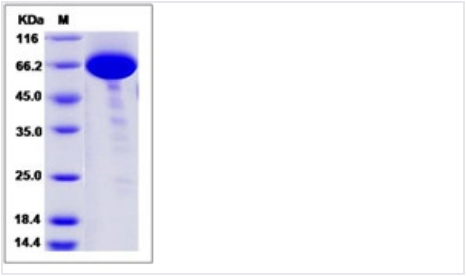
COL9A1 Recombinant Protein

PRODUCT OVERVIEW

Product Name	COL9A1, Recombinant Protein
Full Product Name	COL9A1 Protein, Human, Recombinant (mFc Tag)
Product Gene Name	COL9A1 recombinant protein
Product Synonym Names	Human COL9A1 Protein (Fc Tag); DJ149L1.1.2 Protein, Human; EDM6 Protein, Human; MED Protein, Human; STL4 Protein, Human; collagen, type IX, alpha 1
Sequence	Met1-Pro32 8
Host	HEK293 Cells
Form/Format	Lyophilized from sterile PBS, pH7.4. Normally 5%-8% trehalose, mannitol and 0.01% Tween80 are added as protectants before lyophilization. Please refer to the specific buffer information in the hard copy of CoA.
Purity/Purification	>95% as determined by SDS-PAGE
Preparation and Storage	Samples are stable for up to twelve months from date of receipt at -20 degree C to -80 degree C. Store it under sterile conditions at -20 degree C to -80 degree C. It is recommended that the protein be aliquoted for optimal storage. Avoid repeated freeze-thaw cycles. In general, recombinant proteins are provided as lyophilized powder which are shipped at ambient temperature. Bulk packages of recombinant proteins are provided as frozen liquid. They are shipped out with blue ice.
Molecular Weight	91,869 Da
Chromosome Location	Chromosome: 6; NC_000006.11 (70925743..71012786, complement). Location: 6q13
OMIM	120210
3D Structure	ModBase 3D Structure for P20849
Species	Human
Endotoxin	<1.0EU per ug of the protein as determined by the LAL method
Predicted N Terminal	Ala 24
Tag	C-mouse IgG1-Fc
Protein Construction	A DNA sequence encoding the human COL9A1 (P20849-3)(Met1-Pro328) was fused with Fc region of mouse IgG at the C-terminus.
Reconstitution	A hardcopy of COA with reconstitution instruction is sent along with the products. Please refer to it for detailed information.
Related Product Information for COL9A1 recombinant protein	MBS8121124 is a recombinant protein targeting COL9A1. Also known as COL9A1 Protein, Human, Recombinant (mFc Tag). Host: HEK293 Cells.
Other Notes	Small volumes of COL9A1 recombinant protein vial(s) may occasionally become entrapped in the seal of the product vial during shipment and storage. If necessary, briefly centrifuge the vial on a tabletop centrifuge to dislodge any liquid in the container`s cap. Certain products may require to ship with dry ice and additional dry ice fee may apply.



SDS-Page of COL9A1 recombinant protein

ADDITIONAL DETAILS

NCBI/Uniprot data below describe general gene information for COL9A1. It may not necessarily be applicable to this product.

NCBI GI #	73486666
NCBI GeneID	1297
NCBI Accession #	NP_001842.3
UniProt Primary Accession #	P20849
UniProt Secondary Accession #	Q13699; Q13700; Q5TF52; Q6P467; Q96BM8; Q99225; Q9H151; Q9H152; Q9Y6P2; Q9Y6P3
UniProt Related Accession #	P20849
GenBank Nucleotide #	NM_001851.4
NCBI Official Full Name	collagen alpha-1(IX) chain isoform 1
NCBI Official Synonym Full Names	collagen, type IX, alpha 1
NCBI Official Symbol	COL9A1
NCBI Official Synonym Symbols	MED; EDM6; STL4; DJ149L1.1.2
UniProt Protein Name	Collagen alpha-1(IX) chain
Protein Family	Collagen
UniProt Gene Name	COL9A1
UniProt Entry Name	CO9A1_HUMAN
NCBI Protein Information	collagen alpha-1(IX) chain; collagen alpha-1(IX) chain; alpha-1(IX) collagen chain; collagen IX, alpha-1 polypeptide; cartilage-specific short collagen
NCBI Summary for COL9A1	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]

UniProt Comments for COL9A1	COL9A1: Structural component of hyaline cartilage and vitreous of the eye. Defects in COL9A1 are the cause of multiple epiphyseal dysplasia type 6 (EDM6). A generalized skeletal dysplasia associated with significant morbidity. Joint pain, joint deformity, waddling gait, and short stature are the main clinical signs and symptoms. Radiological examination of the skeleton shows delayed, irregular mineralization of the epiphyseal ossification centers and of the centers of the carpal and tarsal bones. Multiple epiphyseal dysplasia is broadly categorized into the more severe Fairbank and the milder Ribbing types. The Fairbank type is characterized by shortness of stature, short and stubby fingers, small epiphyses in several joints, including the knee, ankle, hand, and hip. The Ribbing type is confined predominantly to the hip joints and is characterized by hands that are normal and stature that is normal or near-normal. Defects in COL9A1 are the cause of Stickler syndrome type 4 (STL4). 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. Belongs to the fibril-associated collagens with interrupted helices (FACIT) family. 3 isoforms of the human protein are produced by alternative splicing. Protein type: Secreted, signal peptide; Secreted Chromosomal Location of Human Ortholog: 6q13 Cellular Component: proteinaceous extracellular matrix; endoplasmic reticulum lumen; extracellular region Molecular Function: metal ion binding; extracellular matrix structural constituent conferring tensile strength Biological Process: extracellular matrix disassembly; axon guidance; collagen catabolic process; organ morphogenesis; extracellular matrix organization and biogenesis; tissue homeostasis Disease: Stickler Syndrome, Type Iv; Epiphyseal Dysplasia, Multiple, 6
Research Articles on COL9A1	**1.** The NC2 domain of type IX collagen determines the chain composition but also the chain register of the triple helix.
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