Further Section

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Book Reviews


The segregation of the new genetic subspeciality, pharmacogenetics, took place at much the same time as another important subspeciality, cytogenetics, was established, and developments have been equally impressive and explosive within both fields in the last ten years. The fact that variations in the response to drugs could be genetically determined got its first substantial consolidation by the explanation of the causes of the rare, but life-threatening reactions to the muscle relaxant suxamethonium, observed in some individuals. This opened the new field of research, and many other similar, genetically determined ‘abnormal’ drug reactions have been described since then, but studies of the first example of this type have continued and are far from being completed. The extent of this intensive research is evidenced by the present volume where more than 250 pages are needed to give a survey of the present knowledge about pseudocholinesterases, primarily with reference to the genetics and the clinical aspects. A short outline of pharmacogenetics introduces the book, the biochemistry of the cholinesterases is discussed and the methods used in these studies are clearly described in the end of the book. The main emphasis is placed on a detailed account of the many variants which have been observed and the present genetic interpretation. Studies of esterases in animals are also reviewed. The chapters on the clinical importance bring a detailed survey of the pseudocholinesterase activity in a number of pathological states, and the contents of the section on the implications of this genetically determined variation in anaesthesiology should be fully familiar to all those who use this group of muscle relaxants in their daily work. The extensive list of more than 500 references is a very valuable part of this important work. Mogens Hauge, Copenhagen


It is well known that ionizing radiation produces chromosome abnormalities among many other biological effects. The Edingurgh group of cytogeneticists demonstrated this in a group of patients treated with X rays for spondylizing ankylitis nearly ten years ago. It was hoped that estimation of chromosome damage could be used as a measure of the biological effects of radiation. But the main obstacle has been the immense number of cells that has to be observed in the individual patient and the difficulty in doing this in an unbiased way. In the present volume most of the relevant problems are discussed. Perhaps the sections on automatic chromosome analysis will attract most general interest (Rutovitz and Wald). The machines used to-day are very complicated and not at all suitable for practical purposes, but there can be no doubt about their success in the future.
In their concluding remarks the editors point out the many difficulties that should still be overcome before chromosome analyses can be used as a quantitative tool in radiation biology. Especially, very little is known about the absorbed radiation dose to the lymphopoietic system.

Cytogeneticists should study this book in order to get a survey of the work done in this field of radiation biology, and also to get an impression of the importance that should or should not be attached to the incidental finding of structural chromosomal abnormalities in single cells. Anders Frøland, Copenhagen


It can be calculated that in the Swedish population of 7.8 million people there are around 159,000 diabetics, 59,000 males and 100,000 females. In the years 1961 to 1963 3,324 deaths with diabetes as a primary cause and 7,273 deaths with diabetes as a contributory cause were registered in Sweden among a total of 226,806 deaths. The aggregate morbidity risk of diabetes up to age 50 is 2.1% for males, 1.9% for females; up to age 90 it is 6.5% for males and 13% for females. The overall excess mortality among persons with clinically manifest diabetes does not exceed 15%, for diabetics with age of onset under 15, however, mortality is about six times population mortality.

Insulin therapy has been applied to 90% of patients with onset under 40 and to 60% with onset after 40. There is a correlation between the duration of diabetes, on the one hand, and the prevalence and severity of the late complications, on the other. The data seem to indicate that no treatment hitherto used will decisively improve or retard the development of retinopathy in diabetics.

The fertility of diabetics before the onset of disease is not higher than fertility in corresponding groups of the general population, and after onset the number of children born to female diabetics is far below expectation.

Among different genetic hypotheses discussed sex-linked dominance is felt to fit in best with the epidemiological data. On the other hand the authors must admit that earlier family studies do not support this hypothesis.

The book has been sponsored by The Medical Research Council of the Swedish Life Office, and it has been one of the purposes to supply reliable data for life-table statistics and rating of diabetes mellitus risks in life and health insurance. The book offers much valuable epidemiological information on diabetes and can be highly recommended to all with an interest in this field. B. Harvald, Copenhagen

W. Leydhecker (ed.): Glaucoma Tutzing Symposium. S. Karger, Basel 1967, VI and 265 p., 93 fig., 54 tab. Price: S.Frs. 75.–, US Dollar 18.–, 137 s. 6 d.

