

## Training of medical geneticists

One of the exciting accompaniments of advance in a particular field is the way in which people from diverse backgrounds combine in a fruitful partnership, conferring hybrid vigour, as it were. The convergence of different disciplines into the field of human genetics has resulted in an explosion of knowledge in the past 2 decades to which this and similar journals are today eloquent evidence.

Such chance convergence has a weakness, however, that cannot be overlooked; many of those working in the field today have not been trained for it. Their knowledge of basic genetics has been obtained later in their career and often is highly selective. That some have, nevertheless, remedied the initial deficiency is shown by their fundamental new contributions which have had repercussions through the whole science of genetics. However, now that advances are likely to be less spectacular, with consolidation and application the keynotes, it has become necessary to consider the training of medical (clinical) geneticists. This has been for some time a concern of the Clinical Genetics Society. The Society's original outline recommendations were incorporated into the second edition of the report of the Joint Committee on Higher Medical Training, and the Clinical Genetics Society subsequently set up a working party<sup>1</sup> to provide more detailed recommendations.

Their first step was to consider and define the functions of the medical geneticist, particularly in relation to other disciplines. The medical geneticist is envisaged as having 4 main functions: to contribute to diagnosis (including prenatal); to counsel patients and their relatives; to maintain genetic registers; and to act as consultant to cytogenetic, biochemical, and other relevant laboratories. In short, he or she would be responsible for organising a comprehensive genetic advisory service. Additional functions would be involvement in teaching and research, which would be particularly important since he would be working mainly in major centres, usually teaching hospitals. Thus a joint NHS/university appointment at consultant level to a Genetic Advisory Centre is preferable.

In a programme designed to prepare for these responsibilities, the working party was emphatic that flexibility was essential and that most of their recommendations were to be regarded as a guide

rather than mandatory. However, because medical genetics crosses so many boundaries, a thorough grounding and wide experience in a clinical subject, such as paediatrics or general medicine, were essential.

The prospective candidate should obtain a membership, normally the MRCP (UK), before starting higher specialist training as a senior registrar. An early component would be a year of basic genetics, with emphasis on human examples. During the remainder of his training he must obtain experience in 3 main fields, genetic counselling, cytogenetics, and biochemical genetics. Opportunity to see the different syndromes, which is particularly important in the dysmorphism group, should be provided by participation in a genetics clinic. By such involvement, particularly in the preparatory work for these consultations, the trainee will learn how to construct a pedigree, how to interpret laboratory data, how to consult old records and, supremely, the art of counselling. Practical experience in karyotyping leucocytes and cells from amniotic fluids, as well as in enzyme and other biochemical assays, and interpretation of the results, would be necessary. Other aspects, such as immunogenetics, population and quantitative genetics, including statistics, should also be covered. Wherever possible he should maintain his interest in the clinical field in which he already has some expertise. To fit all this experience into 3 years will not be easy. To develop a special interest within the field will be more difficult still, and yet should be encouraged. A year in another centre, particularly one abroad, could provide such an opportunity, and would enhance the value of any training programme. Provision has also been made for training those who would work primarily in another field, but who wished to have some experience in genetic aspects of their subject. At present accreditation is the responsibility of the Paediatric Specialist Advisory Committee of the Joint Committee on Higher Medical Training.

The Clinical Genetics Society has suggested that there should eventually be 42 consultant posts, 2 being based in each provincial medical school with 4 centres in London. The present situation, in which a proportion of those working in the specialty do not devote the whole of their time to it, is not expected to change rapidly. As several vacancies for consultant clinical geneticists have not been filled recently, it may be that joint appointments with some sessions in, say, paediatrics will have to be considered, but it would be preferable for most new posts to be wholly in the

<sup>1</sup>Copies of Report of Working Party may be obtained from Dr A. W. Johnston, Ward 8, Woodend General Hospital, Aberdeen AB9 2YS.

specialty. This lack of suitable candidates also emphasises the necessity to implement this training programme, despite the financial situation, by creating a total of 6 senior registrar posts, and a willingness to fund the consultant posts with the appropriate supporting staff and facilities in the genetic advisory centres. In view of the increasing demand for genetic advice, it is essential that Area Health Authorities and Boards should give high priority to the establishment and maintenance of effective genetic advisory services in each main centre.

Such support will be critical to the future of the specialty since, unless there is a reasonable prospect of a career ahead, no-one will embark on highly specialised training. This is a further argument for funding this programme. Judging by other fields, it is apparent that the future development of a specialty, resulting in better service to the patient, has been promoted by the recognition of a training scheme and career structure.

To meet a comparable situation in Canada, workers in the field of medical genetics have recently set up a Canadian College of Medical Geneticists. Its role is to establish and maintain professional standards of health care delivery in the field of medical genetics. The details, not only of their training programme but also of the other functions of medical geneticists, show considerable similarity to the report produced by the Clinical Genetics Society. The main differences relate to aspects relevant to the Canadian situation, including

scientists with a PhD being trained in genetic counselling, and with regard to the funding. They recommend that for the medical graduate there would be 2 years of approved residency training in internal medicine, paediatrics, and family practice, either singly or in combination. Then would follow 2 years in an approved medical genetics programme which must include a basic genetics course. This shorter period of postgraduate training is, of course, in line with that in other specialties in North America.

The pressures which produced these 2 programmes require full recognition from those responsible for both training and service commitments to be followed by implementation. Otherwise the specialty in this country may fail to develop its full and exciting potential in the coming decade, to the detriment of patients and their relatives, whether considered in terms of economics or in that most fundamental of human hopes, a healthy family.

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**Note added in proof**

Six senior registrar posts have been or are about to be created. The Medical Genetics Sub-Committee of the Paediatric Specialist Advisory Committee has also been formally established.