In order to understand genetic services, it is necessary first to discuss the role of science and the role of public health in genetic services. In discussing the role that science can play in the planning and conduct of genetic services, it is necessary to begin by asking the questions “what is science and what are the limits of science?” There is indeed widespread misunderstanding about the meaning of science, even among the health professionals. There are two assumptions that are made about modern science which are incorrect and which greatly mislead people. The first assumption is that the role of science is to collect facts and to arrange them in order to be able to explain how the world works. Neutral, objective scientists observe the world and may experiment with parts of it, but they always remain detached. Provided that they follow the rules of scientific method, their own beliefs, attitudes and desires are irrelevant to the practice of science.¹ This view of science has now been widely discredited. Two major objections to the “neutral facts” view of science are that:

(a) scientists are not detached from the world, but are part of it, so that the very process of science carries the values, prejudices, and beliefs of scientists and the communities in which they live;

(b) facts are a product, at least in part, of the process of searching for them. Information becomes fact only when a scientist working within a particular theoretical framework and using a specific methodology, creates concepts and measurements.¹ In the disciplines of physics, Albert Einstein and Niels Bohr clearly demonstrated over 75 years ago that in physics you cannot keep the scientist out of the equation. Since that time this has been clearly understood by theoretical physicists. In other words, scientists do not escape social influence in their perceptions. In short, science is a social process, an institution, and it must be analysed and understood as such.

It is high time that medical scientists put aside the notion that they are a group of objective scientists who are collecting and organizing facts which are then used in planning and delivering medical services. In the medical world there are frequent organized meetings of “experts”. Calling someone an expert is a form of sympathetic magic, by giving something a name, it becomes what the name symbolises.

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Thus, by organizing a meeting of experts, we merely ensure that the beliefs and assumptions of the group of experts are embodied in the recommendations.

The second assumption which is widespread among medical scientists today is that issues in health can best be addressed by objective scientists who know the facts. Health is all about the quality of life and science does not address issues of quality. In other words, science cannot decide on good versus evil, nor can science tell individuals or groups how they wish to lead their lives. Science properly conducted can provide information which the people can then use to make their own judgements about their own lives. It is important that we all understand the above limits of science when we conduct our work in genetic services. In other words, we certainly must use science in genetic services, but we must use it properly.

There are two kinds of science that we will use in genetic services — clinical science and epidemiology. What is epidemiology? This is perhaps best explained through an analogy. Imagine a football game. The clinicians are the players on the field. As such they are personally involved in the real world of action. They are in direct contact with the ball and the other players. The clinicians know in great detail from personal experience just exactly what is going on in the game and they must repeatedly make quick decisions that may profoundly influence the outcome. They talk with each other as the game progresses, making suggestions, sharing experience and giving encouragement.

Then suddenly the game is half over and what happens? The players leave the field and go to a meeting. At this meeting someone called a coach begins to advise these players on how they should play during the second half of the game. On what basis? The coach has not been on the field in actual contact with the ball or other players. The coach has been sitting on the sideline, carefully observing the overall pattern of play from a distance. The coach is the team’s epidemiologist. While coaches must understand all the rules of the game, they must also have further skills and knowledge — to know how to make correctly observations on groups of players, how to interpret these observations, and how to communicate them to the players in the most effective way.

During the meeting an essential, dynamic interplay takes place between the players and the coach. The players report their experience on the field and their impression of what is going on. The coach respects this important information and takes it seriously, evaluating the players’ reports, making an overall assessment of the situation, and then advising on how to play. The players respect this advice because of the coach’s skills and unique perspective. At the same time there is often a tension between the players and the coach. While it is hoped that each side respects the other, each may also feel envious. The coach may envy the players the excitement of participation in the action, the adrenalin surge of play, the opportunity actually to touch the ball and make those last small but vital judgements on which way to turn. The players may envy the coach the luxury of sitting quietly on the sideline, not running out of breath (staying up all night), not taking the risk of being injured (sued), and yet having so much authority and so much to say about the course the game will take. This tension between the players and coach may at least to some extent simply be a natural consequence of the nature of the game and may even be constructive as long as mutual respect remains.
In football today, no team would want to be without a coach. Sadly we are not there yet with health care. Some players talk only with the other players on their own team and go to conventions where only players attend and talk (or even brag) about their individual play. Some coaches feel superior to their players and will not listen to them, but go to conventions attended only by coaches and talk about the theory of the game or discuss endlessly the details of the rules of the game. Slowly, however, it is becoming clear in health care that both the clinical viewpoint and the epidemiological viewpoint are equally correct and valid. Both viewpoints represent pieces of the truth that complement each other in a way that brings forth the whole truth. We need players and coaches who respect and communicate continuously with each other.

