PREVENTION GENETICS
DISEASE PREVENTION THROUGH GENETIC TESTING

CLIA #: 52D1027685 ● CAP #: 7185561
AU ID: 1407125 ● NPI: 1114140571
3700 Downwind Drive
Marshfield, WI 54449 USA
Phone: 715-387-0484 Fax: 715-384-3661

Osteoarthritis with mild chondrodysplasia via *COL2A1* Gene Sequencing (Test #788)

Brief Description of Clinical Features: Osteoarthritis with mild chondrodysplasia (OMIM#604864) is characterized by early onset osteoarthritis and mild epiphyseal dysplasia and spinal involvement (Knowlton et al. *New Eng J Med* 322: 526-530, 1990). Individuals with this condition usually have normal or near normal height.

Genetics: Osteoarthritis with mild chondrodysplasia is inherited in an autosomal dominant manner. *COL2A1* is the only gene known to be associated with this condition. *COL2A1* encodes the alpha 1 chain of type II collagen, a major structural component of cartilaginous tissues. *COL2A1* mutations can also cause several different skeletal disorders, including Achondrogenesis type II/Hypochondrogenesis (OMIM#200610), Stickler dysplasia type I (OMIM#108300), Spondyloepiphyseal dysplasia congenita (SEDc; OMIM#183900), Spondyloepimetaphyseal dysplasia, Strudwick type (OMIM#184250), Spondyloperipheral dysplasia (OMIM#271700), Kniest dysplasia (OMIM#156550), and Platyspondylic lethal skeletal dysplasia, Torrance type (OMIM#151210). Most of reported *COL2A1* mutations associated with Osteoarthritis with mild chondrodysplasia are missense mutations.

Description of This Particular Test: This test involves bidirectional sequencing using genomic DNA of all coding exons of the *COL2A1* gene plus ~50 bp of flanking non-coding DNA on each side. As indicated, we will also sequence any single exon (Test #100, $190) in family members of patients with a known mutation, or to confirm research results.

Reference Sequences: **Genomic: NC_000012.11** **mRNA: NM_001844.4**
Protein: NP_001835.3 **mRNA and Protein: CCDS 41778.1**

Indications for Test: Candidates for this test are patients with clinical and radiographic findings consistent with Osteoarthritis with mild chondrodysplasia, and family members of patients who have a known *COL2A1* mutation.

Sensitivity of Test: Osteoarthritis with mild chondrodysplasia is a rare condition. Series of patients screened for *COL2A1* mutations in this particular condition have not been described in the literature. Our test has been designed to detect >99% of mutations published to date.

Turnaround Time: Maximum of 40 calendar days, although many tests are completed in 2-3 weeks.

Specimen Requirements: See page four of the Requisition Form.

Prices: **Sequencing of *COL2A1* gene** **$ 1990**

CPT Codes:

Sample Ascertainment x1	83890 $ 30	DNA Isolation x1	83891 $ 40
Amplification x47	83898 $660	Sequencing x47	83904 $990
Separation x1	83894 $140	Interpretation/Report x1	83912 $130

Accreditation: CLIA ID #: 52D1027685 (expires 1/18/13) (CAP#: 7185561, AU ID: 1407125 expires 12/20/12).

Contact: Dr. Ying Wang, ying.wang@preventiongenetics.com, www.preventiongenetics.com