

Clinical practice in medical genetics

The genetic testing of children

Report of a Working Party of the Clinical Genetics Society (UK).
Chairman: Dr Angus Clarke

Conclusions and recommendations

(1) 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 (for example, diet, medication, surveillance for complications).

(2) In contrast, the working party believes that predictive testing for an adult onset disorder should generally not be undertaken if the child is healthy and there are no medical interventions established as useful that can be offered in the event of a positive test result. We would generally advise against such testing, unless there are clear cut and unusual arguments in favour. This does not entail our recommending that families should avoid discussing the issues with younger children, but rather that formal genetic testing should generally wait until the "children" request such tests for themselves, as autonomous adults. This respect for autonomy and confidentiality would entail the deferral of testing until the person is either adult, or is able to appreciate not only the genetic facts of the matter but also the emotional and social consequences of the various possible test results.

In circumstances when this type of testing is being contemplated, there should be full discussions both within the family and between parents and genetic health professionals (clinical geneticists or non-medical genetic counsellors); the more serious the disorder, the stronger the arguments in favour of testing would need to be.

(3) For some disorders, there is insufficient evidence to know whether a diagnosis in childhood is helpful in the medical management of the possibly (not yet) affected child. Research in these areas will be worthwhile and important. When such research is planned, however, it will be important to incorporate a social and psychological evaluation of the genetic testing, as well as a technical and more strictly medical evaluation, because 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. Furthermore, the psychosocial study of testing for these conditions, where the existence of possible medical benefits justifies the study of the testing, may throw light upon the likely psychosocial effects of testing for other dis-

orders; hence such studies may be of more general applicability.

(4) The situation with regard to testing children for their carrier status for recessive disorders and balanced, familial chromosomal rearrangements is more complex. In general, the working party would make a presumption against testing children to determine their carrier status, where this would be of purely reproductive significance to the child in the future.

(A) Circumstances may arise, however, in which the genetic testing of children could be helpful in the provision of accurate information to other family members. Even in families with apparently balanced chromosomal translocations, however, we think that this occurs only occasionally. It is important that children in such families are not tested "as a routine", but that each situation is considered on its merits so that children are tested only when the results will contribute to the counselling of other family members. Otherwise, if the results would only be of future reproductive concern to the child, then it is wiser to defer the testing until the child is able to understand the issues and requests testing in person.

(B) Where such (carrier) testing is, or has been, taking place, it would be useful to institute prospective and retrospective psychosocial evaluations of the impact of the testing on the children and their families, so that future policy can be guided by evidence rather than conjecture and anecdote.

(C) If the testing is not to be performed in childhood, then a certain obligation rests upon the health care system and the family together to ensure that testing is offered when the child is older. While testing in childhood may allow parents and physicians to feel that they have done their duty, this may still leave both parties with an obligation to ensure that the tested child is offered counselling (and possibly an updated genetic test too) when he or she "comes of age".

(5) There are additional factors to be considered with a healthy but "at risk" child referred for adoption, in so far as the results of the testing might influence decisions made on behalf of the child. However, it should not be assumed that genetic (predictive or carrier) testing will be required before a suitable placement can be achieved. In each case, we would

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advise discussion between the medical adviser to the adoption agency and a clinical geneticist. The important factors other than the possible laboratory test results need to be identified for future attention in advance of any test being performed.

(6) Because some of these recommendations (1–5) are likely to diverge from the practice and beliefs of many within the medical profession, and even within clinical genetics, it is important that further discussion and debate take place. We believe that the medical profession should work towards a consensus on these issues before such tests become more widely available through commercial laboratories, which may pay little respect to the goal of coupling laboratory testing with the provision of counselling and support as a package of genetic services. The ability of the working party to arrive at such a consensus, despite our holding different views initially, suggests that this may be a realistic goal. It will also be important to extend this work to achieve a broader consensus across the professions of nursing, social work, and the law, as well as medicine.

Background

Opportunities for genetic testing

Technical advances in recent years have greatly enhanced our ability to identify persons carrying faulty genes. It has been possible for some years to identify those carrying adult polycystic kidney disease by ultrasound scanning before the onset of symptoms; the healthy carriers of some haemoglobin disorders can be identified by routine, automated haematological analysis. However, it is now possible to identify those apparently healthy persons likely to develop one of a large number of inherited disorders by molecular genetic technologies. It is also possible to identify those who carry faulty genes or chromosomes that will cause them no health problem, but which may result in problems for their future children. Much of this new testing has become possible because the techniques of molecular genetics can be applied to a person of any age, and do not depend upon the at risk person manifesting early signs of the disorder. However, the principles involved are no different for molecular genetic testing than for more traditional diagnostic techniques.

