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## *Editorial*

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### Genetics services in the community

The work of clinical geneticists, and much of the work of related laboratory genetics services, is aimed principally at the needs of individual people and families with a risk of genetic disorders. The family may be large and often extended, but the aim of the service, whether genetic counselling, prenatal diagnosis, or carrier detection, has primarily been to resolve the problem of a specific person, couple, or family, with population prevention of the disorder being seen as a separate and often subsidiary issue.

Clinical genetics has thus until now been firmly rooted in the tradition of other medical specialities, with emphasis in training and practice placed on skills of diagnosis, communication, and risk estimation, as well as on the integration of the rapidly evolving range of specific genetic tests. Yet most clinical geneticists have also been aware of the need to consider the population preventive aspects of their field, and have often been acutely aware of the contrast between what could in theory be done to prevent and avoid genetic disorders in families, and the small proportion who are actually aware of the services that are available. It could be argued that the effective and equitable delivery of services that are already feasible is at least as great a challenge as the development of new techniques.

These important and potentially controversial issues are addressed in two items in this issue of *Journal of Medical Genetics*. The article by Dr Bernadette Modell, 'Cystic fibrosis screening and community genetics'<sup>1</sup> examines the existing organisation of population genetics services in the light of the likely advent of mass cystic fibrosis carrier screening, while the report of the Royal College of Physicians of London *Prenatal diagnosis and genetic screening*,<sup>2</sup> reviewed on page 535 by Professor Rodney Harris, is similarly largely devoted to the population aspects of these services.

Several points emerge immediately from these

reports, which will involve equally clinical geneticists and those responsible for laboratory services. First, these services are already widespread and in some communities well developed; the past two decades have seen rapid implementation of prenatal testing of older mothers for Down's syndrome, of neural tube defect screening based on serum AFP and ultrasound, as well as neonatal screening for treatable disorders such as phenylketonuria and congenital hypothyroidism. Carrier screening for recessive disorders has so far been successful only in specific high risk populations for disorders such as the haemoglobinopathies and Tay-Sachs disease, but these now provide a valuable model for the much larger challenge (in northern Europe and America) of cystic fibrosis. Cystic fibrosis screening is also likely to be the first population based application of DNA techniques to inherited disorders.

Despite the real individual success of most of these programmes, when viewed overall there is a striking lack of coordination and evaluation. A variety of different laboratory services may be responsible for these programmes and while the record of quality control has been good, there has frequently been a total lack of long term evaluation of effectiveness and acceptability at a consumer level. Even when such evaluation has been present in the research phase, long term funding has rarely been available to allow its continuation as an integral part of the service. There is thus a real danger of proliferation of uncoordinated services with a risk of confusion for those receiving them and with no clear overall policies or priorities.

If the need for some coordination is accepted, whose duty should this be? As stated at the outset, most clinical geneticists have skills more related to the individual person or family than those required for the implementation or evaluation of population based programmes. Laboratory scientists also are not ideally

trained for the role, though clearly they are best suited for the maintenance of quality in their own services. Obstetricians have so far rarely proved able or willing to assume coordination even of the prenatal genetics services, though they are essential to the delivery of these.

This leaves two groups of medical staff, primary care doctors (in Britain general practitioners) and community (public health) physicians, neither of whom currently has adequate training in genetics. Yet the effective implementation of all these programmes will only be possible with the support of motivated and informed primary care medical and allied health staff. Thus knowledge of genetics and its applications must be considered as essential in both the general medical curriculum and the subsequent training of those in the primary care field. The Journal will be referring to this topic in a future issue in the light of a new report on the teaching of medical genetics in Britain.<sup>3</sup>

The involvement of community physicians and departments of public health medicine can be seen as a logical extension of their traditional role in epidemiology and communicable disease, areas closely allied to the implementation of preventive programmes for genetic disorders. However, at present, in Britain at least, their training contains little or no genetics, while many are preoccupied, even overwhelmed, with administrative duties. A further factor, never far in the background, is the history of abuse of genetics as applied to public health, something that has already

occurred in the past and could well happen again if population goals are given priority over individual decisions.

Perhaps then there is a need for a new speciality, that of 'community geneticists', who could have training both in genetics and community medicine, and whose main remit would be the coordination, audit, and delivery of the overall series of genetics services in the population, though not the management of the individual laboratory components. Such a person would need to be firmly based in the community health structures, working closely with primary care physicians, community nursing staff, and those involved in health education. A close link with the specialist medical genetics centres would be essential and perhaps participation in genetic counselling activities would be a valuable and necessary reminder that, however important the evolving service of community genetics may be, populations are made up of individual persons whose needs and decisions will inevitably and rightly differ, and whose acceptance and support is essential if genetics services are truly to benefit the community.

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- 1 Modell B. Cystic fibrosis screening and community genetics. *J Med Genet* 1990;27:475-9.
- 2 *Prenatal diagnosis and genetic screening*. London: Royal College of Physicians, 1989.
- 3 *Teaching genetics to medical students*. London: Royal College of Physicians, 1990.

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