

Product name:	COL9A1 Rabbit Polyclonal Antibody
Cat number:	ABN09200
Conjugate:	Unconjugated
Size:	100µL
Clone:	Polyclonal
Concentration:	1mg/ml
Host:	Rabbit
Isotype:	IgG
Immunogen:	Synthesized peptide derived from COL9A1 . at AA range: 390-470
Reactivity:	Human,Mouse
Applications:	WB 1:500-1:2000,IHC 1:100-1:300,ICC/IF 1:50-1:200,ELISA 1:5000-1:10000
Molecular Weight:	120kDa
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 one of the three alpha chains of type IX collagen, which is a minor (5-20%) collagen component of hyaline cartilage. Type IX collagen is usually found in tissues containing type II collagen, a fibrillar collagen. Studies in knockout mice have shown that synthesis of the alpha 1 chain is essential for assembly of type IX collagen molecules, a heterotrimeric molecule, and that lack of type IX collagen is associated with early onset osteoarthritis. Mutations in this gene are associated with osteoarthritis in humans, with multiple epiphyseal dysplasia, 6, a form of chondrodysplasia, and with Stickler syndrome, a disease characterized by ophthalmic, orofacial, articular, and auditory defects. Two transcript variants that encode different isoforms have been identified for this gene. [provided by RefSeq, Jul 2008], alternative products: Additional isoforms seem to exist, disease: Defects in COL9A1 are a cause of COL9A1-related multiple epiphyseal dysplasia (COL9A1-MED) [MIM:120210], disease: Defects in COL9A1 are the cause of Stickler syndrome autosomal recessive COL9A1-related (COL9A1ARSTL) [MIM:120210]. COL9A1ARSTL is an autosomal recessive 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., domain: Each subunit is composed of three triple-helical domains interspersed with non-collagenous domains. The globular domain at the N-terminus of type IX collagen molecules represents the NC4 domain which may participate in electrostatic interactions with polyanionic glycosaminoglycans in cartilage., function: Structural component of hyaline cartilage and vitreous of the eye., PTM: Covalently linked to the telopeptides of type II collagen by lysine-derived cross-links., 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 fibril-associated collagens with interrupted helices (FACIT) family., similarity: Contains 1 TSP N-terminal (TSPN) domain., subunit: Heterotrimer of an alpha 1(IX), an alpha 2(IX) and an alpha 3(IX) chain.,