GENETICS IN MEDICAL EDUCATION*

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For the first time in South Africa, an entire plenary session of the Medical Congress is being devoted to genetics. This fact, of itself, is very significant. It means that the organized medical profession of our country has recognized the important part played by heredity in the aetiology, treatment and prevention of disease. To my mind, it is more than an expression of passing interest; it implies a demand on the part of the medical profession to know more about genetics and to have human and clinical genetics systematically taught at our medical schools.

Less than a year ago, the first conference on human and medical genetics to be held in South Africa took place at the Witwatersrand Medical School.[1] On that occasion I urged that South African medical schools should take immediate steps to introduce formal instruction in human genetics into the medical curriculum.[2] I am pleased to say that, to my knowledge, at least three of our medical schools are at present enquiring into the possibility of establishing courses on the subject. Since that conference, too, a South African doctor has attended an international study course on human genetics held in Copenhagen and Aarhus last year, under the auspices of the World Health Organization. He has returned to the Republic equipped to assist in establishing such teaching facilities.[3]

The WHO course at Copenhagen followed the meeting in Geneva in 1961 of a WHO expert committee to enquire into the teaching of genetics in the undergraduate medical curriculum and in postgraduate training. The report of this committee was published in 1962.[4] It is instructive to compare parts of it with the proceedings of a teaching institute held by the Association of American Medical Colleges 8 years before (October 1954);[5] and with the yet earlier report (1948) of the Henry Cohen Committee of the British Medical Association.[6]

In the 1948 British document—a 150-page report on 'The training of a doctor'—one looks in vain for the slightest reference to genetics. The 1954 American report recognizes the value of genetics in medical practice, but the subject still fills only a small corner of a large conference on pathology and microbiology. In the 1962 international document genetics has become the object of a major independent enquiry and of emphatic recommendations to medical educators everywhere. In 1954 the question is still being asked: Has genetics earned a place in the medical curriculum? In 1962 the affirmative answer is taken for granted and pages are devoted to the precise definition of the place of genetics. In 1954 12 - 14 lecture hours are recommended in the entire medical curriculum; by 1962 the plea has changed to one for a preclinical course of 15 - 20 hours, followed by a clinical course of, say, five 3-hour sessions, in all about 30 - 35 hours. In 1954 there is argument whether *practical* tuition should be included. By 1962 there is no question but that case histories, laboratory work and clinical demonstrations are deemed necessary. Such has been the change in the climate of medical opinion from 1948 through 1954 to

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1962.

From these international developments, from the fact of the present symposium being held, from the enquiries that are being conducted by some of our medical schools —it is not really necessary to justify the claim of genetics to be included in the medical curriculum.

At least one survey has been made in an attempt to estimate the size of the load of genetic disability in man. This is a 10-year survey by Stevenson[7] of the population of Northern Ireland. He found that at some stage in their lives about 4% of all infants born suffer ill-health owing to a genetic defect. This is a minimum estimate, as Stevenson has recognized. Dobzhansky[8] has pointed out that Stevenson did not include abortions and stillbirths, some of which are due to genetic causes, and which occurred in at least 14% of the recordable pregnancies. Furthermore, his figure did not include some fairly common and many less common conditions in the aetiology of which genetic factors are involved, such as diabetes, schizophrenia, and many others, the total frequency of which is fairly high. Stevenson has calculated that no less than one-quarter of the hospital beds in Northern Ireland are occupied by genetically handicapped persons; and that at least 6% of consultations with medical practitioners involve such cases. This gives an idea of the *expressed* genetic load in a human population; and there is a much larger *concealed*, or *latent*, genetic load that is *not* expressed, but may declare its presence later in life or in a subsequent generation.

The Position in South Africa Compared with Other Countries

Let me turn now to what has been done in South Africa. As an illustration, I shall cite the example of one medical school, that of the University of the Witwatersrand.

Until 1947 - 48 no systematic teaching of human genetics was included in the medical curriculum, a sporadic lecture or passing reference being the entire genetic training with which our doctors and dentists were speeded on their way. In 1947 a series of lectures on the genetics of congenital abnormalities featured in the course of clinical anatomy given to 3rd-year medical students. A year later 7 - 8 lectures on general genetics were included for the first time in the zoology course given to medical and dental students; this course has continued until the present day. Then human genetical tuition appeared at postgraduate levels, in the courses for the Diploma in Child Health (1949), for the Diploma in Psychological Medicine (1953), and for the Higher Dental Diploma (1954). At about the same time a short series of systematic lectures in *human* genetics was first given as part of the anatomy course to 2nd-year medical students. At present, over and above the basic 7 - 8 lectures in the 1st year of study, about 6 genetics lectures are given to 2nd-year students, and others in clinical anatomy and in various clinical departments, especially psychiatry, in which 6 genetics lectures are given in the 4th year of study. The total coverage of human and medical genetics in the entire medical course is thus about 20 hours, but its organization is higgledy-piggledy, uncoordinated and theoretical. No department or subdepartment is specifically entrusted with the task of planning and organizing the genetics course; many aspects go by default through lack of coordination; no staff member has the task of attending to students' enquiries in human genetics—and, as a result, the enquiries tend not to be made!

