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# THE TEACHING OF GENETICS IN THE UNDERGRADUATE MEDICAL CURRICULUM AND IN POSTGRADUATE TRAINING

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## EXPERT COMMITTEE ON HUMAN GENETICS

Geneva, 28 November - 4 December 1961

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# **THE TEACHING OF GENETICS IN THE UNDERGRADUATE MEDICAL CURRICULUM AND IN POSTGRADUATE TRAINING**

## **First Report of the Expert Committee on Human Genetics**

The Expert Committee on Human Genetics met in Geneva from 28 November to 4 December 1961. Dr P. M. Kaul, Assistant Director-General, opened the meeting on behalf of the Director-General and welcomed the participants. He referred to the significance of genetics in practical health work, pointing out the contribution that it is making to the understanding of endogenous factors in disease causation and of the complex interactions that exist between man and his environment. Recent developments in this field have been so rapid that many medical men find it difficult to appraise the present status of human genetics, its importance to the practising physician, and the contribution that it can make at present to public health thinking. Dr Kaul expressed the hope that the Expert Committee in considering these questions would recommend how the necessary knowledge of genetics could best be presented to different kinds of medical personnel, and how instruction in genetics should be integrated into the medical curriculum.

Dr J. Böök was elected Chairman and Dr M. Lamy, Vice-Chairman ; Dr F. Clarke Fraser and Dr J. A. Fraser Roberts were elected Rapporteurs.

### **1. THE IMPORTANCE OF GENETICS IN THE MEDICAL SCIENCES**

The gene is at the basis of life, and it is self-evident that instruction about its nature and function should be included in the education of those who intend to follow any biological discipline. There are, however, even more cogent reasons for teaching genetics to medical students. Knowledge of the means by which genes influence the development and function of the living organism has advanced to the point where it not only forms an

essential background to a proper understanding of disease but has important practical applications of which physicians must be aware if they are to provide high standards of medical care.

The rapid expansion of genetics as a medical discipline has stemmed largely from :

(1) the increasing control of infectious and other extrinsic causes of disease, which has greatly increased the average life span and enlarged the proportion of medical practice devoted to diseases that are determined, to a greater or lesser degree, by genes ;

(2) the discovery of the human blood groups and the applications of this knowledge in the fields of blood transfusion and treatment of haemolytic disease of the newborn ;

(3) the striking advances in molecular biochemistry which have provided essential technical tools for investigation of gene structure and function and which have led to the description of many inherited diseases in specific biochemical terms ;

(4) the recent public concern about the genetic effects of ionizing radiation ;

(5) the development of improved cytological techniques for the study of human chromosomes, and the consequent discovery that chromosomal aberrations cause a number of pathological conditions.

These developments have led to a greatly increased demand for instruction in genetics among clinicians and research workers in essentially all fields of medicine and medical biology, but this need has not yet been reflected in the curricula of most medical schools. Thus, the paradoxical situation now exists that the teaching of genetics in most schools is either inadequate or non-existent, yet geneticists are receiving many requests from medical practitioners, research workers and medical groups, for lectures in genetics and for collaboration in solving problems.

The science of genetics seeks knowledge of the gene from many aspects, including :

(1) the means by which the information is coded in the chromosomal material ;

(2) the means by which genes affect development and function, both normal and abnormal ;

(3) factors altering gene structure—i.e., gene mutations ;

(4) the causes and pathological effects of chromosomal aberrations ;

(5) the variability in expression of genes in different genetic backgrounds and in different non-genetic environments ;

(6) the distribution of genetic differences within families ;

(7) the distribution of genetic differences within and between populations.

Aspects 1, 2 and 3 give important insights into the nature of human disease, and aspects 4, 5, 6 and 7 already have direct practical applications to patient care.