Ethical concerns raised by genetic testing of children

One early concern addressed the ethics of testing children at risk of developing Huntington's disease (HD) in later (adult) life.¹ A consensus was reached by those involved in the predictive testing of those at risk of HD, who decided that children should not be tested.²³ The knowledge of HD gene status can be burdensome for an adult, even when the result is low risk⁴ and only a minority of at risk adults in fact choose to undergo such testing (about 10–15%).^{5–7} The right of the child to decide in adult life whether or not to be tested would be

removed if their genetic status was determined at the request of parents or others; such requests have been made on many occasions.^{8,9} To what extent is HD unique, and to what extent has the discussion about HD served to awaken us to similar issues raised by the testing of children for other disorders?¹⁰

Similar ethical concerns arise in the case of other adult onset neurodegenerative disorders such as Alzheimer's disease and prion dementia.¹¹ We already know that discrimination against persons on the basis of their genetic constitution has been practised by insurance companies in Britain and North America¹² and has even been practised by employers against healthy workers who happen to carry a recessive disorder, such as sickle cell trait.¹³ If children are to have predictive tests performed, they may lose the opportunity to obtain life insurance before clarifying their genetic status. Harper^{14,15} has suggested a moratorium on the use of genetic testing by insurance companies to avoid generating new problems without an opportunity for reflection and for the development of a socially accepted consensus.

Two areas were regarded as potentially problematical. (1) Predictive testing of apparently healthy children (that is, presymptomatic testing) for late onset disorders, usually of autosomal dominant inheritance, in which clinical manifestations are unlikely until well into adult life (and in which early treatment or surveillance for complications would not be helpful). (2) Testing healthy children to determine their carrier status for inherited disorders that would have no implications for their own health, but might affect the health of their future children (usually autosomal recessive or sex linked inheritance, or balanced chromosomal rearrangements).

In both these areas, children's future autonomy, their ability to decide for themselves at some stage whether or not to be tested, could be undermined. In addition, the confidentiality to which any adult being tested would be entitled, would have been breached when the results were disclosed to the child's parents.

The potential harms caused by childhood genetic testing might include damage to the child's self-esteem, distortion of the family's perceptions of the child, loss of future adult autonomy and confidentiality, discrimination against the child in education, employment, or insurance, and adverse effects on the child's capacity to form future relationships. Such testing breaches the policy of providing counselling before or in parallel with the testing process, when viewed from the perspective of the child being tested; this could damage the professional attempts to provide genetic services of counselling and testing together as a package.

Although concerns about predictive and carrier testing in childhood have been voiced, there may be advantages to such testing, including the opportunity for the child to adjust to circumstances, the fostering of openness within the family, the resolution of parental uncertainty, and (for carrier status tests) ensuring that testing has been offered to the whole family. These points will require consideration.

Clearly, considerations of the possible harm done by genetic testing do not apply to a large number of disorders, where it is to the advantage of the child that the condition be diagnosed at the earliest possible stage. This applies wherever therapy, or surveillance for possible complications of a disorder, can usefully be started in childhood. Also, where an affected person is likely to manifest the disorder during childhood, these ethical difficulties of presymptomatic genetic testing in childhood do not arise.

This working party was constituted to address these concerns.

Working party aims

The working party was asked to: examine current attitudes and practices; focus attention on any difficulties raised by the genetic testing of children; and make appropriate recommendations about future practice. These objectives led us to examine two questions. To what extent does such testing take place now? Under what circumstances is such testing justified, and when might it be better deferred until the child is older?

Even before embarking upon this project, we acknowledged that there were likely to be differences of opinion and practice among members of the medical profession. We found such differences, which were reflected within the working party. On the assumption that it is desirable, is it possible for health professionals to work towards a consensus on these issues? Some apparent differences of approach are likely to have arisen for historical reasons rather than from any major philosophical schism: how can such differences be resolved, so as to avoid arbitrary inconsistencies of practice?

Results

Current practice and attitudes in Britain: a questionnaire survey

We wrote to molecular genetic and cytogenetic laboratories engaged in clinical diagnostic work requesting information about the number and nature of the tests carried out on children. We also asked whether requests were declined on ethical grounds.

We carried out a questionnaire survey of approximately 3000 health professionals in Britain (as described in appendix 1), seeking information about their practice and attitudes concerning the genetic testing of children. Detailed results are given in appendix 1.

