



# Collagen II $\alpha$ 1 (Cleaved-Ala1241) mouse mAb

<b>Catalog No</b>	YP-mAb-15063
<b>Isotype</b>	IgG
<b>Reactivity</b>	Human;Mouse;Rat
<b>Applications</b>	WB
<b>Gene Name</b>	COL2A1
<b>Protein Name</b>	Collagen II $\alpha$ 1 (Cleaved-Ala1241)
<b>Immunogen</b>	Synthesized peptide derived from human Collagen II $\alpha$ 1 (Cleaved-Ala1241)
<b>Specificity</b>	This antibody detects endogenous levels of Human,Mouse,Rat Collagen II $\alpha$ 1 (Cleaved-Ala1241, protein was cleaved amino acid sequence between 1241-1242 )
<b>Formulation</b>	Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% sodium azide.
<b>Source</b>	Monoclonal, Mouse,IgG
<b>Purification</b>	The antibody was affinity-purified from mouse antiserum by affinity-chromatography using epitope-specific immunogen.
<b>Dilution</b>	WB 1:500-1:2000
<b>Concentration</b>	1 mg/ml
<b>Purity</b>	$\geq 90\%$
<b>Storage Stability</b>	-20°C/1 year
<b>Synonyms</b>	Collagen alpha-1(II) chain (Alpha-1 type II collagen) [Cleaved into: Collagen alpha-1(II) chain; Chondrocalcin]
<b>Observed Band</b>	135 160kD
<b>Cell Pathway</b>	Secreted, extracellular space, extracellular matrix .
<b>Tissue Specificity</b>	Isoform 2 is highly expressed in juvenile chondrocyte and low in fetal chondrocyte.
<b>Function</b>	skeletal system development, cartilage condensation, ossification, tissue homeostasis, endochondral ossification, chondrocyte differentiation, heart morphogenesis, proteoglycan metabolic process, cell adhesion, sensory organ development, heart development, sensory perception, visual perception, sensory perception of sound, glycoprotein metabolic process, embryonic development ending in birth or egg hatching, regulation of cell death, cell-cell adhesion, biological adhesion, extracellular matrix organization, collagen fibril organization, appendage morphogenesis, limb morphogenesis, ear morphogenesis, inner ear morphogenesis, homeostatic process, regulation of apoptosis, chordate embryonic development, extracellular structure organization, negative regulation of apoptosis, regulation of programmed cell death, negative regulation of programmed cell death, ear development, embryonic organ morp



## Background

disease: Defects in COL2A1 are a cause of primary avascular necrosis of femoral head (ANFH) [MIM:608805]; also called ischemic necrosis of the femoral head or osteonecrosis of the femoral head. ANFH causes disability that often requires surgical intervention. Most cases are sporadic, but families in which there is an autosomal dominant inheritance of the disease have been identified. It has been estimated that 300,000 to 600,000 people in the United States have ANFH. Approximately 15,000 new cases of this common and disabling disorder are reported annually. The age at the onset is earlier than that for osteoarthritis. The diagnosis is typically made when patients are between the ages of 30 and 60 years. The clinical manifestations, such as pain on exertion, a limping gait, and a discrepancy in leg length, cause considerable disability. Moreover, nearly 10 percent of the 500,000 total-hip arthroplasties performed each year in the United States involve patients with ANFH. As a result, this disease creates a substantial socioeconomic cost as well as a burden for patients and their families.

disease: Defects in COL2A1 are a cause of rhegmatogenous retinal detachment autosomal dominant (DRRD) [MIM:609508]. Rhegmatogenous retinal detachment most frequently results from a break or tear in the retina that allows fluid from the vitreous humor to enter the potential space beneath the retina. It is often associated with pathologic myopia and in most cases leads to visual impairment or blindness if untreated.

disease: Defects in COL2A1 are the cause of a variety of chondrodysplasia including hypochondrogenesis and osteoarthritis.

disease: Defects in COL2A1 are the cause of achondrogenesis hypochondrogenesis type 2 (ACG2) [MIM:200610]. ACG2 is a disease characterized by the absence of ossification in the vertebral column, sacrum and pubic bones.

disease: Defects in COL2A1 are the cause of Kniest syndrome (KS) [MIM:156550]; also known as Kniest dysplasia or metatropic dwarfism type II. KS is a moderately severe chondrodysplasia phenotype that results from mutations in the COL2A1 gene. Characteristics of the disorder include a short trunk and extremities, mid-face hypoplasia, cleft palate, myopia, retinal detachment, and hearing loss.

disease: Defects in COL2A1 are the cause of Legg-Calve-Perthes disease (LCPD) [MIM:150600]; also known as Legg-Perthes disease or Perthes disease. LCPD is characterized by loss of circulation to the femoral head, resulting in avascular necrosis in a growing child. Clinical pictures of the disease vary, depending on the phase of disease progression through ischemia, revascularization, fracture and collapse, and repair and remodeling of the bone.