In connection with the XX International Congress of Ophthalmology the Third International Symposium on Glaucoma took place in Tutzing Castle in 1966. This book is a record of the papers and the discussion which followed.

The general subject of this Symposium was the discussion of difficulties and errors in theoretical and practical work on glaucoma and outstanding scientists participated. Most of the papers deal with ophthalmological problems and the epidemiology and genetics of glaucoma were only discussed briefly. The hereditary types of reaction to topical application of corticosteroids is presented in a paper by M.F.
Abnormality, who demonstrated that the reactions could be divided into three groups, corresponding to three genotypes. Ernst Goldschmidt, Odense

Book Reviews


In a study of 776 children with severe visual handicaps attending special educational institutions in England and Wales, the authors have attempted to ascertain the diagnosis in every case as accurately as possible. The material was investigated for family history, birth rank, birth weight, and various laboratory findings such as screening of urine for abnormal products and chromosome studies.

Nearly half the children had genetically determined lesions, most of them determined by single gene inheritance. At least nineteen autosomal dominant, thirty autosomal recessive and ten sex-linked genetic entities play a part in the causation of childhood blindness and a lot of genetical and other details are presented. The biological investigations which were performed have not proved very productive.

Fifty per cent of the blindness is acquired and the largest group of children owe their handicap to perinatal difficulties. It is amazing that retrolental fibroplasia accounted for visual loss in 177 children, while infectious causes were rare. Prematurity plays an important role not only in cases of retrolental fibroplasia but also in cases of congenital cataract and optic atrophy.

An enormous amount of work was involved in preparing the analysis and an extensive summary of the literature is given. The book is not only of value for those concerned with blind and partially sighted children but also to those concerned with human genetics and genetic counseling. Ernst Goldschmidt, Odense


The last ten years have witnessed a rapidly growing interest in teratology, especially in Britain and the US. The main explanation may be found in the recent gains of knowledge within a number of biological disciplines which in time may be able to throw more light upon the etiology and pathogenesis of the induction of congenital abnormalities. This applies specifically to work on the morphology and function of the cell, the effects of ionizing radiation at the cellular and molecular level and on the mechanisms involved in transplacental passage. Stimuli have undoubtedly also come from the increased use of ionizing radiation in medicine and industry and the demonstration of teratogenic effects of thalidomide and some viral affections. In addition, many animal experiments have shown strong teratogenic influence of a number of drugs used in medical therapy. The first results of research on ‘late effects’ of intrauterine exposure to ionizing radiation, including qualitative and quantitative studies of causes of death in such animals, should also be mentioned in this context. It seems clear that teratology must be accepted as a discipline of its own.

The present volumes indicate very clearly the growing interest in teratology and its clinical implications. The very praiseworthy initiative to introduce this series, which intends to bring annual surveys of advances in teratology, has been taken by Professor Woollam, Cambridge. The first volume brings chapters on Down’s syndrome, the inactivation of the X-chromosome in mammals and on anomalies which may be caused by viral infections. The vast field of the immunological aspects of developmental biology would perhaps have gained in clarity if more subspecialists had contributed. The section on congenital malformations induced experimentally
by ionizing radiation pays little attention to the problem of greatest practical importance: the effects of small doses. The final chapter deals with the possible teratogenic effects of drugs. It is stressed that an uncontrolled teratological experiment is instituted whenever a drug is given to a woman in the first trimester of a pregnancy.

The second volume is largely devoted to an introduction to the specific problems and methods of teratology. Stimulating theories concerning the possible effects of purely mechanical, intrauterine influences are launched. Results are given of chromosomal studies in 466 cases of spontaneous abortions where 120 severe aberrations were found. Intrauterine growth retardation is discussed in detail; although a wealth of facts is given, the present knowledge is clearly incomplete. A valuable chapter on the problems related to the use of inbred experimental animals in teratology should also be mentioned. The technical standard of the volumes is very high. The present series will undoubtedly meet a need which has been felt by workers in experimental teratology as well as by many clinicians. It is warmly recommended as a reference book of high quality. It may also be of value as a textbook in the more advanced medical schools which include teratology in their curriculum. It seems safe to predict that the increased and systematic research in this field will produce results of the greatest practical value in the prevention of congenital abnormalities, in addition to the increased knowledge about the principles underlying the induction and development of congenital defects and abnormalities.

