

## Human COL11A Recombinant Protein

PX-P1081-10

### DESCRIPTION

COL11A1 plays a significant role in fibrillogenesis by controlling lateral growth of collagen II fibrils. Defects in COL11A1 are the source of Stickler syndrome type 2 (STL2); also known as Stickler syndrome vitreous type 2. STL2 is an autosomal dominant form of Stickler syndrome, an inherited disease that combines ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. The degree of hearing loss varies among concerned persons and may become more severe over time. Defects in COL11A1 are also the cause of Marshall Syndrome (MRSHS), an autosomal dominant disorder identified by ocular abnormalities, deafness, craniofacial anomalies, and anhidrotic ectodermal dysplasia etc. Besides, defects in COL11A1 can be the cause of fibrochondrogenesis type 1 (FBCG1). A severe short-limbed skeletal dysplasia characterized by pear-shaped vertebral bodies, and characteristic morphology of the growth plate, in which the chondrocytes have a fibroblastic appearance and there are regions of fibrous cartilage extracellular matrix. The protein is also expressed in cancer-activated stroma and could be an anti-cancer target.

### OVERVIEW

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<b>SIZE</b>	10 ug
<b>ORIGIN SPECIES</b>	Human
<b>FRAGMENT</b>	Partial (Small part)
<b>PROTEIN DELIVERED WITH TAG</b>	Yes
<b>MOLECULAR WEIGHT WITH TAG IF ANY</b>	48,19 kDa
<b>DELIVERY CONDITION</b>	Dry Ice

### PRODUCT INFORMATION

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<b>EXPRESSION SYSTEM</b>	Prokaryotic expression
<b>HOST</b>	E.coli
<b>PURITY</b>	>95%
<b>PROTEIN ACCESSION</b>	AAF04724.1
<b>FORM</b>	liquid
<b>BUFFER</b>	TrisHCl 50mM. PBS if dialysis
<b>STABILITY &amp; STORAGE</b>	4°C for short term (1 week), -20°C or -80°C for long term (avoid freezing/thawing cycles; addition of 20-40% glycerol improves cryoprotection)

## MORE INFO

<b>GENE ID</b>	1301
<b>SWISSPROTID</b>	P12107
<b>UNIPROT ID</b>	P12107
<b>UNIPROT LINK</b>	<a href="http://www.uniprot.org/uniprot/P12107">http://www.uniprot.org/uniprot/P12107</a>
<b>NCBI GENE ALIASES</b>	COLL6, CO11A1, STL2
<b>SYNONYMS</b>	CO11A1; COBA1; COL11A1; COLL6; Collagen alpha-1(XI) chain; collagen XI, alpha-1 polypeptide; collagen, type XI, alpha 1; STL2

## PROTEIN SEQUENCE

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MSPILGYWKIKGLVQPTRLLEYLEEKYEEHLYERDEGDKWRNKKFELGLEFPNLPYYIDGDVKLTQSMAIRYIADKHN  
MLGGCPKERAEISMLEGAVLDIRYGVSRIAYSKDFETLKVDFLSKLPPEMLKMFEDRLCHKTYLNGDHVTHPDFMLYDALD  
VVLYMDPMCLDAFPKLVCFKKRIEAIQIDKYLKSSKYIAWPLQGQWQATFGGGDHPPKSDGSTSGSGHHHHHSAGLVPR  
GSTAIGMKETAA

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