The fourth, completely revised edition of this classical reference and textbook presents a cohesive and up-to-date exposition of the concepts, results, and problems underlying the theory and practice in human and medical genetics. In the 10 years since the appearance of the third edition, many new data and insights have emerged for understanding the genetic and genomic basis of development and function in human health and disease. Human genetics, with its emphasis on molecular concepts and techniques, has become a key discipline in medicine and the biomedical sciences. While the chapters of this edition are written by multiple experts, the general spirit of this book highlighting problems, approaches, and history continues. The fourth edition has been extensively expanded by new chapters on timely topics such as epigenetics, pharmacogenetics, gene therapy, cloning, genetic epidemiology, and databases for basic and clinical genetics. In addition, a multi-chapter section giving an overview on comparative genetics and the main model organisms useful for human genetics (mouse, dog, worm, fly, fish) has been introduced. This book will be of interest to all human and medical geneticists, scientists in all biomedical sciences, physicians and epidemiologists, as well as to graduate and postgraduate students who desire to learn the fundamentals of this exciting and fascinating field.

Michael R. Speicher, MD, is internationally acknowledged for his research and teaching in medical genetics. He is professor of human genetics and chairman of the Institute of Human Genetics at the Medical University of Graz (Austria) where he manages an extensive genetic counseling and diagnostic program. His research concerns the analysis of chromosome structure, chromosomal instability, and consequences of chromosomal aberrations for phenotype and disease susceptibility. He is the author of more than 100 research articles and multiple book chapters.

Stylianos E. Antonarakis, MD, DSc, is internationally known for research and training in genetic medicine and genome analysis. He is professor of genetic medicine at the University of Geneva in Switzerland. His research includes the molecular basis of monogenic disorders, genome analysis of chromosome 21 and Down syndrome, functional characterization of conserved genomic sequences, and genomic variability. A highly cited author of 570 scientific papers, he directs the clinical genetics and diagnostics program in Geneva.

Arno G. Motulsky, MD, is internationally recognized for research, training, and policy setting in medical genetics. He is considered a founder of pharmacogenetics. In 1957, he established a pioneering medical genetics division and genetics clinic. His research included hematological genetics, human population genetics, and heart disease genetics. He authored over 400 articles and with F. Vogel as sole authors coauthored three editions of this text. Multiple awards include the National Academy of Science (USA), Institute of Medicine, American Academy of Arts and Sciences, and American Philosophical Society.

ISBN 978-3-540-37653-8

springer.com