DNA and RNA Profiling in Human Blood

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METHODS IN MOLECULAR BIOLOGY ™

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Methods and Protocols

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Preface

Blood samples are widely used as biological specimens for diagnostic or research purposes. There are many examples of important disease markers which can be investigated using peripheral blood. In the context of blood transfusion and immunohematology, blood itself is the target of investigation with regard to the determination of blood groups and the screening for antibodies. Furthermore, blood samples represent the main source of genetic material, i.e., DNA and RNA, for analyzing gene mutations, polymorphisms, and expression at the molecular level.

The development of novel bioanalytical technologies for complex and quantitative molecular analysis of DNA, RNA, proteins, and cell functions led to the introduction of '-omics' terms such as genomics, transcriptomics or proteomics. These molecular profiling approaches have opened up a broad field of research and may help to identify further disease markers in blood. Recently developed techniques, such as microarrays or bead arrays, represent the basis of high-throughput multiplex DNA typing and RNA profiling. Such methods have been applied already to blood cells or plasma and many of them were adapted to the special characteristics of blood cells. Thus, protocols have been developed to achieve diagnostic systems in the fields of genotyping for blood cell antigens including Human Leukocyte Antigens (HLA) and blood groups. The diagnostic systems are of great importance in blood transfusion and organ transplantation. Furthermore, special protocols were adapted to particularities of certain blood cell types such as platelets or reticulocytes in order to address questions in the field of gene expression analysis.

The aim of DNA AND RNA PROFILING IN HUMAN BLOOD is to bring together established, standardized, and recently developed protocols for complex and/or high-throughput DNA and RNA profiling. This book consists of two sections, Part I: *DNA Profiling for Blood Cell Antigens*, and Part II: *RNA Profiling in Blood Cells*. In Part I, a number of methods and protocols describe high-throughput multiplex approaches for genotyping of various blood cell antigens (*see* **Chapters 1–5, 8,** and **9**). Blood grouping by DNA typing also includes a step-by-step protocol for prenatal RhD determination using of maternal plasma (*see* **Chapter 11**). Other DNA protocols describe modern techniques for SNP typing other than blood cell antigen SNPs (*see* **Chapters 6, 7, 10** and **12**) that may serve as examples to establish protocols for the own purposes.

Part II is focused on RNA profiling methods and protocols that have been adapted to the special characteristics of certain blood cell types such as platelets (*see* **Chapters 16–18**), reticulocytes (*see* **Chapter 20**) or megakaryocytes (*see* **Chapter 19**). Furthermore, methods and protocols are included to describe recently developed techniques which have been applied to blood samples (see **Chapters 13, 14, 21,** and **22**) or which may be applied to RNA samples of any type of biological source (see **Chapters 12** and **15**).

This book summarizes contributions from leading international experts in the fields of DNA and RNA profiling. As editor of this volume, I am very grateful indeed to themfor

their willingness to provide an insight into their knowledge and to provide the detailed step-by-step protocols. I also wish to thank Steffanie Bickelhaupt and Daniela Griffiths for considerable editorial and secretarial assistance.

Peter Bugert

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