

GIG Response to the Clinical Genetics Society Report: "The Genetic Testing of Children".

The Genetic Interest Group is the national umbrella organisation for the charities, voluntary organizations, and support groups which serve those affected by specific genetic disorders and their families. GIG was officially launched in 1989 by a group of voluntary organisations concerned with genetic disorders who saw the need to coordinate action on the issues the groups have in common. Primarily, these are to improve support and services for people affected by genetic disorders and to advance the knowledge and understanding of human genetics throughout the population.

GIG now has a membership of over 100 ranging from large, well established organisations to small support groups for very rare conditions. Research to find the gene(s) responsible, and treatments for the resulting condition, is at a different stage in each of the many disorders being studied. There is, therefore, a wealth of knowledge based on experience which can be passed on from those who have already met the dilemmas of genetic testing to those who are still awaiting the possibility. Guidelines must take account of this knowledge. They must also look to the future. The field of genetics is developing fast. More and more conditions are becoming testable, and, hopefully, treatable. Different disorders will have some different implications for testing, depending on the usefulness of test results, the severity of the symptoms, and the availability of treatment. However, GIG believes that it is both possible to draw up standard, basic, guidelines for testing, and also necessary to do so if best practice is to survive the extra burdens that will result from the expansion of the field.

The genetic testing of children is an issue that has been much debated by GIG and its member organisations. Accordingly, GIG welcomes the Clinical Genetics Society's report as a first step in raising the issue for consideration before a larger audience. This is essential, as many families affected by a genetic disorder are never referred to a geneticist, and this trend will continue. For example, children with Friedreich's ataxia are almost always seen by a paediatric neurologist. Information about the at-risk status of brothers and sisters is usually given by the same consultant. The issue of presymptomatic or carrier testing is rarely discussed. GIG therefore particularly welcomes the work done in surveying paediatricians and others, even if the reply rate was rather disappointing. Also welcome is the research on the Children's Act in preparation for the report which puts the rights and well-being of the child to the forefront. An issue that needs further consideration in this context is how to draw a dividing line between childhood and adulthood, and

the consequences this might have for issues related to the genetic testing of children. The age at which a young person becomes sexually active might be a better indicator than any fixed age. It is certainly the age at which genetic information becomes important in many circumstances.

However, GIG considers the report to be deficient and flawed in a number of areas. For example, no specific questions on childhood-onset were even included in the survey sent out. Because of this, GIG set up its own working group to highlight areas of particular concern. GIG has a different perspective from those who wrote the original report - a perspective informed by the experience of families directly affected by genetic disorders. GIG wishes to be assured that this perspective will be taken account of by those involved in clinical practice and legislators, if the issues under consideration become a matter for legislation.

1. ISSUES OF CONCERN

1.1 In attempting to reach a 'consensus', the report fails to reach firm conclusions. Whereas GIG totally agrees that every situation should be treated individually, we still believe that there are basic principles which can and should be adhered to. These issues are taken up in section 2 below. Being aware of the different procedures and protocols followed by different genetics centres and other units involved in genetic testing, GIG is also concerned that if there are not generally applicable rules and procedures, people may have to shop around, their access to services dependent on the views of individual clinicians. People who can afford it may go private if they cannot get satisfaction. This is not only inequitable, but also bad for those who do go private, as appropriate genetic counselling is often not provided in the private sector.

1.2 The consultation with GIG members was poor. A number of the larger organisations as well as the smaller ones found it difficult to respond to the questionnaire as it stood. This possibly accounts for the report's failure to quantify the response from GIG members. It is also felt that the responses quoted were not all well chosen and showed an outdated and patronising attitude towards voluntary organisations. Comments from individuals are valid for that individual - organisations, however, are able to report from a range of experiences. We feel that GIG and its members were badly represented.

1.3 In its efforts to take on board the Children's Act, the report fails to give proper consideration to the concerns of parents who are in fact responsible for the wellbeing of their children. The overall tone is patronising to parents. Because a few may not have the best interests of their children at heart, or have a different perspective as to what those best interests may be, this is no reason to frame recommendations as if all children require protection from parents.

1.4 The report is overly preoccupied with psychological considerations, and the harm that knowledge of genetic disorders can cause within families. With little evidence, this seems to reflect more the fears of doctors that they will be held responsible for negative reactions, rather than the needs of families. For example, ten years after testing for Huntington's disease was introduced, there has so far been only one known suicide. Even then, the protocol for this particular case was not adhered to¹. Whilst we totally uphold the principle that families need counselling and support, we also believe they should be given credit for being responsible and having coping capacities. Although the vast majority would prefer there NOT to be a genetic disorder in their family, knowledge comes to be accepted as a fact of life in the same way that

other issues are recognised to be individual and integral to any family. It is also our experience that children can cope with information about themselves from an early age and that it is much more often the adult who has a problem in giving information. We feel ourselves to be in a strange position in this argument - it is often the role of the voluntary sector to educate the medical profession in the need to understand psychological factors - perhaps the pendulum has swung too far the other way in genetics.

