COL9A1 cDNA Clone – Collagen, Type IX, Alpha 1 (Human)

PRODUCT OVERVIEW

Product Name	COL9A1, cDNA Clone
Full Product Name	COL9A1 cDNA Clone
Product Gene Name	COL9A1 cdna clone
Sequence	atgaagacct gctggaaaat tccagttttc ttctttgtgt gcagtttcct ggaaccctgg gcatctgcag ctgtcaagcg tcgccccaga ttccctgtca attccaattc taatggtgga aatgaactct gtccaaagat caggattggc caagatgact taccagggtt tgatctgatc tctcagttcc aggtagataa agcagcatct agaagagcta tccagagagt agtgggatca gctacattgc aggtggctta caagttggga aataatgtag acttcaggat tccaactagg aatttatatc ccagtggact gcctgaagaa tactccttct tgacgacgtt tcgaatgact ggaagcactc tcaaaaagaa ctggaacatt tggcagattc aggattcctc tgggaaggag caagttggca taaagattaa tggccaaaca caatctgttg tattttcata caagggactg gatggaagtc tccaaacagc agccttttcg aatttgtcct ccttgtttga ttcccagtgg cataagatca tgattggcgt ggagaggagt agtgctactc tttttgttga ctgcaacagg attgaatctt tacctataaa gccaagaggc ccaattgaca ttgatggctt tgctgtgctg ggaaaacttg cagataatcc tcaagtttct gttccatttg aacttcaatg gatgctgatc cattgtgacc ccctgcggcc caggagagaa acttgccatg agctgccagc cagaataacg cccagccaga ccaccgacga gagaggtccc ccgggtgagc agggtcctcc cgggcctccg ggccccccctg gagttccagg catcgatggc atcgacggtg accgaggtcc taagggcccc ccgggccccc cgggtcctgc aggtgaaccg ggaaagccag gagctccagg caagcctggc acacctggcg ctgataccag tccttga
Molecular Weight	35,509 Da
OMIM	120210
Vector	pENTR223.1 or pUC
Clone Sequence Report	Provided with product shipment
ISO Certification	Manufactured in an ISO 9001:2015 Certified Laboratory.
Related Product Information for COL9A1 cdna clone	MBS1265882 is a cDNA clone targeting COL9A1.
Other Notes	Small volumes of COL9A1 cdna clone 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.

ADDITIONAL DETAILS

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

NCBI GI #	15929965
NCBI GeneID	1297
NCBI Accession #	BC015409
UniProt Secondary Accession #	Q13699; Q13700; Q5TF52; Q6P467; Q96BM8; Q99225; Q9H151; Q9H152; Q9Y6P2; Q9Y6P3
UniProt Related Accession #	P20849
NCBI Official Full Name	Homo sapiens collagen, type IX, alpha 1, mRNA
NCBI Official Synonym Full Names	collagen type IX alpha 1 chain
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
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: endoplasmic reticulum lumen; extracellular region Molecular Function: extracellular matrix structural constituent conferring tensile strength Biological Process: extracellular matrix organization and biogenesis; organ morphogenesis Disease: Epiphyseal Dysplasia, Multiple, 6; Stickler Syndrome, Type Iv
Research Articles on COL9A1	**1.** We observed a significant association between rs6910140 of COL9A1 and KBD, suggesting a role of COL9A1 in the development of KBD.
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