### 1.1 The present status of genetics as related to medicine

The concerted efforts of geneticists, biochemists, and physicists have yielded a remarkable degree of insight into the mechanisms by which genetic information is coded for transmission from generation to generation and determines (in conjunction with the environment) the nature of the organism. Evidence from a multitude of living things, ranging from viruses to man, has shown that—except in the ribonucleic acid (RNA) viruses—the genetic code resides in the sequences of nucleotides in the chromosomal deoxyribonucleic acid (DNA), that RNA molecules are synthesized in the nucleus under the influence of the DNA in such a way that they carry a corresponding sequence of base pairs, and that this RNA migrates to the sites of protein synthesis in the cytoplasm, where it controls the sequence of amino-acids being assembled into proteins. This knowledge underlies the concept of “molecular diseases”. For example, the cause of sickle-cell disease is a change in a single gene which leads to an alteration of one specific amino-acid out of the approximately 300 that constitute the globin fraction of the haemoglobin molecule.

The concept of molecular disease can be extended to many other variants, be they malformations or metabolic diseases: the “inborn errors of metabolism”, for instance, which are being identified at the rate of about three a year, are characterized by the fact that they show simple Mendelian inheritance, and that each results from a specific enzymatic defect. Thus, when the geneticist establishes the genetic basis for a disease, this is an indication for the biochemist that there is probably an underlying specific biochemical defect. Identification of the defect provides the basis for sound treatment and may also improve the precision of genetic counselling through the biochemical detection of normal carriers of the gene. Furthermore, study of the effects of a block at a specific step in a metabolic sequence can provide a great deal of information about normal metabolic pathways. This approach has been effectively utilized, for instance, in phenylketonuria.

Most of our understanding of the nature of the genetic code and its role in controlling protein specificity comes from microbial genetics and this work has far-reaching implications for such problems as the pathogenesis of cancer, the aging process, controlled alteration of the genetic material and even space biology.

On the developmental level, the use of mutant genes in mice and other organisms provides an opportunity, still largely unexploited, to analyse the pathogenesis of malformations and to study the mechanisms by which genes control the complicated processes of morphogenesis. Inbred strains of various experimental organisms (particularly mice) are now being utilized to study the etiology and pathogenesis of neoplasms and degenerative diseases such as arteriosclerosis, the genetic components of reproductive performance and longevity, and the biological effects of various environmental factors such as ionizing radiation.

Rapid progress is also being made in the analysis of factors controlling the frequency of genes in populations. The interaction of mutation, selection, population size, migration and patterns of mating has an important bearing on the distribution of inherited diseases in human populations. The concepts of population genetics have also helped to clarify other important medical problems, such as the increasing resistance of bacteria to antibiotics, and of insects to insecticides.

These few examples demonstrate the importance of genetic knowledge to the medical practitioner and its importance to further advances in many branches of medical science.

## 1.2 Applications of genetics in clinical practice

In spite of the growing interest in medical genetics, physicians are often not sufficiently aware of the many ways in which genetic knowledge can have direct clinical applications. These include :

### 1.2.1 *Prediction of recurrence risks for specific diseases*

There are a great many pathological conditions that arouse in the patient and his near relatives anxiety that the same condition may occur again in the family. Information about the genetic (or non-genetic) nature of the disease and the statistical probability of recurrence, if given sympathetically, is almost always helpful in aiding the family to understand the disease and to adjust to the situation. In the experience of members of the Committee, the average risk estimate given by the counsellor is frequently lower than that suspected by the family and therefore reassuring.

A great many diseases are now known that follow the Mendelian laws of inheritance. Although each of them may be rare (with some notable exceptions in particular areas), there are so many that any physician is likely to encounter an appreciable number in his practice, and he should be aware that the probability of occurrence of the same disease in the sibs, children or other near relatives of the patient can be predicted, often very precisely. For a great many other diseases which do not follow simple patterns of inheritance, estimates of risks of recurrence in various groups of relatives have been obtained empirically and are available for use in

advising families. It must be recognized, however, that, even in the case of diseases with predominantly non-genetic causes, irregularities in gene expression and other complications sometimes make the estimation of recurrence risks difficult. The physician who is aware that such complications exist is in a position to know when to seek consultation with a geneticist.