1.5 The report tends to notice only the down-side of testing when there is no medical treatment available. Apart from adult-onset conditions (see section 2 below), there are many non-medical reasons for testing. Although there obviously ARE potential disadvantages to having knowledge prior to the event, for some families these would be outweighed by the advantages (see 2.1 and 2.2 below). If families are making a properly informed choice, then again the problem may be that professionals feel over-responsible.

1.6 The main focus of our response is to argue for the right of parents to have their children tested, except in the case of adult onset conditions. However, it should also be re-affirmed that tests should not be carried out without parents' consent.

1.7 Communication between professionals and children is often difficult. To help overcome this problem, we suggest that a booklet is produced and given to all children who are tested for childhood onset conditions or carrier status. To be most appropriate, the information contained should be of a general character, covering the different kinds of conditions in terms of onset and type (primarily dominant, recessive, and X-linked). We also suggest that each child so tested be given a record of which lab did the test and what kind of test was carried out. Beyond supplying the child with basic information, the intention is that the child should have a record of the results in case they lose touch with their family or GP, and that s/he also has a record of what kind of test was carried out in case a better, more reliable, test is developed for his/her particular condition at a later date.

2. SPECIFIC ISSUES

2.1 Presymptomatic diagnosis of childhood-onset conditions.

A family may wish to test a child presymptomatically for a wide range of reasons. One reason might be that presymptomatic medical procedures may be of benefit to the child. The case for testing is clear in this case. However, there are valid reasons to test in the case of disorders for which there is no presymptomatic medical intervention. Reasons include: possible freedom from anxiety; facilitating open relationships; and the parents' need to secure the best environment they can for themselves, the child who will develop the disorder, and other children in the family. 'Best environment' might mean a house with suitable access, located near a school and hospital. It might also mean securing particular kinds of work. It is a major defect of the report that it makes little mention of the non-medical reasons for testing.

More than this, the report is vague and refuses to come to firm conclusions about testing in this case. In some places the report seems to propose that testing for childhood-onset conditions should be possible, whilst in other places it seems to argue the opposite. This indecisiveness is a consequence of the fact that those who carried out the study for the CGS neither asked anyone for their opinions on the subject of testing for childhood-onset conditions (the set of questions sent

around by the CGS do not ask for opinions on this issue), nor considered it as a case in its own right. The result is that some sections of the report can be read as favouring testing for childhood onset conditions: 'The predictive genetic testing of children is clearly appropriate where onset of the condition regularly occurs in childhood or there are useful medical interventions that can be offered' (p.1); 'where an affected individual is likely to manifest the disorder during childhood, these ethical difficulties of presymptomatic genetic testing in childhood do not arise' (p.5). Whilst on the other hand, sections of the report can be read as questioning parents' rights to have their child tested for a childhood-onset condition: It is argued that 'the results of the psychosocial evaluation may be critical in future clinical judgements if the medical benefits remain uncertain or are shown to be minor' (p.2). In reality, the report argues for neither position as the issue was not properly considered.

There are a host of different situations that can be cited as requiring different solutions - a parent's desire to test for a condition when onset is likely to be in the near future may be treated more sympathetically than one when it may not be for a few years. Most certainly counselling needs to take account of these differences and allow parents to consider potentially negative effects such as the child being treated as 'ill' long before there are any problems. However, parents are responsible for the welfare of their children, and at the end of the day most of them are better equipped to decide what is in the best interest of a particular child, and the family as a whole, than are outsiders. Denying them the right to cope in the way that they see as best may have the opposite effect to that intended. Parents generally may be good, bad, or indifferent in the way they relate to their children - genetic testing is simply another factor, albeit an important one, in the way they operate.

PRINCIPLE: Subject to the limitations of existing law, parents have the right to make an informed choice on whether or not to have their child tested for childhood-onset conditions²

2.2 Testing for carrier status

Although there are differences to be considered, the arguments in favour of a right to test for childhood-onset also hold good in the case of testing for carrier status. (We note in passing that presymptomatic testing may well reveal a child to be unaffected but a carrier. This information should be treated according to the principles set out below.)

In many, perhaps most, cases, the issue of carrier status will be best dealt with at puberty or when the child becomes sexually active. The child, or young adult, as s/he will then be, would discuss the issues with parents and professionals, and make a decision based upon this. However, in other cases, early knowledge of carrier status could help a child adapt to the consequences of being a carrier over a period of time, rather than having the information presented at puberty, when s/he is going through a time of emotional adjustment and may not best handle the information.

There are additional reasons as to why it might be appropriate to test early for carrier status. Children are often astute, and may well enquire if there is something wrong with themselves or whether they could have an affected child because of the experience of the extended family. For example, a brother may have Duchenne muscular dystrophy, or an elder sister might have given birth to a baby boy with fragile X. Or two cousins may have cystic fibrosis. The CGS certainly advocates openness in answering questions, but seems to prefer answers with a 'worry about it

later' slant. This is not unreasonable, and it is important that a child realises that s/he is not ill and that the issue WILL be addressed later. However, a straight answer, provided that it is well informed, could well relieve pressure in some families. The argument in the report that a child may be treated differently if known to be of carrier status or given erroneous information is unsubstantiated - the vast majority of people are better able to understand the implications than they are often given credit for. Professionals do have a serious responsibility to make sure that the information is understood, and the voluntary organisations can play a large part here. Indeed, our experience is that clinicians often refer families to support groups as they are often best able to provide information in the most appropriate way.