This survey has confirmed that there is widespread testing of children for genetic disorders, but most of it falls outside our remit: it either involves testing children for disorders that usually become manifest during childhood (or in which there is a significant risk of this happening), or it is otherwise clearly appropriate as part of good medical practice. By this, we mean that these tests are used in management decisions concerning therapy, or the surveillance for complications of genetic disorders.

Among geneticists and co-workers, the pre-

dominant view is that no requests for testing but those initiated by a child's parents or medical advisers should be entertained. This view is shared by paediatricians, except that they would be willing for adoption agencies to initiate such testing. Respondents consider genetic tests in childhood to be justified if a useful intervention can be made as a result (diet, treatment, surveillance for complications), or if medical management may be otherwise improved. There is also general agreement that predictive testing is best avoided among untreatable late onset disorders such as HD, prion protein and Alzheimer dementias, and other neurodegenerative disorders.

In contrast to the consensus on those topics, there was a lack of consensus concerning the wisdom of testing for other late onset disorders for which no useful interventions would be available; 16% of respondents considered that predictive testing in childhood was generally undesirable. There was even some disagreement about whether or not such tests were justified even where some clinical intervention is possible. Predictive testing, it was thought by some, might also be helpful in clarifying the natural history of some conditions.

There was a wide range of views on the question of testing children for gene carrier status where this would have no health repercussions for the child, as with the carrier state for recessive disorders and chromosomal rearrangements. It is certainly common practice among paediatricians to test the healthy sibs of affected subjects to see if they may be carriers of, for example, cystic fibrosis or a balanced chromosomal translocation.

There were also differences of attitude concerning the right of parents to arrange genetic testing for their children. Geneticists and co-workers were less likely to view parental wishes alone as sufficient to justify testing than were paediatricians and others. Only a minority of respondents had a firm age limit below which they did not test children's genetic status, and these age limits varied widely.

Finally, it was clear from the questionnaire replies that geneticists and co-workers are more wary than paediatricians or haematologists of using phrases that could be regarded as paternalistic, eugenicist, or directive. Thus, geneticists and co-workers did not support the notion of genetic testing in childhood to increase "responsible" attitudes to reproduction, whereas other groups did support such sentiments. These differences may arise from different understandings of the meaning or value implications of the word "responsible", or may indicate that the genetics community has become sensitised to such language, and no longer regard it as acceptable terminology. Alternatively, those involved in clinical genetics may have decided that "responsible" reproductive behaviour should be encouraged, but that genetic testing in childhood does not, or in practice is unlikely to, result in such "responsible" adult reproductive behaviour. We cannot distinguish these possible explanations from the replies we received.

Current practice and attitudes in Britain: a prospective study

Because the retrospective gathering of data is difficult, and is likely to be unreliable, we initiated a prospective survey of genetic testing in children (under 16 years) for 12 months from 1.1.92. Seventeen molecular genetic laboratories supplied data, although it was conceded that these were incomplete even when considerable pains had been taken to gather information on all cases. We also circulated clinical geneticists, CGS members, and consultant paediatricians. These data therefore only provide an indication of the pattern of childhood testing in Britain, not an accurate measure of current activity, and the numbers listed are likely to be an underestimate. No attempt was made to enquire into the tests carried out by biochemistry or haematology laboratories, and the generation of carrier status results by newborn screening programmes (for example, for certain haemoglobinopathies) was not studied.

We have listed in table 5 in appendix 2 the numbers of tests reported to us as either performed or deferred in 1992, grouped under the major categories of test. We were informed about 165 tests performed and 37 tests deferred. The most frequent test performed was that for cystic fibrosis carrier status. Forty-two tests (one quarter) were predictive, the rest being carrier status tests. There was no evidence that the tests on children were usually performed in later childhood; the age distribution of children tested appeared to be random, and was not concentrated in the early teenage years. Two exceptions to the apparently random distribution of ages were the tests for fragile X normal transmitting males (the youngest reported was at age 4; before 4 years, the test would also identify children likely to manifest learning problems, and would therefore be diagnostic) and for familial adenomatous polyposis coli (where nine of the children were tested in the age range 9–11 years, presumably as an aid to management, and the remaining 13 children were tested in the age range 0–6 years).