Having illustrated the epidemiological approach, now what must be said is that the public health approach to genetic services or for that matter to any other health services, is to combine the scientific assessment of the problem using epidemiology with the planning, evaluation and monitoring of the health services directed at that problem. What happens then if we apply this public health approach to genetic services?

Genetics began as a laboratory science and, when the technology became applicable to humans, evolved to a combination laboratory/clinical medical specialty. The clinical geneticist, like all clinicians, has focused on individual cases. Still today most clinical geneticists practise differential diagnosis and treatment, although the nature of the genetic diseases demands greater emphasis on family history during diagnosis and greater emphasis on counselling as part of the treatment.

Any clinical practice, including clinical genetics, is deeply influenced by the system of health care in which the practice occurs. It doesn’t take long for a new physician in practice to discover that he is not an island unto himself but part of a network of physicians who in turn are part of a system of services.

More recent advances in genetics are now forcing a broadening of the purely clinical approach. For example, the possibility of preventing genetic disease, through, for example, prenatal screening and neonatal screening has brought the public health approach into genetic services. Screening for genetic diseases began with neonatal screening which was quite straightforward with not too many ethical complications. When prenatal screening came into the picture, however, there were immediately all kinds of problems. For example, there is no use in doing prenatal diagnosis on a pregnant woman if she would never consider therapeutic abortion. These prenatal screening programmes have proven to have significant false positive and false negative rates and these can be quite devastating mistakes for a family to experience. Nevertheless, the prenatal screening programmes have been effective and it has, for example, been possible completely to eliminate new cases of thalassaemia major from certain regions in Italy where it was previously of considerable prevalence. However, elimination of thalassaemia in Italy was only possible through extensive community education programmes so that all of the necessary screening could be done. It is also interesting to note that although there was a high prevalence of Catholic families in these regions, if the need for screening for thalassaemia was explained to the parents, there was a high rate of acceptance of the procedure and also a high rate
of asking for therapeutic abortion if the prenatal diagnosis was positive. This Italian experience is certainly relevant to the organization of genetic services in Northern Ireland.

A new procedure offers the possibility for eliminating genetic diseases without abortion. This is pre-implantation genetic diagnosis combined with in vitro fertilization. There are studies going on in the UK and in the USA which illustrate this new possibility. In both cases, couples who are known to be carriers of a severe sex-linked genetic disease are included. In other words, the child will only have the genetic disease if it is a male. So using IVF the egg is removed from the woman and fertilized in vitro with the partner’s sperm. When the fertilized egg has reached the four-cell stage, one of the cells is removed and examined to determine the gender of the fertilized egg. If it turns out to be male, the egg is destroyed and if it turns out to be a female, the fertilized egg is re-implanted into the woman. Since at the four-cell stage there is not yet differentiation into fetus or placenta, it is argued that this destruction of a four-celled egg is not in fact abortion.

But we are immediately faced by some serious problems. First of all, this procedure demands IVF. Another epidemiologist and I carefully studied the world’s literature on IVF and found that the efficacy of this procedure was very low — between 5 - 10% of the times one can expect to have a live baby after an IVF cycle. Furthermore, there are serious risks which have been markedly underestimated by the clinicians involved. IVF is a sad example of modern salesmanship in health services. The public and the politicians have been fed a great deal of bias, if not false information, about success rates and safety with regard to IVF. Fortunately the Director of one IVF clinic in Northern Ireland has been actively involved in the monitoring of IVF services in the UK.

A second problem with regard to pre-implantation genetic diagnosis is an ethical one — where to draw the line. If we can use this procedure to eliminate serious sex-linked genetic diseases, we can also use this procedure to eliminate female children. As you probably know, there have already been cases in India of couples getting prenatal diagnosis in order to eliminate any females so that they can have a son. But where do we draw the line? Do we eliminate offspring with crossed eyes? Even if government services have ethical controls, commercial genetic services have few or none.

While there are a number of ethical dilemmas involved with therapeutic medicine which are quite commonly discussed, there are some ethical dilemmas associated with preventive medicine and screening which are quite different and have received very little attention.

1. Screening involves large numbers of people, so unethical decisions, whether at a technical, clinical, social or legal level, can have widespread effects.

2. Prevention is usually addressed to healthy people, and unethical decisions can interfere with an entirely or apparently normal life.

3. Preventive medicine and screening procedures are aimed to influence healthy people’s behaviour — to motivate them to seek genetic screening. It is likely to reach the better educated sections of society preferentially so the issue of equity is present. This is particularly important for genetic diseases like
thallasaemia and sickle cell disease which occur mostly in lower socio-economic groups.

