. The encoded preproprotein is proteolytically processed to generate two proteins including the extracellular matrix component fibrillin-1 and the protein hormone asprosin. Fibrillin-1 is an extracellular matrix glycoprotein that serves as a structural component of calcium-binding microfibrils. These microfibrils provide force-bearing structural support in elastic and nonelastic connective tissue throughout the body. Asprosin, secreted by white adipose tissue, has been shown to regulate glucose homeostasis. Mutations in this gene are associated with Marfan syndrome and the related MASS phenotype, as well as ectopia lentis syndrome, Weill-Marchesani syndrome, Shprintzen-Goldberg syndrome and neonatal progeroid syndrome. [provided by RefSeq, Apr 2016],disease: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.,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. 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.,disease:Defects in FBN1 are a cause of MASS syndrome [MIM:604308]. MASS syndrome is 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.,disease: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.,disease:Defects in FBN1 are the cause of autosomal dominant Weill-Marchesani syndrome (WMS) [MIM:608328]. WMS is a rare connective tissue disorder characterized by short stature, brachydactyly, joint stiffness, and eye abnormalities including microspherophakia, ectopia lentis, severe myopia and glaucoma.,function:Fibrillins are structural components of 10-12 nm extracellular calcium-binding microfibrils, which occur either in association with elastin or in elastin-free bundles. Fibrillin-1-containing microfibrils provide long-term force bearing structural support.,online information:Fibrillin 1 mutation database,PTM:Forms intermolecular disulfide bonds either with other fibrillin-1 molecules or with other components of the microfibrils.,similarity:Belongs to the fibrillin family.,similarity:Contains 47 EGF-like domains.,similarity:Contains 9 TB (TGF-beta binding) domains.,subunit:Interacts with COL16A1.,