Attitudes of family and patient support groups

A letter and a brief set of questions were sent to 108 separate family support groups affiliated to GIG, the Genetic Interest Group. We hoped to arouse interest in these issues, to elicit accounts of the experiences of families in which tests had been performed in childhood, and to discover the attitudes of those affected by genetic disorders and of members of their families. We received 78 replies; some were from whole groups or local branches of a group, and others were from individual persons within groups. The purpose of this enquiry was not to ballot the members of these specific disease oriented groups, but to obtain information about the range of opinions held within these groups by those whose lives have been touched by the various genetic conditions.

It is possible to group the replies into very

broad categories. Forty-one replies favoured a policy of not performing predictive or carrier genetic tests in childhood.

Another group of 14 replies indicated a willingness to carry out carrier tests in childhood but not predictive tests, although some reservations were made by a few respondents, such as the child having the right to decide at an appropriate age.

The third group of 19 replies indicated a willingness for predictive tests to be performed in childhood. Interestingly, this group was about evenly divided as to whether or not tests for carrier status should also be performed on children.

Four replies were unclassifiable.

Specific comments from respondents are listed in appendix 3.

Legal considerations

Recent developments in child law, most importantly the Gillick decision in 1985¹⁶ and the Children Act 1989, have considerably altered the legal status of children. Whereas it was previously possible to regard children as little more than the property of their parents, this is no longer the case. Instead, parents have responsibility for the care of their children, rather than rights over them, and are expected to act in their best interests. Parental decisions about testing for genetic disorders should therefore be made according to whether the child will benefit, not in order to relieve the anxieties of the parents. If necessary, in cases of dispute, a court can be called upon to determine whether a particular child should be tested, although this would be rare in practice. The courts have suggested that children should be given the chance to take decisions on medical care for themselves if it is possible to wait until they are able to do so without risking their health.¹⁷

The law ensures that decisions about testing are taken jointly by parents and professionals.¹⁸ In effect, parents have a veto because testing cannot be carried out without their consent, although a general consent to diagnostic testing would be sufficient without a full explanation of the conditions being tested for. The consent of any one person with parental responsibility will suffice, unless a court order specifically deals with the matter. Fathers who have never been married to the mother will not normally have parental responsibility. However, in England and Wales, they may obtain it by applying for a court order giving it to them, or making a "parental responsibility agreement" with the mother and having it registered with the court.

Where older children are concerned, they will be able to consent on their own behalf if they have sufficient understanding of the issue to make a choice¹⁶ (Age of Legal Capacity (Scotland) Act 1991, s 2 (4)). By statute, it is presumed that children of 16 have this capacity (Family Law Reform Act 1969, s 8; Age of Legal Capacity (Scotland) Act, s 1), but if a child below that age can understand the test being proposed and its significance then they too will be able to consent. Even if a child is

mature enough to be able to give a consent, recent English cases have established that parents can still authorise treatment, even against the child's wishes.¹⁹⁻²¹ However, even if it is legal to do so, it may well be considered unethical to test for genetic status against the wishes of a child with a good understanding of the issues. Where children do not have the intellectual capacity to agree to treatment, parental consent must be obtained. Such consent may be given in respect of children until they reach the age of 18 years.

Although children cannot be tested without consent, either their own or that of their parents, health professionals cannot be forced to offer care that they consider inappropriate.²² Thus in cases where it would be contrary to the child's interests to be tested, it is legitimate to refuse to do so even if the parents request it. Nor are professionals obliged to reveal information on the genetic status of a child when they believe it would harm the well being of the child. In principle, parents are entitled to see their children's records under the Access to Health Records Act 1990 (applying to manual records created after 1 November 1991) and the Data Protection Act 1984 (for computerised records). However, health professionals are entitled to refuse access where they believe that revealing the information would cause serious harm to the physical or mental health of the child or others.

The general principles of malpractice law also require health professionals to give due consideration to the welfare of the child patient. They will be negligent if they fail to practise in a manner accepted as proper by a responsible body of professional opinion in their specialty.²³ Thus, legal standards reflect professional ones. As long as practitioners conform to responsible medical practices, they will be safe from legal action. This means that as professional ethics develop, legal requirements will also change to reflect the greater understanding. Given the general consensus that it is unwise to test children for HD, to do so might well be regarded as negligent except in very particular circumstances. In other areas, however, both testing and refusing to test would probably be acceptable providing the child's position was considered. However, an over rigid policy might be found negligent because it was not addressed to the particular child's needs and would therefore be unacceptable to professional opinion. Withholding testing could only leave a doctor open to litigation if it appeared to be negligent on the above test.

It should be emphasised at this point that most potential disagreements among family members, or between family members and health professionals, about the desirability of genetic testing for individual children can be avoided by sensitive counselling. This discussion of the legal issues should not be taken to imply that recourse to the law is likely to be required on a regular basis.