#### 1.2.2 *Genetics as an aid in diagnosis and prognosis*

Taking the family history is a time-honoured procedure of medicine, but full advantage of it can be taken only if the physician is aware of its value and is familiar with the recent developments in medical genetics. For instance, the knowledge that a patient has a mentally retarded sib with cataracts can lead the physician to identify the cause of the patient's vomiting and failure to thrive before the irreversible changes of galactosaemia occur, and a family history of consanguinity may be the clue to the etiology of deaf-mutism.

It is particularly important that the physician should recognize that the manifestations of the same mutant gene may vary widely in different individuals. Thus, the recognition of "formes frustes" of a syndrome in a patient's relatives may be a clue to the patient's diagnosis—e.g., ectopia lentis in a relative may suggest the diagnosis of dissecting aneurysm of the aorta in a patient who does not have the characteristic features of Marfan's syndrome. On the other hand, many genetic diseases tend to display a similar development, response to therapy, and final outcome when occurring in different members of the same family. The detailed family history can then be used as a guide in evaluating the prognosis and determining the best therapy for the actual patient.

#### 1.2.3 *Cytogenetics in medical practice*

The recent discovery that certain abnormalities in man are caused by gross chromosomal aberrations has opened a new field for investigation which has already yielded results of practical value, both diagnostic and predictive. Such aberrations are found in an appreciable portion of patients with mental retardation, with amenorrhoea, and with sterility. Sex chromatin and sex chromosome analyses are important aids to the diagnosis of abnormalities in sexual development; chromosomal analysis in cases of mongolism makes it possible to distinguish families with a high risk from those with a very low risk of recurrence.

#### 1.2.4 *Drug-induced diseases*

Many instances are now known of genetically determined susceptibility to particular drugs, the consequences being of great practical importance. Thus, a high proportion of individuals in certain populations have a

deficiency of glucose-6-phosphate dehydrogenase. Under natural conditions the possession of this sex-linked gene is of little moment, apart from the development of favism. Certain drugs, however, such as primaquine or sulfonamides, induce a haemolytic anaemia in males who have this gene (or in females who have it in double dose), making indiscriminate use of such drugs in a susceptible population a hazardous procedure.

When this disease could be diagnosed only by an abnormal response to particular drugs or to fava beans, it usually appeared to occur sporadically, and was only occasionally detected more than once in a family. Measurement of enzyme activity shows that there is, in fact, a clear-cut genetic basis, but clinical disease only appears in a very special environment, namely, exposure to the drug. This raises the question of how many other diseases that are familial, but do not appear to have a simple genetic basis, may result from single mutant genes causing metabolic defects which express themselves only in certain environments.

In the white and coloured populations of South Africa there are estimated to be about 8000 persons suffering from a hereditary porphyria due to a dominant gene. Again, under natural conditions the possession of the gene is not a serious disadvantage as witnessed by the fact that all 8000 sufferers are descended from a single couple who married in 1688: a fantastic rate of gene multiplication. But certain drugs, notably barbiturates, and particularly a barbiturate anaesthetic, may induce very severe reactions. Many fatalities have been reported. It is now routine at some South African hospitals to test for porphyria before giving an anaesthetic. Increasingly, the possessors of the gene are being provided with cards emphasizing that the dangerous drugs must on no account be administered.

Another example of a genetically determined difference in reaction to a drug concerns the rate of breakdown of isoniazid. This has an important bearing on the clinical management of patients being treated with the drug—slow inactivators develop toxic reactions at the dosages necessary to maintain adequate blood levels in fast inactivators, and it may be that the slow inactivators respond well to treatment at doses inadequate for fast inactivators. It can be confidently predicted that many other inherited variations in reaction to drugs will be found and that other important applications to medical practice will result.

#### 1.2.5 *Immunogenetics*

The discovery of blood groups, with its immediate application to blood transfusion, was one of the first and most important examples of the implications that individual inherited differences may have for practical medicine. Another outstanding example was the discovery that haemolytic disease of the newborn is caused by maternal iso-immunization, due to special mother-child combinations of normal inherited traits.

