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Editorial

Fourteen volumes of *Monographs in Human Genetics* were published between 1966 and 1992. Since then a plethora of experimental data obtained by the new molecular techniques has led to paradigmatic changes in understanding the mechanisms operating in human heredity. Currently human genetics presents as a highly diversified field covering an ever increasing number of topics, ranging from basic research to medical practice. This calls for a specialized forum where the rapid advances in human genetics are reviewed by experts of different fields. In response to these developments, the traditional book series *Monographs in Human Genetics* will be revived with two volumes per year, focusing on important hereditary diseases, their molecular basis, their clinical impact and their eventual treatment. With its concise but highly informative reviews, *Monographs in Human Genetics* provides essential reading not only for researchers but also for physicians and students interested in specific genetic diseases. All articles published in this book series are reviewed according to classical standards.

The present volume is devoted to 'Fanconi Anemia', a chromosome instability disorder whose molecular basis has been all but elucidated in recent years. By their very nature, monograph-types of publications are more comprehensive and synthetic rather than hot-off-the-press reports. A case in point is the topic 'Fanconi anemia' where the pace of gene discovery has accelerated during recent years. While this volume was in preparation three new disease causing genes have been discovered, the most recent of which (FANCI) was too recent to be included. Even though monographs may not cover the very last developments in a given field, their undisputed value arises from their unhurried and in

depth treatment of a given subject that can hardly be achieved with the usual publish-or-perish types of publications. As such, monographs fulfill the important task to remind the reader that any type of scientific progress has its roots in a multifaceted landscape of prior insights and achievements that need to be collected and preserved in order to provide fertile ground for future growth.

I would like to thank all the authors for their interesting contributions, the Editors Holger Hoehn and Detlev Schindler for their invaluable support and help in the organization of this book, and the Publisher Thomas Karger for having offered the opportunity to reinitiate this book series.

Michael Schmid
Würzburg, January 2007

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Preface

The revival of the series ‘Monographs in Human Genetics’ starts with a volume on Fanconi anemia, a rare inherited disease that was discovered 80 years ago by an eminent Swiss pediatrician, Guido Fanconi, then working in Geneva. The Editors of the volume gratefully acknowledge the initiative by Karger Publishers (Basel, Switzerland), and by Michael Schmid of the University of Würzburg, Germany, in getting this new series started. The volume not only pays tribute to Guido Fanconi’s discovery of a disease that teaches us much about the connection between genetic instability, cancer and premature aging, but also pays tribute to Traute Schroeder-Kurth who in 1964 discovered the chromosome instability in Fanconi anemia, and who has contributed numerous important observations on the clinical and genetic aspects of the disease. On the occasion of Traute Schroeder-Kurth’s 75th birthday we convened a small meeting in Würzburg in order to honor her seminal contributions to the diagnosis and pathophysiology of Fanconi anemia. The talks presented at the Schroeder-Kurth symposium provide the basis for this volume. We are very grateful to the contributing authors for complementing and updating their previous oral presentations. Following introductory chapters that include a historical account, exemplary case reports, and a summary of the current status of FA genes and their mutations, there are two chapters on neoplasia in FA, three chapters reviewing diagnostic approaches in FA, including prenatal diagnosis, and one chapter each on the phenomenon of revertant mosaicism or ‘natural gene therapy’ and on hematopoietic stem cell transplantation as the only curative approach in FA. The final three chapters deal with evolutionary aspects of the FA genes with special emphasis on the avian genome, with recombinational

types of DNA repair in FA, and with the establishment of a test system for the Rad51 recombinase in homology-directed DNA repair. Even though it is impossible to cover all aspects of Fanconi anemia within the space available for such a volume, we hope that it may serve as useful introduction to a disease that provides unique insights into the complex network of genomic maintenance systems which protect us from cancer and premature aging.

We dedicate this volume to George M. Martin, M.D., Professor Emeritus of Pathology and Genetics at the University of Washington, Seattle, USA, on the occasion of his 80th birthday. Like Traute Schroeder-Kurth with respect to Fanconi anemia, George Martin was one of the first scientists to recognize the intimate relationship between genetic instability, cancer and aging by studying the Werner progeria syndrome. Very early on he was convinced of the model character and unique value of the rare human chromosomal breakage syndromes. He emphasized that these rare experiments of nature would provide unprecedented insights into the complex mechanisms of DNA damage recognition and repair which are essential for long lived, warm-blooded species such as ours. George Martin encouraged us to study the human caretaker gene syndromes, including Fanconi anemia, and we owe him much in terms of motivation, mentorship, and guidance.

Detlev Schindler
Holger Hoehn
Würzburg, January 2007