PEDIATRIC RHEUMATOLOGY

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Genetic polymorphism of COL1A1 gene and bone mineralisation in juvenile idiopathic arthritis

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Background

Bone mineralization disturbances are common in children with juvenile idiopathic arthritis (JIA) and depend on inflammation, medications, lack of motion and genetic factors.

Aim

To evaluate the role of genetic markers in bone metabolism and mineralization in JIA children.

Methods

We included 196 JIA children, 81 boys and 115 girls. Bone mineralization parameters were detected by dual-energy X-ray absorptiometry of lumbar spine L_1-L_4. Bone biochemical markers were osteocalcine, C-terminal telopeptides (CTT), parathyroid hormone (PTH), Ca, Ca^{++}, P, total alkaline phosphatase (TAP) activity. We have detected Sp1 (rs1800012) and -1997G/T (rs1107946) polymorphisms in type I alpha-1 chain collagen gene (COL1A1).

Results

We revealed gender differences in Sp1 genotype distribution in children with low bone mineral density (LBMD): boys had GG genotype in 89.5% and girls in 54.2% (p=0.03). In boys GG genotype presence increased LBMD – OR=2.96 (95%CI: 0.59-14.9) compare in girls in which GG presence decreased LBMD – OR=0.56 (95% CI: 0.36-2.7). Also, children carried T allele (GT and TT genotypes) despite on higher inflammatory parameters had better mineralization dates. In total group children with GG genotype had higher osteocalcine (111.0 ±56.1 ng\ml and 85.9±39.9 ng/ml in GT+TT, p=0.02) and CTT levels (1.22±0.45 ng\ml and 0.99±0.38 ng\ml in GT+TT, p=0.02). In children, who have not been treated with steroids GG genotype was associated with lower BMD-Zscore in boys (-1.24±0.14SD and 0.29±0.98SD in GT+TT, p=0.006) and lower height in girls (142.9± 28.0 cm and 156.3±21.6 cm in GT+TT, p=0.025). In children with Tanner stage I GG genotype was associated with more rare LBMD (12.8% vs 36.4% in GT+TT, p=0.05) and with frequent LBMD in children with Tanner stage II-III (37.8% and 5.9% in GT+TT, p=0.01).

GG genotype of -1997G/T polymorphism was associated with lower Ca^{++} (1.1±0.11 mmol\l and 1.15±0.006 mmol/l in GT+TT, p=0.03), inorganic phosphate (1.67± 0.16 mmol/l and 1.57±0.22 mmol/l in GT+TT, p=0.04) and osteocalcine level (82.3±18.4 ng/ml and 115.5± 24.2 ng/ml, p=0.01) in children with Tanner stage II-III and lower BMD (0.84±0.14 g\cm^2 and 0.91±0.1 in GT+TT, p=0.04) and lower BMD-Zscore (-1.275±1.25SD and -0.5±1.0SD in GT+TT, p=0.009)

Conclusion

We have revealed different changes in mineralization and metabolism, associated with sex, Tanner stage and treatment due to COL1A1 gene polymorphisms in children.

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