Lars Jacobsen, Copenhagen


Recent advances in teratology have demonstrated the diversity of approach which is needed in efficient studies of teratological problems and have stressed that teratology now covers such a wide area of the biology that it must be considered a separate discipline which should take a more central position in modern medical research and education. The present volume reflects the great interest of a clinician in the principles underlying the induction of congenital disorders and abnormalities. The most important chapters seem to be those which deal with the damage caused in the fetus by bacteria, viruses and chemical agents, including drugs of more or less common use. Other important factors such as prenatal oxygen deficiency, toxoplasmosis and prenatal exposure to ionizing radiation have also been included in the survey.

The author has largely mastered the many difficulties involved in outlining the extent of the problems and the nomenclature and systematics of prenatal pathology. Minor criticism may of course be raised in relation to some of the sections which is quite natural as no single author could be expected to be able to cover all aspects of prenatal disorders completely, and some collaboration with specialists within the various fields might have added to the value of the volume. As an example, fetal damage caused by ionizing radiation may be mentioned. This subject is dealt with in two different sections, ‘Vergiftungsembryopathien’ and ‘Fetale Vergiftungen’. Too much emphasis has been put on casuistic reports of single human cases, and too few references are given to the systematic studies in genetically homogeneous strains of animals. It would have been valuable if more of the available knowledge about dose-effect relations in animals had been included.

In general, it is found that the book gives a valuable survey, of interest especially to paediatricians and gynecologists. Many important teratological aspects have been
presented in a way which may inspire continued and more profound research. For those who are already actively engaged in experimental or human teratology, the main value of the book is probably found in the very extensive collection of references.  

L. Jacobsen, Copenhagen

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Book Reviews


Medical geneticists who are involved in epidemiological studies may find many useful hints in this stimulating book, written by a statistician. It may be considered as a critical review of the methodological problems of epidemiological investigations with one of the more important chronic diseases, varicose veins, taken as an example. The great variety of approaches to an answer to the simple question of the prevalence of this disease has been analysed and the inadequacies and limitations of the methods employed have been discussed in detail. As comparative epidemiological studies may also be expected to be of growing value in genetic analyses, information on the methodology of such investigations as presented in the present book is definitely of great importance in increasing the value and usefulness of the results.

M. Hauge, Copenhagen


Extensive and systematic studies of non-human primates are still rare, but facilities have recently been made available in the US for investigations on a large colony of chimpanzees. The present report gives a survey of the results of serological and immunological studies, primarily on these but also on some other primates. Dr. Wiener and Dr. Moor-Jankowski give a very useful survey of the blood groups of non-human primates, and a number of valuable papers discuss the Gm, the Inv and the Tf groups as well as the haemoglobin and the gamma-globulins, their structure and immunology. Much of the knowledge obtained from serological studies of the other primates will obviously be of immediate value to human serology and genetics.  

M. Hauge, Copenhagen


Medical geneticists with interest in neurology and ophthalmology should be aware of this valuable volume (in French, with summaries in English) which includes some general surveys and a number of discussions of important hereditary syndromes, based mainly on reports of single cases. Among the surveys special attention is given to the ophthalmological anomalies in cases of chromosomal aberrations; among the cases discussed, the Marinesco-Sjögren and the Morquio syndromes, optic atrophies and dystrophies of the cornea may be mentioned. Two interesting pairs of probably monozygous female twin pairs, discordant for colour blindness, are reviewed in the light of the Lyon hypothesis, and they are taken as support for the randomness of inactivation of one of the X chromosomes in the female. The volume is of a high technical standard, but a subject index would have increased the value of the book tremendously  

M. Hauge, Copenhagen