A recent cDNA analysis performed at UMASS’ CPCT identified differential expression of the COL1A1 gene. COL1A1 and collagen type I alpha 1 are the official symbol and official gene name corresponding to RefSeq accession # NG\_007400.1. RefSeq also includes EDSC, OI1, OI2, OI3, and OI4 as aliases of the COL1A1 gene.

The COL1A1 gene codes for collagen alpha-1(I) chain protein, also known as alpha-1 type I collagen, alpha1(I) procollagen, collagen alpha 1 chain type I, collagen alpha-1(I) chain preproprotein, collagen of skin, tendon and bone, alpha-1 chain pro-alpha-1 collagen type 1, type I proalpha 1, and type I procollagen alpha 1 chain. The protein coded by COL1A2, alpha 2 type I procollagen chain combines in a 1:2 ratio with the protein coded by the COL1A1 gene to form type I collagen in a triple helix structure (1). Small alpha 1 chains act on either end of the three chain arrangement to twist the chains into the helical structure (1). Type I collagen is a fibril protein and a structural component of bones, ligaments, tendons and the dermal layer of epithelial tissue (1).

The dbSNP database of single nucleotide polymorphisms lists 2966 insertions, deletions, multi-nucleotide polymorphisms, and single nucleotide polymorphisms for the COL1A1 gene of which 707 are validated by the 1000 genomes project. Mutations of the COL1A1 gene are linked to numerous degenerative bone disease such as osteogenesis imperfecta, Ehlers-Danlos syndrome, infantile cortical hyperostosis, and osteoporosis. 90% of Osteogenesis imperfecta type I (the least severe), II, III, and IV (the most severe) are coupled with COL1A1 or COL1A2 mutations (1). The disease can be associated with a single point mutation or many, with the most severe cases of the disease arising from mutations in the most highly conserved region of homologous proteins (3). The mechanism of degeneration is impaired inter-chain disulfide bonds and subsequent abnormal chain integration (3).

COL1A1 may also have suppressor function for particular cancers. In a study regarding Hepatocellular carcinoma (HCC), COL1A1 was found to be significantly downregulated at tumor sites (5). No chromosomal mutation could be found and the study identified promoter methylation as the mechanism of expression interference (5) Upregulation of COL1A1 may also be associated with particular forms of cancer. Reciprocal translocation of the COL1A1 gene on chromosome 17 with the platelet-derived growth factor beta gene on chromosome 22 is associated with the skin tumor dermatofibrosarcoma protuberans (4). This is attributable to the growth factor beta gene’s subsequent unregulated expression (4). The many disorders associated with up/down regulation and mutation of COL1A1 indicate its importance in eukaryote vertebrates. COL1A1 is highly conserved in homologous proteins for (now) obvious reasons.

1) [Xiran Wang](http://www.ncbi.nlm.nih.gov/pubmed/?term=Wang%20X%5Bauth%5D),[Yu Pei](http://www.ncbi.nlm.nih.gov/pubmed/?term=Pei%20Y%5Bauth%5D),[Jingtao Dou](http://www.ncbi.nlm.nih.gov/pubmed/?term=Dou%20J%5Bauth%5D),[Juming Lu](http://www.ncbi.nlm.nih.gov/pubmed/?term=Lu%20J%5Bauth%5D),[Jian Li](http://www.ncbi.nlm.nih.gov/pubmed/?term=Li%20J%5Bauth%5D),and [Zhaohui Lv](http://www.ncbi.nlm.nih.gov/pubmed/?term=Lv%20Z%5Bauth%5D)Genet Mol Biol. 2015 Mar; 38(1): 1–7 “Identification of a novel *COL1A1* frameshift mutation, c.700delG, in a Chinese osteogenesis imperfecta family”

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3) [Chessler SD](http://www.ncbi.nlm.nih.gov/pubmed/?term=Chessler%20SD%5BAuthor%5D&cauthor=true&cauthor_uid=8349697)1, [Wallis GA](http://www.ncbi.nlm.nih.gov/pubmed/?term=Wallis%20GA%5BAuthor%5D&cauthor=true&cauthor_uid=8349697), [Byers PH](http://www.ncbi.nlm.nih.gov/pubmed/?term=Byers%20PH%5BAuthor%5D&cauthor=true&cauthor_uid=8349697). [J Biol Chem.](http://www.ncbi.nlm.nih.gov/pubmed/8349697) 1993 Aug 25; 268(24):18218-25. “Mutations in the carboxyl-terminal propeptide of the pro alpha 1(I) chain of type I collagen result in defective chain association and produce lethal osteogenesis imperfecta.”

# 4) [Nakamura I](http://www.ncbi.nlm.nih.gov/pubmed/?term=Nakamura%20I%5BAuthor%5D&cauthor=true&cauthor_uid=26332510), [Kariya Y](http://www.ncbi.nlm.nih.gov/pubmed/?term=Kariya%20Y%5BAuthor%5D&cauthor=true&cauthor_uid=26332510), [Okada E](http://www.ncbi.nlm.nih.gov/pubmed/?term=Okada%20E%5BAuthor%5D&cauthor=true&cauthor_uid=26332510), [Yasuda M](http://www.ncbi.nlm.nih.gov/pubmed/?term=Yasuda%20M%5BAuthor%5D&cauthor=true&cauthor_uid=26332510), [Matori S](http://www.ncbi.nlm.nih.gov/pubmed/?term=Matori%20S%5BAuthor%5D&cauthor=true&cauthor_uid=26332510), [Ishikawa O](http://www.ncbi.nlm.nih.gov/pubmed/?term=Ishikawa%20O%5BAuthor%5D&cauthor=true&cauthor_uid=26332510), [Uezato H](http://www.ncbi.nlm.nih.gov/pubmed/?term=Uezato%20H%5BAuthor%5D&cauthor=true&cauthor_uid=26332510), [Takahashi K](http://www.ncbi.nlm.nih.gov/pubmed/?term=Takahashi%20K%5BAuthor%5D&cauthor=true&cauthor_uid=26332510).[JAMA Dermatol.](http://www.ncbi.nlm.nih.gov/pubmed/26332510) 2015 Sep 2 “A Novel Chromosomal Translocation Associated With COL1A2- PDGFB Gene Fusion in Dermatofibrosarcoma Protuberans: PDGF Expression as a New Diagnostic Tool.”

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