4. Since it involves testing large numbers of unaffected people in the search for an affected minority, screening should be practised to the highest possible standard with suitable training, a quality control system, and a professional code of practice.

5. In case of the slightest doubt, because of its serious and life-long implications, a genetic diagnosis should, as far as possible, be confirmed by an independent approach.

6. Results should be monitored regionally and nationally, with particular emphasis on false-positives and false-negatives.

Thus we see the need for the public health approach in screening for genetic diseases. One of the most important applications of the public health approach to genetic diseases is for the rational planning of these services. Sadly it must be pointed out that there is very little rational planning of any kind of health service today. Very often it is only by accident that a particular service becomes available. If an interested clinician at a hospital wants to start a genetic clinic or an IVF clinic, then he or she can set about doing it, but in no systemic or rational way.

In the rational planning of genetic services, the first step is to determine the need for such service. This involves using epidemiology to determine the prevalence of genetic diseases. For example the World Health Organization publication on genetic services\(^5\) gives epidemiological estimates of the number of people with certain genetic diseases in Europe. Every year in Europe 4,500 children are born with cystic fibrosis. There are now approximately 23,000 living cystic fibrosis individuals and it is estimated that if these same numbers continue to be born and receive the same type of care now available, we will eventually have 112,000 individuals with cystic fibrosis in Europe. This illustrates that we can in fact estimate the need for genetic services.

The second step in rational planning is to determine the ability of the present services to meet the need that has been determined. In other words, can the services meet the need, and if so, how well and at what cost and what danger? There is usually very little done in any country to answer these questions. This involves the assessment of the technology used in genetic services. In every country in Europe there is a careful system to evaluate any new drug before it is allowed on the market. On the other hand, a new procedure or a new machine can be used tomorrow in any country in Europe without any necessity for its careful assessment. As a result, many procedures and technologies come into widespread use before any adequate assessment. The World Health Organization did a study of routine obstetrical procedures and discovered that only about 10% of them have been adequately scientifically evaluated as to whether or not they are of any value.\(^6\) This is an example of the urgent need to bring good solid scientific methodology into the assessment of what were are doing, including genetic services.

The same holds with regard to assessment of the safety of new procedures and technologies. Many IVF clinics advertise or state that IVF is safe, when in fact we now know that there are all kinds of risks both to the woman and to the baby.\(^4\)
For example, the perinatal mortality rate for babies from this procedure is four times higher than usual. We need to look more carefully at the risks of procedures and technologies and be more honest with our results. We also need to assess the cost of procedures. This is complicated and must involve health economists in order to include indirect costs and the costs of failures.

Once we have information on the ability of the service to meet the estimated need, we come to the moment of decision on whether or not we want such a service. What should be the priority for funding such a service in a health care system and who should decide? Until recently most of these decisions were made by physicians, but this is inappropriate since these are not medical decisions. No country in the world today can afford to do everything that is possible to do with medical procedures and technologies — transplant all of the hearts, dialyze all the kidneys, give IVF to all the infertile couples. It is only the public and its representatives who are in a position to make the decision with regard to who shall live. Incidentally, the people who are suffering from a condition are not the ones who should decide whether or not there should be services for this condition. IVF clinicians all quote the demand for their services from infertile people. And yet when a poll of the general public asked for priorities on how to spend money for various medical procedures, treatment of cancer was at the top and IVF at the bottom.7

Once a decision has been made to deliver a particular service, the next question is how to deliver this service. Here it is necessary to involve the consumers of those services so that they will be delivered in a way that is acceptable to the public. Thus, to open a clinic for thalassaemia you need to approach the families with thalassaemia in your community, and involve them actively in planning and assessing and monitoring this service. Most countries have active lay organizations for the various genetic diseases and these organizations can play an important role in determining the nature of services.

Once the services are in place, it is then necessary to have quality assurance programmes. Whether medical people like it or not, this is the wave of the future. Quality assurance protects the public from bad practices and also helps to be sure that the money is well spent. One of the goals of such quality assurance is to pay only for appropriate care. In the case of genetic diseases this would ideally get to the point where the clinicians would be paid according to their role in reducing the number of cases of genetic diseases. It could be that some day in the future we will get to the ideal that was proposed by Hippocrates several thousand years ago — that the physician should be reimbursed for all of the well people rather than for the sick people in his practice.

It is clear from what I said that genetic services need to involve a close collaboration between the clinicians, the epidemiologists, the other public health personnel and the lay public. This is the direction that all health care is going today. Physicians cannot make ethical decisions nor can they make priority decisions with regard to health services. But we can use the best science to assess our procedures and practices and then give this information totally and honestly to the public and their representatives, so that they can decide what they would like us to do.

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