New red cell antigenic systems continue to be discovered and "groups" for white cells, platelets, and some serum fractions are also being recognized—for instance the antigenic differences between gamma globulins give rise to a number of "types" and are controlled by at least two genetic systems. Blood groups, along with other simply inherited traits, also provide important medico-legal evidence in cases of disputed paternity and personal identification. Furthermore, immunogenetics has contributed greatly to our knowledge of gene distribution in populations and to a new rational approach to physical anthropology.

Experiments in animals have firmly established that the rejection of transplanted tissues between individuals of different genotypes is due to immunogenetic incompatibility; in mice, a number of genetic systems controlling histocompatibility, some of them expressing themselves also as red cell agglutinogens, have been identified. In man, the successful permanent take of transplanted kidneys between identical twins clearly shows that the present practical limits to homografting are not surgical but immunogenetic.

Further advances are to be expected, for instance with regard to autoimmune disease (e.g., the specificity of some haemolytic autoantibodies, and the distribution of specific antibodies in the relatives of patients with lupus erythematosus, rheumatoid arthritis, and thyroiditis).

#### 1.2.6 *Genetics of specific diseases*

Many diseases have a significant genetic component and failure to take account of genetic implications may make many researches in a wide variety of fields less profitable than they could otherwise be. Often a genetic inquiry can profitably be incorporated in the more general study of a particular disease entity. There is little doubt that it will be increasingly recognized that the incorporation of genetic research in a wider scheme is an excellent approach: each specialist helps the others, and in economic terms the return is much greater than if clinicians or pathologists on the one hand and geneticists on the other investigate the same conditions independently and from their own points of view only.

#### 1.2.7 *Genetics, preventive medicine, and public health*

It is now possible to prevent the harmful effects of a gene in a number of diseases by applying prophylactic measures to genetically susceptible individuals—e.g., exchange transfusion for haemolytic diseases of the newborn, the dietary treatment of galactosaemia, and colonic resection for multiple polyposis, to mention only a few. This approach can be fully exploited by physicians only if they are aware that the diseases concerned have a genetic basis and if they know how to predict which relatives are likely to be susceptible or, alternatively, when and where to seek help in

making such predictions. This aspect of preventive medicine will become progressively more important with increasing knowledge of gene action in some more common diseases, such as coronary artery disease, different types of diabetes, and many types of mental illness or mental defect.

Genetics is also important to the epidemiologist because of the high proportion of individuals with specific inherited diseases in some geographic areas and ethnic groups. The high frequency of thalassaemia in certain Mediterranean populations may be cited as an example. The dangers of large-scale administration of primaquine, sulfonamides, or barbiturates in certain populations have been referred to in section 1.2.4. The genetics of human pathogens and disease vectors also have an important bearing on public health—for example, the appearance of penicillin-resistant strains of staphylococcus, DDT-resistant mosquitoes, and virulent strains of influenza virus. Schools of public health and government public health agencies would benefit from having a geneticist on their staffs.

#### 1.2.7.1 *Collection of data*

One of the most difficult tasks in medical genetics is the collection of reliable data on the frequencies of specific pathological conditions and their ethnic and geographical variations. Such measurements are vital to a proper evaluation of the role of genes in human health and disease, and in particular to the estimation of mutation rates. The Committee felt that more attention should be given to the collection of data in a form that can be used for such purposes wherever data concerning human disease, reproductive performance, or longevity are being routinely recorded. Consultation with geneticists by those responsible for designing hospital record-keeping systems, registries for handicapped children, national vital statistics, etc., would be helpful in this regard. Further relevant considerations will be found in the first report of the WHO Expert Committee on Radiation.<sup>1</sup>

#### 1.2.7.2 *Mutation and mutagenic agents*

The sudden changes in the genetic material known as mutations provide the genetic variability necessary for evolution, but are also a source of a largely undetermined amount of human disease. The fact that ionizing radiation increases the frequency of mutations has recently become a source of considerable public concern which, incidentally, has been a great impetus to research in genetics. The extensive literature on the subject needs no documentation here. To evaluate the significance of the mutational load and increases in this load on the health of future generations, a great deal more needs to be known about mutation rates in man, the factors that alter them and the fate of fresh mutations in the population.

