ADVANCES IN HUMAN GENETICS 15

Edited by

Harry Harris

and

Kurt Hirschhorn

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Harry Harris

Harnwell Professor of Human Genetics
University of Pennsylvania, Philadelphia

and

Kurt Hirschhorn

Herbert H. Lehman Professor and Chairman of Pediatrics
Mount Sinai School of Medicine of The City University of New York

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CONTRIBUTORS TO THIS VOLUME

Charles Auffray
Institut d'Embryologie du CNRS et du Collège de France
Nogent sur Marne, France

Arthur L. Beaudet
Departments of Pediatrics and Cell Biology
Baylor College of Medicine
Houston, Texas

Hans-Georg O. Bock
Department of Pediatrics
Baylor College of Medicine
Houston, Texas

Svend O. Freytag
Department of Pediatrics
Baylor College of Medicine
Houston, Texas

Michelle M. Le Beau
Department of Medicine
Section of Hematology/Oncology
University of Chicago
Chicago, Illinois

James V. Neel
Department of Human Genetics
University of Michigan Medical School
Ann Arbor, Michigan

William E. O'Brien
Departments of Pediatrics and Biochemistry
Baylor College of Medicine
Houston, Texas

Janet D. Rowley
Department of Medicine
Section of Hematology/Oncology
University of Chicago
Chicago, Illinois

Michael M. Skolnick
Department of Human Genetics
University of Michigan Medical School
Ann Arbor, Michigan

Moyra Smith
Department of Pediatrics
University of California at Irvine
Irvine, California

Jack L. Strominger
Department of Biochemistry and Molecular Biology
Harvard University
Cambridge, Massachusetts

Tsung-Sheng Su
Department of Pediatrics
Baylor College of Medicine
Houston, Texas

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Preface to Volume 1

During the last few years the science of human genetics has been expanding almost explosively. Original papers dealing with different aspects of the subject are appearing at an increasing rapid rate in a very wide range of journals, and it becomes more and more difficult for the geneticist and virtually impossible for the nongeneticist to keep track of the developments. Furthermore, new observations and discoveries relevant to an overall understanding of the subject result from investigations using very diverse techniques and methodologies and originating in a variety of different disciplines. Thus, investigations in such various fields as enzymology, immunology, protein chemistry, cytology, pediatrics, neurology, internal medicine, anthropology, and mathematical and statistical genetics, to name but a few, have each contributed results and ideas of general significance to the study of human genetics. Not surprisingly it is often difficult for workers in one branch of the subject to assess and assimilate findings made in another. This can be a serious limiting factor on the rate of progress.

Thus, there appears to be a real need for critical review which summarizes the positions reached in different areas, and it is hoped that *Advances in Human Genetics* will help to meet this requirement.

Each of the contributors has been asked to write an account of the position that has been reached in the investigations of a specific topic in one of the branches of human genetics. The reviews are intended to be critical and to deal with the topic in depth from the writer's own point of view. It is hoped that the articles will provide workers in other branches of the subject, and in related disciplines, with a detailed account of the results so far obtained in the particular area, and help them to assess the relevance of these discoveries to aspects of their own work, as well as to the science as a whole. The reviews are also intended to give the reader

some idea of the nature of the technical and methodological problems involved, and to indicate new directions stemming from recent advances.

The contributors have not been restricted in the arrangement or organization of their material or in the manner of its presentation, so that the reader should be able to appreciate something of the individuality of approach which goes to make up the subject of human genetics, and which, indeed, gives it much of its fascination.

HARRY HARRIS
The Galton Laboratory
University College London

KURT HIRSCHHORN
Division of Medical Genetics
Department of Pediatrics
Mount Sinai School of Medicine

Preface to Volume 10

This is the tenth volume of *Advances in Human Genetics* and some fifty different reviews covering a very wide range of topics have now appeared. Many of the earlier articles still stand as valuable sources of reference. But the subject continues to move forward at an increasing speed and its vitality is indicated by its remarkable recruitment of young investigators. New areas of research which could hardly have been envisaged only a few years ago have emerged, and quite unexpectedly dicoveries have been made in parts of the subject which only recently had come to be thought as full explored. So there continues to be a need for authoritative and critical reviews intended to keep workers in the various branches of this seemingly ever-expanding subject fully informed about the progress that is being made and also, of course, to provide a ready and accessible account of new developments in human genetics for those whose primary interests are in other fields of biological and medical research.

We see no reason to alter the general policy which was outlined in the preface to the first volume. We believe that it has served our readers well. The subject seems to us to be just as exciting and intellectually stimulating and rewarding as it did when this series was first started. We expect the next decade of research in human genetics to be as innovative and productive as the last and our aim is to record its progress in *Advances in Human Genetics*.

HARRY HARRIS
University of Pennsylvania, Philadelphia

KURT HIRSCHHORN
Mount Sinai School of Medicine of the City University of New York

NOTE ABOUT ADDENDUM

To make the volume as up-to-date as possible, each author was given the opportunity to write a short Addendum at the time he or she received the page proofs of that particular chapter. This allows for any important new material to be presented at the latest possible time in the publication process. The Addendum is presented at the end of the book, beginning on page 291.

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Chapter 1

Chromosomal Abnormalities in Leukemia and Lymphoma: Clinical and Biological Significance

Michelle M. Le Beau and Janet D. Rowley
Department of Medicine
Section of Hematology/Oncology
University of Chicago
Chicago, Illinois 60637

INTRODUCTION

Cytogenetic analysis of human tumors is one of the most rapidly progressing and exciting areas of cancer research. Major advances in our understanding of the specificity of some of the abnormalities observed have occurred within the last 10 years with the application of both chromosome banding techniques and improved methods of cell culture. Thus, the hypothesis put forward by Boveri at the turn of the century, that an abnormal chromosome pattern was intimately associated with the malignant phenotype of the tumor cell, can now be tested with the substantial hope of obtaining a valid answer (Boveri, 1914).

The study of the chromosome pattern in human leukemias can be divided into two periods, each one covering 10 years. The first lasted from 1960 to 1970, and the second from 1970 to 1980. During the first period, the chromosome abnormalities seen in leukemic cells were identified without banding, and therefore they include only structural rearrangements that resulted in a change in morphology and abnormal modal chromosome numbers. The most significant observation during this initial period was the identification of the Philadelphia (Ph^1) chromosome in leukemic cells from patients with chronic myelogenous leukemia (CML)