Chapter 4: Biomarkers, genetic association and genomic studies Mehmet Tevfik DORAK & Yusuf YAZICI

Mehmet Tevfik DORAK

Professor in Health Sciences School of Health Sciences Liverpool Hope University Hope Park Liverpool L16 9JD United Kingdom e-mail: <u>dorakm@hope.ac.uk</u>

Yusuf YAZICI

Division of Rheumatology NYU Hospital for Joint Diseases New York University New York USA

Abstract

Rheumatoid arthritis (RA) is a common autoimmune disorder which shows clinical heterogeneity. IT has multiple treatment options and there is individual variation in response to treatment. These features make RA an ideal condition to develop biomarkers for its pre-clinical detection, diagnosis, subtyping, prognostic stratification and selection of most optimal treatment. While a number of markers have been assessed for their biomarker quality, currently no marker has the statistical properties of a biomarker to be considered as a good classifier. In this chapter, a general review of biomarkers is followed by a detailed discussion of biomarker candidates for various aspects of RA. It is unlikely that a single marker will ever be sufficiently powerful as a biomarker, but combinations of clinical, biochemical, genetic, epigenetic, proteomic and metabolomic markers have the strongest potential to fulfill the requirements of biomarkers. Given the high heritability of RA and the progress in methodology of genome-wide association studies, genetic markers are the most promising group to be developed as biomarkers, in particular when epigenetic markers become more widely used. It is possible that in the near future, biomarkers with documented clinical utility will be available for use in clinical decision making and will most probably use multiple omics platforms.