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<sup>1</sup> *Wld Hlth Org. techn. Rep. Ser.*, 1959, 166.

In addition to radiation, there are a number of possible mutagens about which we know virtually nothing. The mutation rate in *Drosophila* varies with temperature; caffeine, antibiotics and other drugs, and a number of industrial chemicals are mutagenic in experimental organisms and possibly in man. This is a problem that deserves attention from geneticists, physicians, and public health authorities.

## 2. THE UNDERGRADUATE TEACHING OF GENETICS

The preceding sections of this report have endeavoured to show why the teaching of genetics to the medical undergraduate is essential. It is a basic discipline in biology, an exercise in scientific method, and is fundamental to the understanding of the subjects the undergraduate is studying.

At an early stage in his preclinical training the medical student needs to be familiar with the hereditary code, the mechanisms by which it controls the synthesis of proteins and particularly of enzymes, and its relation to the development of normal traits and inherited disease. He must become familiar with chromosome and gene behaviour in meiosis and recombination—the alphabet of the Mendelian scheme. He must be led to an intelligent appreciation of the determination of the phenotype—the finished product—through the interaction of genetic and environmental factors. He must appreciate the importance of mutation—gene and chromosomal—and its relation to selection in determining gene frequencies in populations.

At a later stage, during his clinical years, and preferably rather late in his clinical years, he should be required to use this knowledge in the study and understanding of genetically controlled diseases, many of which are common conditions, and of the more numerous instances in which genetic differences are potent agents in determining whether or not disease develops. If he knows less than this, his equipment is seriously incomplete; indeed, with the popularization of scientific knowledge in the press, and especially perhaps on the television screen, he is in danger of knowing less about particular points than some of his more intelligent patients.

It is significant that a similar conclusion was reached by a previous WHO Expert Committee<sup>1</sup> who in their discussion of "The Teaching of the Basic Medical Sciences in the Light of Modern Medicine" stated: "If [the student] is made aware of the genetic factors in disease he will be better prepared not only for the clinical handling of patients but also for raising the general health of the community he serves. Modern medical teaching and research problems require the participation of qualified geneticists within the medical faculty."

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<sup>1</sup> *Wld Hlth Org. techn. Rep. Ser.*, 1961, 209, 27.

### 2.1 Preliminary training

The Committee agreed that students entering medical school should already have a thorough grounding in biology, including the elementary principles of genetics. In many areas of the world this would require a marked improvement in the teaching of biology, and in particular, the inclusion of genetics in the curriculum of biology teachers.

### 2.2 Preclinical training

At some time before the student reaches the clinical years of medical school he should have a course in basic genetics, including an introduction to the special methodology of human genetics. This should normally be a part of the medical school curriculum and, provided there has been satisfactory teaching at the preliminary level, should take a minimum of 15-20 hours. If there has been no teaching at the preliminary level, this time should be proportionately increased. The distribution of the hours devoted to genetics would of course depend on the local situation, but the Committee felt that the elementary principles should be presented very early, since they have applications to some of the other basic courses, such as biochemistry, and that the teaching of applications to medical genetics might better be done at a later stage of the preclinical training, when the student has become familiar with the concepts of disease. The course should be compulsory, and the students' knowledge should be tested by formal examinations.

The following headings cover the minimum content of an adequate preclinical course :

#### 2.2.1 *The chemistry of genes and chromosomes*

Introducing the course by a description of the chemistry of DNA and RNA and the role of these compounds in the control of protein synthesis plunges the student at once into one of the most exciting recent developments of biology, gives him an immediate insight into possible clinical applications of genetics, and should arouse in him a desire to learn more about genetics.

