Background: Genetic factors accounts for about 90% pathogenesis of ankylosing spondylitis (AS), and may predict the disease progression. MRI is an important tool for evaluating disease progression and assessing treatment response in patients with AS.

Objectives: This study aims to define correlation between AS-associated genetic variations and MRI scores of AS patients in Chinese population.

Methods: A total of 62 AS patients treated with TNF blockers from Shanghai, China were recruited in this study. All of AS patients were evaluated for disease progression with MRI at baseline and six months’ follow-up. The changes of MRI scores were defined as the score of six months’ follow-up minus that of baseline. All of patients were examined with whole exon sequencing for genetic polymorphisms. Logistic regression analysis was performed and genetic variants with p < 1E-4 were considered as significant.

Results: The results showed that seven genetic variants significantly enriched in extracellular matrix pathways were associated with the changes of BME score of SIJ and spine. Four genetic variants regulating bone homeostasis, such as ABCA4, were significantly associated with the changes of fat metaplasia. Twelve variants were associated with the changes of erosion, one of which regulated expression of FAT4 (p-value = 6.8/1.4E-8). Mutation of FAT4 caused two bone related diseases and mouse with Fat4 knockout showed abnormality in insulin metabolism and skeleton (MGI database).

Conclusion: Overall, the results suggested that the polymorphisms of genes involved in extracellular matrix were associated with BME, and genetic variants involved in adipose and bone metabolism were associated with structural damage in AS patients.

REFERENCES:

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