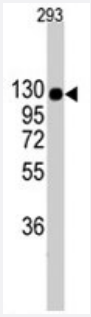
COL2A1 polyclonal antibody

Catalog # PAB3065 Size 400 uL

Applications



Western Blot (Cell lysate)

Western blot analysis of COL2A1 polyclonal antibody (Cat # PAB3065) in 293 cell line lysates (35 ug/lane). COL2A1 (arrow) was detected using the purified polyclonal antibody.

Specification

Product Description	Rabbit polyclonal antibody raised against synthetic peptide of COL2A1.
Immunogen	A synthetic peptide (conjugated with KLH) corresponding to C-terminus of human COL2A1.
Host	Rabbit
Reactivity	Human
Form	Liquid
Purification	Protein A purification
Recommend Usage	Western Blot (1:1000) The optimal working dilution should be determined by the end user.
Storage Buffer	In PBS (0.09% sodium azide)
Storage Instruction	Store at 4°C. For long term storage store at -20°C. Aliquot to avoid repeated freezing and thawing.
Note	This product contains sodium azide: a POISONOUS AND HAZARDOUS SUBSTANCE which shoul d be handled by trained staff only.

Applications

- Western Blot (Cell lysate)

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Gene Info — COL2A1

Entrez GeneID	1280
Protein Accession#	NP_001835;P02458
Gene Name	COL2A1
Gene Alias	ANFH, AOM, COL11A3, MGC131516, SEDC
Gene Description	collagen, type II, alpha 1
Omim ID	108300 120140 132450 151210 156550 165720 183900 184250 184252 200610 215150 271700 608805
Gene Ontology	Hyperlink
Gene Summary	This gene encodes the alpha-1 chain of type II collagen, a fibrillar collagen found in cartilage and the vitreous humor of the eye. Mutations in this gene are associated with achondrogenesis, chondrodysplasia, early onset familial osteoarthritis, SED congenita, Langer-Saldino achondrogenesis, Kniest dysplasia, Stickler syndrome type I, and spondyloepimetaphyseal dysplasia Strudwick type. In addition, defects in processing chondrocalcin, a calcium binding protein that is the C-propeptide of this collagen molecule, are also associated with chondrodysplasia. There are two transcripts identified for this gene. [provided by RefSeq
Other Designations	OTTHUMP00000195063|cartilage collagen|chondrocalcin|collagen II, alpha-1 polypeptide

Publication Reference

- Missense and nonsense mutations in the alternatively-spliced exon 2 of COL2A1 cause the ocular variant of Stickler syndrome.

  McAlinden A, Majava M, Bishop PN, Perveen R, Black GC, Pierpont ME, Ala-Kokko L, Mannikko M.

  Human Mutation 2008 Jan; 29(1):83.

- Stickler and branchio-oto-renal syndromes in a patient with mutations in EYA1 and COL2A1 genes.

  Olavarrieta L, Morales-Angulo C, del Castillo I, Moreno F, Moreno-Pelayo MA.

  Clinical Genetics 2008 Mar; 73(3):262.

- A familial case of achondrogenesis type II caused by a dominant COL2A1 mutation and "patchy" expression in the mosaic father.

  Forzano F, Lituania M, Viassolo A, Superti-Furga V, Wildhardt G, Zabel B, Faravelli F.

  American Journal of Medical Genetics. Part A 2007 Dec; 143A(23):2815.

Pathway

- ECM-receptor interaction
- Focal adhesion

Disease

- Asthma
- Cardiovascular Diseases
- Cleft Lip
- Cleft Palate
- Cumulative Trauma Disorders
- Diabetes Mellitus
- Edema
- Gaucher disease
- Genetic Predisposition to Disease
- Hip Dislocation
- Intervertebral Disk Displacement
- Legg-Perthes Disease
- Myopia

- Occupational Diseases
- Osteoarthritis
- Thrombophilia
- Toxoplasmosis