There is no reason to suppose that the experience of this medical school is not typical of other medical schools in the country; in fact, from preliminary enquiries, it is probable that this is a somewhat richer experience than that gained by medical students in most other schools in the Republic. In short, it is extremely doubtful whether any South African medical school gives an adequate genetical training to its students.

It is high time that this state of affairs was remedied. No South African medical school can be said to be fulfilling its duty adequately unless it trains doctors who have a clear understanding of the role of heredity in disease.

The contrast with what is being done abroad is striking and not a little humiliating. All over Europe and America, and in some parts of Asia, human and medical genetics departments have recently sprung into being.[9] For instance, courses in general human genetics have been included in the regular medical curriculum at Uppsala in Sweden; at Oslo in Norway; in France; in Italy at Rome, Florence and Milan, while at Turin a course of 40 lectures is given in the 3rd year of study. In Denmark the course comprises 25 lectures during the 1st year in medical school, and 12 teaching sessions, mainly clinical, near the end of the degree course. In Japan most medical schools have regular courses in human genetics. In Brazil 45 lectures are given at Sao Paulo. At McGill University, in Canada, the course comprises 12 lectures in the 1st medical year and 3 hours of clinical genetics during the paediatrics 'block' in the 4th year.

In Great Britain the General Medical Council has recommended for about a dozen years that medical students should be taught the elements of genetics, but it has not suggested who ought to do the teaching.[10] It is seldom a separate course, but has been brought into the teaching of embryology, histology, bacteriology, or the discussion of blood groups.

In the United States several medical schools (such as Bowman Gray in North Carolina and the University of Michigan at Ann Arbor) have full-fledged departments of human or medical genetics as integral parts of the medical faculty; in many others a geneticist is attached to some or other department. This is itself a marked improvement, for it is not yet 25 years since formal teaching of genetics was first injected into the body of medical education in America. Ten years ago only 6 American medical schools —out of 80 to 90—offered a proper course in genetics, consisting of from 6 to 24 hours. Clearly genetics is the 'baby brother' of the medical subjects; but it *can* be said that in many parts of the world, after passing through a difficult infancy and an indifferent puberty, medical genetics is now a lusty and blooming adolescent, calling ever more attention to itself.

By these international standards South Africa has much leeway to make up.

Recommendations

I should like to make a series of recommendations. Some of these are adopted from the WHO committee, some from the American teaching institute for 1954; and some are simply thrown in by myself.

1. Every South African medical school should be urged to institute comprehensive, systematic tuition in human and medical genetics. At least some part of this tuition should be in the preclinical years and some part in the clinical years. For the content of the course the detailed recommendations of the WHO committee could well be adopted as a standard, and the course should include practical as well as theoretical work.

2. Initially the tuition would devolve upon the one or two genetics enthusiasts who are probably to be found on the staff of every medical school. To give such people a more thorough preparation it is suggested that, where one or more reasonably-trained geneticists exist at a centre, a summer school or workshop for teacher-training should be held, with participants from other schools not so favourably placed. This would be a stopgap arrangement until the country had acquired sufficient qualified human geneticists.

3. From a long-range point of view it is recommended that the minimal qualifications for a medical geneticist should be both a medical degree and a scientific genetical qualification. An example of the latter is the medical B.Sc. and honours courses, with a heavy genetical bias, at present being offered in the Department of Anatomy at the Witwatersrand University. Such courses may, in fact, provide some of the trained personnel who will ultimately be needed in the country. For some little time, however, it is likely that a spell of intensive training abroad, perhaps in the famous human genetical institutes and departments of Great Britain, the United States, and Scandinavia, will be necessary to provide the personnel.

4. In the early stages a starting point may be provided by a lectureship and a counselling clinic or service, such as that which Prof. L. A. Hurst has established in Johannesburg. A logical subsequent development might be a full-fledged *heredity clinic* under the aegis of the *clinical* departments, and a *human genetics unit* under the *preclinical* departments. Preferable, however, would be an all-embracing, interdepartmental human genetics unit, covering both the *basic* and the *applied* aspects of human genetics. The *basic* aspects include population genetics, biochemical genetics, twin studies, serological genetics, and chromosomal studies, while the *applied* aspects include advisory or counselling services for patients, and consultative services for the medical profession. Ultimately such a unit must evolve into a self-contained teaching institute or university department.

