Abstract

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Based on scientific progress in the research of human genome and the discovery of polymorphisms, which are involved in the interindividual differences in human population, there is also a growing interest in pharmacogenetics. It is a field combining pharmacology and genetics with the aim of identifying specific features that could explain the different responses of patients to treatment by clinically used drugs. Applying this knowledge could contribute to a simpler choice of medication for a particular patient and it could reduce the risk of side effects or poor response. In this diploma thesis I dealt with the latest scientific knowledge on pharmacogenetics in rheumatology, in particular the rheumatoid arthritis. From available studies, reviews, and meta-analyzes that have been published, I summarized current data on the relationship between polymorphisms and disease modifying drugs (DMARDs) used for the treatment of this disease. The largest amount of data was found on the most commonly used methotrexate. Further, the work examines the leflunomide and other substances, including biological agents. Studies show promising association of polymorphismsin case of biological DMARDs and genes such as TNF-α (-308 A / G), ATIC 347C> G and RFC-1 in case of methotrexate, CYP2C19 * 2 in case of leflunomide or NAT2 in the treatment with sulfasalazine. Apart from azathioprine, where the FDA recommends genotyping of patients before starting treatment, these studies are relatively small, sometimes with conflicting results and must be verified by a larger number of patients.