disease: Defects in COL2A1 are the cause of multiple epiphyseal dysplasia with myopia and conductive deafness (EDMMD) [MIM:132450]. Multiple epiphyseal dysplasia is a generalized skeletal dysplasia associated with significant morbidity. Joint pain, joint deformity, waddling gait, and short stature are the main clinical signs and symptoms. EDMMD is an autosomal dominant disorder characterized by epiphyseal dysplasia associated with progressive myopia, retinal thinning, crenated cataracts, conductive deafness.

disease: Defects in COL2A1 are the cause of osteoarthritis with mild chondrodysplasia [MIM:604864]. Osteoarthritis is a common disease that produces joint pain and stiffness together with radiologic evidence of progressive degeneration of joint cartilage. Some forms of osteoarthritis are secondary to events such as trauma, infections, metabolic disorders, or congenital or heritable conditions that deform the epiphyses or related structures. In most patients, however, there is no readily identifiable cause of osteoarthritis. Inheritance in a Mendelian dominant manner has been demonstrated in some families with primary generalized osteoarthritis. Reports demonstrate coinheritance of primary generalized osteoarthritis with specific alleles of the gene COL2A1, the precursor of the major protein of cartilage.

disease: Defects in COL2A1 are the cause of platyspondylic lethal skeletal dysplasia Torrance type (PLSD-T) [MIM:151210]. Platyspondylic lethal skeletal dysplasias (PLSDs) are a heterogeneous group of chondrodysplasias characterized by severe platyspondyly and limb shortening. PLSD-T is characterized by varying platyspondyly, short ribs with anterior cupping, hypoplasia of the lower ilia with broad ischial and pubic bones, and shortening of the tubular bones with splayed and cupped metaphyses. Histology of the growth plate typically shows focal hypercellularity with slightly enlarged chondrocytes in the resting cartilage and relatively well-preserved columnar formation and ossification at the chondro-osseous junction. PLSD-T is generally a perinatally lethal disease, but a few long-term survivors have been reported.

disease: Defects in COL2A1 are the cause of spondyloepiphyseal dysplasia congenital type (SEDC) [MIM:183900]. This disorder is characterized by disproportionate short stature and pleiotropic involvement of the skeletal and ocular systems.

disease: Defects in COL2A1 are the cause of spondyloperipheral dysplasia (SPD) [MIM:271700]. SPD patients manifest short stature, midface hypoplasia, sensorineural hearing loss, spondyloepiphyseal dysplasia, platyspondyly and brachydactyly.

disease: Defects in COL2A1 are the cause of Stickler syndrome type 1 (STL1) [MIM:108300]; also known as vitreous type 1, or



membranous vitreous type. STL1 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 platyspondylitis 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. disease: Defects in COL2A1 are the cause of Stickler syndrome type 1 non-syndromic ocular (STL1O) [MIM:609508]. STL1O is an autosomal dominant form of Stickler syndrome characterized by the ocular signs typically seen in STL1 such as cataract, myopia, retinal detachment. STL1 systemic features of premature osteoarthritis, cleft palate, hearing impairment, and craniofacial abnormalities are either absent or very mild in STL1O patients. disease: Defects in COL2A1 are the cause of Strudwick type spondyloepimetaphyseal dysplasia (SEMD) [MIM:184250]. SEMD is characterized by disproportionate short stature, pectus carinatum, and scoliosis, as well as dappled metaphyses (which is not seen in SEDC). disease: Defects in COL2A1 are the cause of Wagner syndrome type II (WS-II); a disease characterized by early-onset cataracts, lattice degeneration of the retina, and retinal detachment without involvement of monocular tissues. disease: Of special interest are three different variants that replace arginine codons at positions 275, 719 and 989 in the triple-helical domain with codons for cysteine, an amino acid not normally found in the triple-helical domain of type II collagen from any species. They are of special interest, because they are the only amino acid substitutions in the triple-helical domain that replaces a Y-position amino acid and cause a disease phenotype. Also, they are recurrent in that they have been found in more than one unrelated individual. function: Type II collagen is specific for cartilaginous tissues. It is essential for the normal embryonic development of the skeleton, for linear growth and for the ability of cartilage to resist compressive forces. PTM: Prolines at the third position of the tripeptide repeating unit (G-X-Y) are hydroxylated in some or all of the chains. PTM: The N-telopeptide is covalently linked to the helical COL2 region of alpha 1(IX), alpha 2(IX) and alpha 3(IX) chain. The C-telopeptide is covalently linked to an another site in the helical region of alpha 3(IX) COL2. similarity: Belongs to the fibrillar collagen family. similarity: Contains 1 VWFC domain. subunit: Homotrimers of alpha 1(II) chains. tissue specificity: High expression of isoform 2 in juvenile chondrocyte and low in fetal chondrocyte.

**matters needing attention**

Avoid repeated freezing and thawing!

**Usage suggestions**

This product can be used in immunological reaction related experiments. For more information, please consult technical personnel.

**Products Images**