#### 2.2.2 *Gene transmission in man*

Having learned what the gene is and a little about its relation to protein synthesis, the student is then shown how it is transmitted from parent to offspring, and is introduced to some of the complications of the genetic analysis of human pedigrees.

### 2.2.3 *The actions of genes in development*

A discussion of the role of the gene in normal and pathological development follows this somewhat mathematical preceding section—partly to satisfy the student's natural desire to deal with human disease, but, even more important, to impress upon him how ubiquitous the known effects of the gene are and how, if understood, they may be controlled.

### 2.2.4 *Cytogenetics*

The student is now introduced to the important recent findings in human cytogenetics and to the fact that alterations in the chromosomes (as well as in individual genes) may have profound effects on the organism.

### 2.2.5 *Gene and chromosome mutation : spontaneous and induced*

Having learned something about the chemistry of genes and chromosomes, about their transmission within families, and about their effects on development, the student is then told how often changes in genes and chromosomes arise spontaneously (including a discussion of methods of determining these rates) and about agents (physical and chemical) that cause mutations to occur. Opportunity is taken here to inform the student of the potential genetic dangers of the careless use of X-rays, various drugs, food additives, etc.

### 2.2.6 *Population genetics*

The importance of population genetics for an understanding of geographic medicine and for a better understanding of the relation of populations to each other is indicated and the basic principles are presented. If time permits, some elements of the study of selection may be introduced to help explain population differences and to reinforce earlier illustrations of the interaction of the genotype with environment.

### 2.2.7 *Elements of microbial genetics*

This subject, if not adequately covered by instruction in medical microbiology, should be included in the genetics course, both for its practical implications (e.g., resistance to antibiotics) and its theoretical ones (e.g., the production of a change in host heredity by a viral parasite or other infection and its possible significance in the pathogenesis of neoplastic diseases).

### 2.2.8 *Practical application : diagnosis, treatment, and genetic counselling*

Although it is recognized that the students will have had little or no clinical experience at this stage of their training, the course concludes with

a discussion of the applications to diagnosis and treatment in order to relate the basic material to practical medical work.

### 2.2.9 *Demonstrations and laboratory work*

It is highly desirable that some laboratory work and other practical activities be included in the course to impress the students with the reality of the problems discussed and to introduce them to specific techniques. These practical activities will vary according to local facilities and the case material available. The following items are provided as useful examples :

1. Determination in all students and in a few pairs of twins of :
  - (a) blood groups (ABO and, if possible, Rh and MN systems) and colour blindness as examples of characters determined by single gene differences ;
  - (b) phenylthiocarbamide taste threshold, to illustrate the action of modifiers on a single gene difference, resulting in a bimodal distribution ;
  - (c) a quantitative trait such as stature ;
  - (d) sex ratios of the sibs of the male and female students respectively, illustrating the bias caused by inclusion of the proband.
2. Microscopic observations of :
  - (a) sex chromatin and human chromosomes ;
  - (b) blood smears from cases of hereditary anaemias (sickling, elliptocytosis, Pelger anomaly, etc.).
3. Demonstration of simple biochemical phenotypes such as electrophoretic separation of haemoglobins and serum haptoglobins ; glucose-6-phosphate dehydrogenase deficiency.
4. Presentation of patients with inherited diseases, and their pedigrees, chosen to illustrate the principles discussed. These might include examples of dominant inheritance (hyperdontia, polydactyly), autosomal recessive inheritance (fibrocystic disease of the pancreas, albinism), sex-linked inheritance (haemophilia, muscular dystrophy), mutation (chondrodystrophy), pleiotropy (Laurence-Moon-Biedl-Bardet syndrome), penetrance and expressivity (Marfan's syndrome), and the "pedigree of causes" (sickle-cell disease, galactosaemia). The effects of chromosomal aberrations may be illustrated by mongolism, Klinefelter's syndrome, and Turner's syndrome.
5. Groups of two or three students could each investigate one case of an inherited disease or abnormality, prepare a pedigree, and write a short report for presentation in class, including a review of some pertinent genetic literature.

