LABOR RENNER

Gene test information

COL1A1 POLYMORPHISM OSTEOPOROSIS-RISK

- **Background**

  Osteoporosis is one of the most common diseases in old age. The pathogenesis of osteoporosis is associated with many factors: genetic, environmental, biomechanical, chronic disease states, and the effects of endogenous hormones.

  One of the best examined candidate genes is COL1A1, which encodes the alpha-1 chain of type I collagen, the major protein of bone. A polymorphism (Sp1 polymorphism) in the COL1A1 gene is associated with bone mineral density and osteoporotic fracture risk.

- **COL1A1 2046G>T genotypes**

Genotype	Frequency	Commentary
COL1A1 2046 GG (SS):	61%	Wildtype ("normal") genotype
COL1A1 2046 GT (Ss):	36%	Heterozygous carrier of the Sp1 variant. Modestly increased risk for osteoporosis. Osteoporotic fracture risk 26% higher compared to the wildtype genotype
COL1A1 2046 TT (ss):	3%	Homozygous carrier of the Sp1 variant. Increased risk for osteoporosis. Osteoporotic fracture risk 78% higher compared to the wildtype genotype

- **Indications for testing**

  Determination of the COL1A1 genotype may help to assess to individual risk for osteoporosis. Preventive therapy in subjects at risk may help to reduce the risk for osteoporotic fractures.

References:

Mann V, Ralston SH. Meta-analysis of COL1A1 Sp1 polymorphism in relation to bone mineral density and osteoporotic fracture. Bone. 2003;32:711-7.

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