5. Refresher courses— or should one rather call them *fresher* courses?—should be provided for general practitioners and specialists at centres competent to give them. These courses would stress heredity in relation to disease. After a week's tuition practising members of the profession would have become more genetics-conscious in their practices, and would have been trained to recognize where a genetical situation exists and where the help of a medical geneticist needs to be invoked. Where at present *general* refresher courses are given to practitioners, it is urged that the organizers should try to include some small genetical emphasis in their programmes.

6. My final recommendation is that the Medical Association should institute a new *scientific section* on *medical genetics* at its biennial congresses. Besides the 7 papers in this symposium, no fewer than 10 other papers presented at this congress are of genetical interest or have a major genetical aspect. This number will grow, and ultimately one looks forward to a South African Society of Human Genetics to foster interest and research in the subject.

One Aspect of the Importance of Genetics in Medicine

I should like to dwell briefly on one aspect of genetics which deserves to receive your attention and that of medical educators. I refer to the changing pattern of disease that has been brought about especially by sulphonamides and the antibiotics. These drugs have ushered in a new era in medical biology in which the old balance between man and his germs and parasites is slowly but surely being upset. Already in advanced societies the bacterium has lost its place as Man's No. 1 killer. As it topples from its inglorious pedestal, a new series of killing diseases takes its place. These new major killers include *cardiac disease, other vascular disorders,* and *cancer*. Along with *accidents* they have moved to the top of our list of killers, while infections have moved low down—pneumonia and influenza are 6th, tuberculosis 7th. The diarrhoeal diseases, which were the 3rd biggest killers at the beginning of the century, do not even appear in the 10 top killers of today.

What has all this to do with genetics?

The diseases that have been unseated from the dubious distinction of killing most people in 1900, are the *germ-caused* diseases; while the diseases that have moved up to take their place, though of complex aetiology, all feature a major genetic component. Though hereditary susceptibility is a factor in death from infection, nevertheless the evidence suggests that the new top killers depend to a much greater extent on the genetic component. The bad gene is competing to take the place of the bad germ. In this way, a fantastic revolution in Man's welfare and survival is occurring before our eyes. It is a revolution that has still not started in many under-developed parts of the world, such as most of sub-Saharan Africa, with its appalling ratio of one medical school to every 15,000,000 people! But where hospital facilities, medical services, personal welfare and hygiene are adequate, suddenly Man has altered a pattern of adjustment to Nature, a pattern to which he has been subject for his first two million years on earth.

In the foreseeable future gene-controlled conditions will increasingly determine the cause of death. Not only are we now in possession of the knowledge to prevent most of the preventable diseases, but by successfully treating infection medical science is keeping people alive to older ages. In turn, this is giving an opportunity for genes, which exert their effects later in life, to declare their presence—such as the genes associated with some forms of cancer. This is further increasing the toll of death to be laid at the door of the genes.

What is more, the very drugs we are using to bring about this revolution are uncovering the presence of yet other bad genes, which might otherwise have escaped detection altogether. For example, in some populations a high proportion of individuals are deficient in an enzyme called glucose-6-phosphate dehydrogenase. Under natural conditions, it seems that the possession of this gene makes little difference, apart from the development of favism (an abnormal response to fava beans). But sulphonamides and primaquine induce a haemolytic anaemia in males who have this gene (or females who have it in the homozygous state, or double dose). In other words, our drugs are uncovering the existence of harmful genes whose effects become apparent only under the very special environment created by exposure to the drug.

Thus, the new medicine, while giving man a longer lease of life, is leaving him more blatantly exposed to his own genes. This process will inevitably continue and intensify. The medical practice of the new era to which we have become heir finds the gene an increasingly important factor. And we are forced to the realization that it is much harder to eliminate a bad gene than a bad germ.

Even if there were no other reasons for studying genetics, this changing face of medicine is sufficient reason to justify our acknowledging the importance of the gene, and informing ourselves and our new prospective doctors about it.

SUMMARY

There is an urgent need for the introduction of formal instruction in human genetics into the medical curriculum, both undergraduate and postgraduate. The history and present position of genetics in medical education both in South Africa and elsewhere are considered. Recommendations are submitted for the development of the subject in the South African medical schools. An aspect of the changing importance of heredity in modern medicine is considered.

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