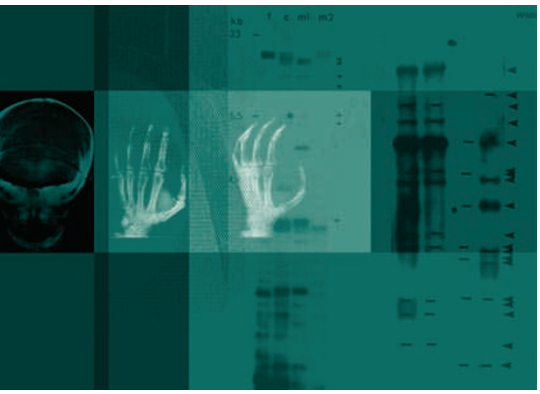
GENETICS

Polymorphisms in collagen genes affect fracture risk

Single nucleotide polymorphisms in the genes *COL1A1* and *COL1A2* have opposing effects on fracture risk, states a report published in *Bone*. The association of these variants with fracture risk is greatest during periods of predominantly appendicular bone growth.

Fractures are a common occurrence during childhood, with a peak incidence at 14 years in boys and 11 years in girls—around the time of peak height velocity when the rate of bone growth is at its greatest.



Blades *et al.* assessed the relationships between bone mass acquisition or childhood fractures and single nucleotide polymorphisms in the genes *COL1A1* and *COL1A2,* which encode the components of type 1 collagen—a major contributor to bone strength. In a noninterventional case–control study, the researchers recruited 394 children and adolescents aged 4–16 years who had been admitted to hospital as the result of an episode of trauma, for example, a fall. Of these, 52% had sustained a fracture.

Blades and co-workers determined the frequency of single nucleotide polymorphisms at the *COL1A1* Sp1 binding site and *COL1A2 Pvu*II restriction site and compared lumbar spine BMD measurements between individuals with previous fractures and controls according to genotype. Subgroup analyses were performed according to sex, pubertal status and site of injury.

No difference in the distribution of the *COL1A1* genotypes was found in the study cohort as a whole; however, possession of the Sp1 binding site *s* allele was associated with a threefold increase in the odds of fracture in prepubertal children (Tanner stage 1).

The results also showed that the *COL1A2 PP* genotype reduced the odds of fracture by about 50%. In individuals with Tanner stages 2–3 (early to midpuberty), this genotype was also associated with a significantly increased lumbar spine bone mineral content and areal BMD.

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Original article Blades, H. Z. *et al.* Collagen gene polymorphisms influence fracture risk and bone mass acquisition during childhood and adolescent growth. *Bone* **47**, 989–994 (2010)