We believe that the interests of the child must be put to the fore, and we appreciate the ethical point of not imposing information on a child, but we believe the seriousness of this information has been exaggerated because it is still relatively new. The child, as an adult, still has the option of whether they want to use it, or not, when they have children of their own. Problems of insurance, employment etc. are problems of public policy and need to be addressed as such.

Overall, we believe that there are distinct advantages for some families who wish to have their children tested for carrier status. Facts of life are best absorbed slowly and when the moment is right rather than during a crisis over a pregnancy. It is distinctly preferable for the child to be involved in the decision to have the test, which could mean delay.

PRINCIPLE: After suitable counselling, parents have the right to make an informed choice about whether or not to have their children tested for carrier status. Ideally, children should only be tested when of an age to be involved in the decision.

2.3 Adult-onset conditions

We believe that adult onset conditions, for which there are no pre-symptomatic medical treatments, are totally different from childhood ones.

The argument that testing of the child takes away their right to make an informed decision as an adult overrides all other considerations. The low uptake in testing for Huntington's disease shows that many people would prefer not to know that they will be affected at some time in the future. We also agree with the report when it argues that genetic testing as a diagnostic tool for possible childhood onset of a condition which normally affects adults should not be allowed. A positive result does not necessarily confirm the diagnosis - there could be other problems, or the child may be reacting to stress in the family and mimicking symptoms. Time, anyhow, will clarify the situation. We appreciate that some parents are so anxious about their children that they put a great deal of pressure on doctors to have them tested. Parents need support, and again the voluntary organisations can be particularly helpful here, but the rights of the affected individual to make an informed choice at a later date have to be held paramount.

PRINCIPLE: Children should not be tested for adult-onset conditions for which there are no pre-symptomatic medical treatments

2.4 Adoption

We fail to understand why the report leaves any loop-holes for adoption. The same rules on testing should apply to children who are adopted as to those who are not.

Additionally, and not mentioned in the report, the same rules should apply to children who are fostered or in residential care. In this case, it may be the Social Services Department rather than the parent who will be responsible.³

PRINCIPLE: The same rules apply for children who are adopted or in care

2.5 Pre-natal testing

Although outside the remit of this report, pre-natal testing needs to be mentioned.

A particular concern is that for childhood-onset conditions, parents should be under no pressure to have a termination if the results are positive. Parents may wish to have a child regardless of disability, but may want to know in advance whether or not the child is disabled so that they can make the necessary practical and psychological preparations. Or, parents may not be sure before a test what to do, but may decide after a positive test that they wish to have the child.

In general, the same rules should apply in the case of pre-natal testing as apply in the case of the testing of children. This implies the following about testing for adult-onset conditions such as Huntington's disease: that it would be unethical to acquire knowledge that a child will be affected as an adult unless there is early treatment available. Thus parents need to consider whether they would want a test at all, and if they do, their conclusion should be that they intend to have a termination on being given a positive result. This is a difficult concept for some as people to accept, as it unequivocally puts the right of the child to make its own decision when s/he is an adult above any wishes of the parents. In the final analysis, of course, parents may change their minds after testing, but careful counselling beforehand may lead some people to take the option of not having the test if they are ambivalent about having a termination. Although this has been debated at length by geneticists and the voluntary organisations concerned with adult-onset conditions, there may be a need for further discussion in the genetic field as a whole.

PRINCIPLE: The same rules apply for pre-natal testing as for children

3. CONCLUSIONS

We have welcomed the CGS report with its wealth of detail and discussion. In our response we have selected the areas we feel are important to clarify. We feel confident, after consultation with the member groups of GIG, that we reflect the opinion of the majority of those that the issues affect most closely. We look forward to further debate on the subject, but believe that there is a need for firm principles which should be adhered to by everybody working in the field.

NOTES

1. As reported by Professor Mike Conneally at the European Huntington's Disease Association meeting, Dublin, September 1994.

2. The relevant Acts which place constraints on this principle include the Mental Health Act. An informed choice includes access to: non-directive genetic counselling (over as many sessions as is required by the family); written and verbal information; information on professional and voluntary support networks; reassurance of ongoing support after the test results.

3. There is a need for reliable and confidential repositories of genetic information. This is true for all children to ensure that they have the opportunity to gain information about themselves

which may not have been passed on earlier. Particular attention needs to be given to children who are adopted or in care. We have suggested that children are given a booklet containing the results of a tests. It is also important that GPs are given the information: they are more likely than anyone else to provide pro-active help, and the results of tests are most likely to follow the individual around if they are held by GPs.

Members of the GIG working party

Shirley Dalby, Chair

Barbara Carmichael (Fragile X Society)

John Gillott (Policy Officer, GIG)

Julie Lambert (Honorary Secretary, Neurofibromatosis Association)

Christine Lavery (Director, Society for Mucopolysaccharide Diseases)