Relation of PvuII site polymorphism in the COL1A2 gene to the risk of fractures in prepubertal Finnish girls.

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Genetic susceptibility to fractures may be detectable in early childhood. We evaluated the associations between the polymorphic PvuII site of the COL1A2 gene and bone properties assessed by different modalities (DXA; pQCT; QUS-2; Omnisense), bone turnover markers, and the occurrence of fractures in 244 prepubertal Finnish girls. Tanner stage and physical characteristics did not differ significantly among girls with different COL1A2 genotypes. The polymorphism was not significantly associated with different bone properties or any of the bone turnover markers when girls at Tanner stage I (prepuberty) and II (early puberty) were considered together, but there was a significant association with spine bone mineral content (BMC) and bone mineral density (BMD), as well as with speed of sound (SOS) (p < 0.05) when girls at Tanner stage I were considered separately, as a purpose to avoid the confounding effect that the pubertal growth spurt has on skeletal development. The distribution of fractures was different between the three genotype groups (p = 0.023). The P alleles were over-represented in girls who had been fractured at least once; 88% of them had at least one copy of the P allele (either PP or Pp). Girls with the PP genotype had 4.9 times higher relative risk for fractures than girls with the pp genotype (95% confidence interval, 1.4 to 17.4; p = 0.015). No significant difference was found between fractured and non-fractured girls in anthropometric measurements, physical activity, or bone mass. However, BMD of the spine and SOS at the radius and tibia were significantly lower in the fractured girls. We conclude that the COL1A2 polymorphism is associated with non-osteoporotic fractures in prepubertal girls independently of bone density.

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