Items 1, 2 and 3 can be included during a special laboratory period and the remainder as demonstrations during the lecture classes.

### **2.3 Clinical training**

In the clinical years the specific applications of genetics to human diseases should be taught. This can best be done in collaboration with one or more clinical departments, such as paediatrics, neurology, ophthalmology, etc., where suitable cases are available, and the relation of the genetic principles to the clinical problems can be presented as a matter of practical importance. Clinics or institutes of genetics may also provide valuable teaching material. A method that has already demonstrated its efficacy makes use of 5 three-hour sessions, in which cases of genetically determined diseases are presented and a clinician, geneticist and other appropriate specialists discuss the manifold aspects of the disease in question.

## **3. POSTGRADUATE TRAINING IN MEDICAL GENETICS**

Postgraduate training in genetics is needed at three levels :

- (1) as part of the regular training in the various medical and surgical specialities ;
- (2) more advanced training for those who consider genetics as a valuable adjunct to their research or practice, but do not wish to make it a full-time occupation ;
- (3) advanced training to qualify candidates for full-time research and teaching in medical genetics.

### **3.1 Medical and surgical specialists**

The importance of some familiarity with genetic concepts in the practice of the majority of the medical and surgical specialities is now so well recognized that, at least in some countries, there is an active demand at this level for instruction in genetics. At present, and unfortunately perhaps for some years to come, most of those who have entered training in a speciality have had little or no previous introduction to genetics. In this case, a course with a content similar to that outlined in section 2.2 would be desirable, but the presentation might make more use of the seminar method, guided literature reviews, and special projects, with emphasis on the particular aspects and diseases appropriate to the speciality concerned.

When adequate instruction in genetics has become routine in undergraduate medical schools, it will be possible for teaching at the postgraduate level to be reduced in amount and increased in depth.

### 3.2 Medical specialists with a particular interest in genetics

There are some medical specialists who have a particular interest in medical genetics and who wish to make genetics within their own speciality their research interest. The Committee was of the opinion that these specialists should secure all the encouragement possible ; their work should add greatly to the development of the subject, especially in clarifying the genetics of particular diseases.

In addition to the instruction in genetics recommended for undergraduates and for specialists in general, the chief requirements for training specialists with a particular interest in genetics are facilities for study within a department of medical genetics. There would seldom be a sufficient number of such candidates at any one time and place to justify a formal course, but a period of about six months to a year could be used to familiarize them with research methods. They could undertake under supervision a research project of their own and could also take advantage of the seminars and other courses available. Sometimes the candidate could join the department on a part-time basis, in which case the period of study would have to be longer. It is anticipated that such people would thereafter resume the full practice of their speciality, devoting what time they found possible to genetic research. In addition, for some time to come they could usefully help in genetic teaching at centres where trained full-time medical geneticists were not available.

### 3.3 Specialists in medical genetics

These are the people who wish to devote themselves entirely to medical genetics and who will provide the teachers and research workers so urgently needed if genetics is to take its proper place in the medical curriculum.

The training of a medical geneticist should cover the fields outlined for the training in genetics of medical undergraduates, with each topic developed in considerably more detail. In addition, the Committee agreed that a well-trained teacher and practitioner of medical genetics should be conversant with the genetics, not only of man, but also of other animals and of micro-organisms. This will enable him to appreciate the contributions to the theory of genetics that can be made by the study of these organisms, to understand the importance of the genetics of pathogens in the control and treatment of disease, to recognize the usefulness of mammals other than man in the analysis of genetically controlled diseases of man, and to familiarize himself with the special techniques required to use these organisms in his research should he find it useful to do so.

It is further recommended that the student be made familiar with as many of the tools of modern biological research as are available in the

institute where he is training or in other institutes to which he may be sent for special training.

