Genomics and epigenomics in rheumatic diseases: what do they provide in terms of diagnosis and disease management?

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Most rheumatic diseases are complex or multifactorial entities with pathogeneses that interact with both multiple genetic factors and a high number of diverse environmental factors. Knowledge of the human genome sequence and its diversity among populations has provided a crucial step forward in our understanding of genetic diseases, identifying many genetic loci or genes associated with diverse phenotypes. In general, susceptibility to autoimmunity is associated with multiple risk factors, but the mechanism of the environmental component influence is poorly understood. Studies in twins have demonstrated that genetics do not explain the totality of the pathogenesis of rheumatic diseases. One method of modulating gene expression through environmental effects is via epigenetic modifications. These techniques open a new field for identifying useful new biomarkers and therapeutic targets. In this context, the development of ?-omics? techniques is an opportunity to progress in our knowledge of complex diseases, impacting the discovery of new potential biomarkers suitable for their introduction into clinical practice. In this review, we focus on the recent advances in the fields of genomics and epigenomics in rheumatic diseases and their potential to be useful for the diagnosis, follow-up, and treatment of these diseases. The ultimate aim of genomic studies in any human disease is to understand its pathogenesis, thereby enabling the prediction of the evolution of the disease to establish new treatments and address the development of personalized therapies.

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