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Osteoporosis/osteopenia in Crohn's disease patients the ATG16L1 variants

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Introduction: Osteopenia / osteoporosis are common in Crohn's patients. Ileal disease in Crohn's, especially fistulating / stricturing disease, or disease requiring small bowel surgery, can cause malabsorption which can cause osteoporosis. Since ATG16L1 polymorphisms have been shown to give a higher risk of ileal disease, one would expect a higher incidence of osteoporosis in patients with this genotype.

Methods: We have analysed the risk for osteoporosis / osteopoenia among Crohn's patients expressing the ATG16L1 polymorphism. Patients diagnosed with Crohn's disease through histological, radiological and endoscopic findings were recruited. Informed consent was obtained to take blood samples for genotyping for the rs2241880 variant of the ATG16L1 gene and to perform a DEXA scan. Genotyping for the rs2241880 variant involved: - DNA extraction - Gradient Polymerase Chain Reaction (PCR) - PCR - Quantitative PCR and High Resolution Melt - Restriction Fragment Length Polymorphism of PCR product

Results: Patients with the wild type allele (n=33) had a mean T score (hip) of -1.48, T score (spine) of -0.96, Z score (hip) of -0.75, and Z score (spine) of -0.45. Patients heterozygous for the rs2241880 polymorphism (n=44) had a mean T score (hip) of -1.68, T score (spine) of -0.94, Z score (hip) of -0.92, and Z score (spine) of -0.34. Patients homozygous for the rs2241880 polymorphism (n=6) had a mean T score (hip) of -1.84, T score (spine) of -0.14, Z score (hip) of -0.99, and Z score (spine) of -0.34. Using t-test, there was no statistical difference between the homozygous, heterozygous and wild type patients' hip T scores (p=0.314), Z scores (p=0.441), and spine T scores (p=0.751) and Z scores (p=0.822). Using $\chi 2$ test, the relationship between the 3 different variants (homozygous, heterozygous and wild type) and the risk of osteoporosis (T score <-2.5), osteopenia (T score: -1.0 to -2.5) and normal (Tscore >-1.0) was not statistically significant (p=0.978).

Conclusion: We found no significant difference between the T and Z scores of patients with ATG16L1 homozygous, heterozygous and wild type alleles. However, there is a trend in the mean T and Z scores at the hip with lower T and Z scores in patients with heterozygous/homozygous alleles. Such a trend is not present in the spine. While the authors can offer no explanation for this difference, studies on larger populations are needed to better investigate the relationship between ATG16L1 mutations and the risk of osteoporosis.