

Product name:	COL11A1 Rabbit Polyclonal Antibody
Cat number:	ABN09163
Conjugate:	Unconjugated
Size:	100µL
Clone:	Polyclonal
Concentration:	1mg/ml
Host:	Rabbit
Isotype:	IgG
Immunogen:	The antiserum was produced against synthesized peptide derived from human Collagen XI alpha1. AA range:581-630
Reactivity:	Human,Mouse
Applications:	WB 1:500-1:2000,IHC 1:100-1:300,ICC/IF 1:100-1:300,ELISA 1:10000-1:20000
Molecular Weight:	181kDa
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:

collagen type XI alpha 1 chain(COL11A1) Homo sapiens This gene encodes one of the two alpha chains of type XI collagen, a minor fibrillar collagen. Type XI collagen is a heterotrimer but the third alpha chain is a post-translationally modified alpha 1 type II chain. Mutations in this gene are associated with type II Stickler syndrome and with Marshall syndrome. A single-nucleotide polymorphism in this gene is also associated with susceptibility to lumbar disc herniation. Multiple transcript variants have been identified for this gene. [provided by RefSeq, Nov 2009],alternative products:Additional isoforms seem to exist. There is alternative usage of exon IIA or exon IIB. Transcripts containing exon IIA or IIB are present in cartilage, but exon IIB is preferentially utilized in transcripts from tendon,disease:Defects in COL11A1 are the cause of Marshall syndrome [MIM:154780]. It is an autosomal dominant disorder with ocular, orofacial, auditory and skeletal manifestations. It shares several features with Stickler syndrome, such as midfacial hypoplasia, high myopia, and sensorineural-hearing deficit.,disease:Defects in COL11A1 are the cause of Stickler syndrome type 2 (STL2) [MIM:604841]; also known as Stickler syndrome vitreous type 2. STL2 is an autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable.,function:May play an important role in fibrillogenesis by controlling lateral growth of collagen II fibrils.,PTM:Prolines at the third position of the tripeptide repeating unit (G-X-Y) are hydroxylated in some or all of the chains.,similarity:Belongs to the fibrillar collagen family.,similarity:Contains 1 TSP N-terminal (TSPN) domain.,subunit:Trimers composed of three different chains: alpha 1(XI), alpha 2(XI), and alpha 3(XI). Alpha 3(XI) is a post-translational modification of alpha 1(II). Alpha 1(V) can also be found instead of alpha 3(XI)=1(II).,tissue specificity:Cartilage, placenta and some tumor or virally transformed cell lines. Isoforms using exon IIA or IIB are found in the cartilage while isoforms using only exon IIB are found in the tendon.,