EXPERIENTIA

Volume 42/No. 10 Pages 1069–1178 15 October 1986

Human genetic research

The Editors wish to thank Dr E. M. Bühler for coordinating this review.

Introductory remarks

The latter part of this century has witnessed a markedly rapid development in the area of molecular biology, and in the following collection of papers we shall try to document some of the major advances made in several key areas of this exciting discipline.

The review opens with H.J. Evans' Introduction which provides a very comprehensive overview of recent developments in the different areas of human genetics. It is obvious from this Introduction that one technique has revolutionized genetics as no other method before: recombinant DNA. There is practically no paper in which these new techniques are not at least mentioned as a major breakthrough in the respective field. Human genetics has long been confined to the observation of pedigrees and populations with respect to phenotypic traits, as experimentation with humans was not possible. In the last few decades, however, advances in cell biology, biochemistry, cytogenetics, and immunology have enabled geneticists to study the human genome more directly.

In the following selection of papers, we shall try to review these developments without going into excessive detail. The papers have been designed to give the interested reader working in science an impression of what has been achieved in the 20th century. We have tried to get capacities in their respective fields from all over the world to participate in this review and I would like to thank all contributors for their support and cooperation.

Dr. E.M. Bühler
Coordinator

New trends in human genetic research – An introduction and overview

by H. J. Evans

Medical Research Council, Clinical and Population Cytogenetics Unit, Western General Hospital, Edinburgh (Scotland)

Key words. Mapping; linkages; mutant alleles; antenatal diagnosis; genetic therapy; genetics and cancer; mutagenesis.

A history of advances in human genetics would recall a long list of enterprising individuals who made remarkable discoveries through astute observation and penetrating analysis of nature's experiments in human variation. Advances in modern human genetics depend to no less an extent on individual enterprise and ingenuity in studying such variation, but the rapid pace in the extension of our knowledge of the genetics of man has been stimulated by developments in two areas. First, the introduction of new approaches and new techniques in cellular and molecular biology and in biochemistry and cytogenetics, which have opened up new ways of analysing, dissecting, probing and indeed modifying the human genome. Second, the evergrowing realisation outside the genetics laboratory, or clinic, that a considerable burden of human disease is attributable to our genetic inheritance and that modern advances enable us to detect an increasing variety of such diseases early in fœtal development and in some instances provide a basis for successful therapy.

Looking at the state of human genetics at the beginning of the 1960s it was evident that the techniques of biochemistry that were available at that time had contributed greatly to our understanding of the nature of some of the mutations that were then known to be involved in inherited disease, as well as to knowledge of the frequencies and distribution of allelic forms of a range of