In those countries where the profession of a speciality is controlled and certain requirements have to be met, medical genetics should be placed in the same position as are the other specialities, and qualification as a specialist in medical genetics should be governed in the same way. A thesis or its equivalent in published work should be required.

The following programme for training medical geneticists, which is already in operation at one university, is presented as an example. The Committee did not, of course, suggest that this is the only way to train medical geneticists, or that all the facilities referred to below would have to be available initially.

*Initial training* (common to all graduate students in biological sciences) : Courses are offered in organic chemistry and biochemistry, cytology, physiology, mathematics and statistics, and in the use of modern aids to research. The latter acquaints the students with the principles and applications of advanced microscopy (i.e., phase-contrast, ultra-violet, electron) ; zone electrophoresis ; chromatography (paper, column, gas) ; radioisotope studies ; cell-culture methods ; and other techniques that may be found relevant. It is emphasized that the student does not become expert in the use of any of these at this time, but he does learn their principles and their uses.

*Subsequent training* : the student must take an extensive course in basic genetics (if he has not already done so), perform simple experiments with *Drosophila*, paramoecium and bacteria or yeast or viruses, and become familiar with the techniques of breeding mice. Following this, more specialized courses are taken in the problems and methods of human genetics, population genetics, developmental genetics, etc.

In addition to being familiar with the principles, terminology and tools of biometry, biochemistry, microbiology, and immunology, the student is required to take advanced training in one or more of these subjects. This is made possible by the co-operation of several departments of the faculties of science and medicine, i.e., biology, chemistry, statistics, preventive medicine, biochemistry, microbiology, and pathology.

Each candidate is required to present a thesis based on his own research. The research programme determines the field or fields chosen by the student for intensive training in theory and methodology.

The course work and the research programme are supplemented by attendance at the usual clinics and ward rounds pertinent to the student's medical interests, lectures by visiting professors, seminars, visits to other laboratories, and attendance at meetings of the national genetics society.

In addition all students are required to teach as laboratory assistants for a minimum of one year.

#### 4. THE NEED FOR TRAINED TEACHERS IN MEDICAL GENETICS

It must be admitted that in most places the kind of programmes suggested in this report would not at present be possible because of the lack of trained teachers. The most appropriate teacher would be a specialist in medical genetics with the training outlined in the previous chapter. An effort should be made to accelerate the preparation of such specialists by sending interested graduates to some of the centres where this training is already offered, and in this regard the co-operation of national and international institutions concerned with such matters is highly desirable.

Since it is important that inclusion of genetics in the medical curriculum should not be unduly delayed by the lack of fully trained teachers, the following interim measures are suggested. The preclinical course might be given, as a part-time activity, by a geneticist not belonging to the medical school staff. The clinical training might be in the care of a clinical teacher with a special interest in the genetic study of diseases. Short training courses now available at a number of centres would help to improve the preparation of such teachers.

The Committee felt that, in order to ensure the maintenance of a satisfactory teaching programme in genetics, it would be highly desirable, as trained personnel become available, to have a chair or department of medical genetics at each medical school, with a staff to participate in the teaching and to create interest by their research activities. It is desirable that such a department should be located in the university hospital, or that hospital facilities should be available.

#### 5. CONCLUSION

It is quite clear that the development of genetics in this century has clarified considerably the understanding of all biological sciences. Since modern medicine is based on these sciences, a knowledge of genetics is indispensable for the understanding of man in health and disease and in his relation to different types of environment.

Genetics is, perhaps, the most important element in biology, with a large number of specific ramifications in clinical pathology and practical medicine. Many examples of the importance of genetic knowledge for medical practice and research have been given in the previous sections of

this report. The Committee was convinced that a medical curriculum that does not include instruction in genetics at different levels (preclinical and clinical) will produce physicians who are severely handicapped in their understanding and handling of many pathological conditions, whether they are dealing with individual patients or with the same conditions from the wider point of view of public health.

The Committee urged that all medical educators and others who determine the details of medical education in different parts of the world should take fully into account the necessity of ensuring the best possible teaching in genetics for all students of medicine.

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