Native Human Collagen I protein

**Product name**: Native Human Collagen I protein

**Expression system**: Native

**Accession**: P02452

**Protein length**: Full length protein

**Animal free**: No

**Nature**: Native

**Species**: Human

**Applications**: ELISA, SDS-PAGE

**Form**: Lyophilized

**Reconstitution**: Solubilize the white, hygroscopic powder slowly in 10 mM acetic acid (0.2% w/v) and adjust to neutral pH by adding 2/5 vol. 2.5X Carbonate buffer (0.25 M NaHCO₃, 1.25 M NaCl, 0.1% NaN₃).

**Stability and Storage**: Shipped on dry ice. Upon delivery aliquot and store at -80°C. Avoid freeze / thaw cycles.

**Function**: Type I collagen is a member of group I collagen (fibrillar forming collagen).

**Tissue specificity**: Forms the fibrils of tendon, ligaments and bones. In bones the fibrils are mineralized with calcium hydroxyapatite.

**Involvement in disease**: Defects in COL1A1 are the cause of Caffey disease (CAFFD) [MIM:114000]; also known as infantile cortical hyperostosis. Caffey disease is characterized by an infantile episode of massive subperiosteal new bone formation that typically involves the diaphyses of the long bones.
mandible, and clavicles. The involved bones may also appear inflamed, with painful swelling and systemic fever often accompanying the illness. The bone changes usually begin before 5 months of age and resolve before 2 years of age.

Defects in COL1A1 are a cause of Ehlers-Danlos syndrome type 1 (EDS1) [MIM:130000]; also known as Ehlers-Danlos syndrome gravis. EDS is a connective tissue disorder characterized by hyperextensible skin, atrophic cutaneous scars due to tissue fragility and joint hyperlaxity. EDS1 is the severe form of classic Ehlers-Danlos syndrome.

Defects in COL1A1 are the cause of Ehlers-Danlos syndrome type 7A (EDS7A) [MIM:130060]; also known as autosomal dominant Ehlers-Danlos syndrome type VII. EDS is a connective tissue disorder characterized by hyperextensible skin, atrophic cutaneous scars due to tissue fragility and joint hyperlaxity. EDS7A is marked by bilateral congenital hip dislocation, hyperlaxity of the joints, and recurrent partial dislocations.

Defects in COL1A1 are a cause of osteogenesis imperfecta type 1 (OI1) [MIM:166200]. A dominantly inherited connective tissue disorder characterized by bone fragility and blue sclerae. Osteogenesis imperfecta type 1 is non-deforming with normal height or mild short stature, and no dentinogenesis imperfecta.

Defects in COL1A1 are a cause of osteogenesis imperfecta type 2A (OI2A) [MIM:166210]; also known as osteogenesis imperfecta congenita. A connective tissue disorder characterized by bone fragility, with many perinatal fractures, severe bowing of long bones, undermineralization, and death in the perinatal period due to respiratory insufficiency.

Defects in COL1A1 are a cause of osteogenesis imperfecta type 3 (OII) [MIM:259420]. A connective tissue disorder characterized by progressively deforming bones, very short stature, a triangular face, severe scoliosis, grayish sclera, and dentinogenesis imperfecta.

Defects in COL1A1 are a cause of osteogenesis imperfecta type 4 (OII) [MIM:166220]; also known as osteogenesis imperfecta with normal sclerae. A connective tissue disorder characterized by moderately short stature, mild to moderate scoliosis, grayish or white sclera and dentinogenesis imperfecta.

Genetic variations in COL1A1 are a cause of susceptibility to osteoporosis (OSTEOP) [MIM:166710]; also known as involutional or senile osteoporosis or postmenopausal osteoporosis. Osteoporosis is characterized by reduced bone mass, disruption of bone microarchitecture without alteration in the composition of bone. Osteoporotic bones are more at risk of fracture.

Note=A chromosomal aberration involving COL1A1 is found in dermatofibrosarcoma protuberans. Translocation t(17;22)(q22;q13) with PDGF.

**Sequence similarities**

Belongs to the fibrillar collagen family.

Contains 1 fibrillar collagen NC1 domain.

Contains 1 VWFC domain.

**Post-translational modifications**

Proline residues at the third position of the tripeptide repeating unit (G-X-Y) are hydroxylated in some or all of the chains. Proline residues at the second position of the tripeptide repeating unit (G-X-Y) are hydroxylated in some of the chains.

O-linked glycan consists of a Glc-Gal disaccharide bound to the oxygen atom of a post-translationally added hydroxyl group.

**Cellular localization**

Secreted > extracellular space > extracellular matrix.

**Images**
SDS PAGE analysis of ab134516.

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