Issues relating to adoption

The issues relating to genetic disease and genetic testing in the context of adoption are com-

plex, and are discussed more extensively elsewhere.^{24,25} However, we raise the question as to whether there are particular considerations that might justify the genetic testing of a child being considered for adoption, restricting our attention (as elsewhere in this report) to tests of (unaffected) carrier status, and to predictive tests for adult onset disorders.

Prospective adoptive parents may have a keen interest in the genetic status of a child that they are considering for adoption. Like most people (if given the choice) adopters will usually want healthy children, and the adoption agency is under an obligation to gather information about the child's circumstances, including the health of the child and of members of the family. All relevant available information is then given to prospective adopters so that they can make as informed a decision as possible when deciding whether to accept a child. Good practice indicates that all available information should be passed on.

Adoptive parents are mostly from the 10% of involuntarily infertile couples, but some will have made a conscious decision to limit their family because of a heritable disorder. Their personal experiences are likely to affect their views on genetic conditions in a prospective adoptive child.

TESTS OF CARRIER STATUS

It is our view that there will usually be no particular justification for testing the child any earlier than would be the case with a child still in the birth family. Adoptive parents have the same legitimate interests in the health of their children and grandchildren as do biological parents. However, there are specific difficulties that may arise relating to carrier testing in the context of adoption.

First, the adoptive family may be informed about a family history of possible relevance to the adopted child's future reproductive plans, but may fail to remember this or to pass on the information to the child at an appropriate age.

Secondly, the adoptive parents may focus on the possible carrier state of their child and accord it more emotional significance than is warranted.

In these situations, the lack of familiarity with the condition in question may complicate the task of discussing the issues with the growing child, leading to over- or under-emphasis on the possibility of "genetic risk". Difficulties of communication may also arise when a genetic disease in the birth family comes to light after the adopted child has been settled in the new family. Similar problems of communication may arise in reverse if the adopted child develops a genetic disease after settling in the adoptive family.

Openness between adoptive parents and child, and the maintenance of links between the adoption agency and both the adoptive and the birth families, may help to minimise these problems. However, the questions of principle relating to carrier testing in childhood are no different in the context of adoption from the

issues encountered in other families, even if the pathways of communication are more vulnerable to rupture. They do not amount to a reason for ensuring that the tests are carried out preplacement.

PREDICTIVE TESTING

For adult onset conditions where predictive genetic testing is available, and where the adoptive child is at risk, the question arises as to whether or not the fact of the child's adoption (prospective or established) is a justification for carrying out a predictive test which might not otherwise be performed until the adopted child became an adult and chose to undergo testing.

Arguments for testing include those in table 1, and the specific point that appropriate carers may more easily be found for the child (if preplacement).

Arguments against testing also include the list in table 1, and some specific points related to adoption: that the diagnosis will label the child and affect the (already difficult) process of identity development, that it is irrelevant to the needs of the child for acceptance as he/she is, and that it may cause more problems by excluding paternity.

The arguments will have to be weighed in each case, but their force will not differ greatly from the standard case of a child in the original birth family, unless it proves difficult to find suitable prospective adoptive parents for a child at risk of a late onset genetic disorder because of the uncertainty surrounding the child's possible genetic status. Then, either the decision to put the child forward for adoption, or the decision about genetic testing, will need to be reconsidered. In practice, this situation may arise only infrequently, but will call for a careful consideration of the child's overall best interests when it does so.

In general, it would seem best, wherever possible, to find adopters who can accept the child as a whole, and subsequently participate in any testing that is appropriate for the child as a confirmed member of their family.

Finally, it should be noted that the status of

a child at risk of developing a genetic disorder (even in adult life) has some parallels with the status of a child with special needs. These include financial implications for the local authority, as a child who is at "substantial risk of developing a serious disorder, known at the time of adoption" may require an adoption allowance, payable until the age of 18.

SUMMARY

Inherited disorders have a wide range of significance in adoption because of the multiplicity of viewpoints that exist between the members of the adoption triangle (adopted person, birth family, adoptive family) and the Adoption Agency. When inherited disorders arise in the context of adoption placement decisions, they may be highly significant and have an important bearing on the outcome of an adoption.

The management of genetic disorders in adoption should begin with determining the attitudes and expectations of adopters with regard to present or future disability. Specific issues such as predictive and carrier testing are the same as for any similarly situated person at genetic risk, with the additional consideration of the effect of the decision (and, if a test is performed, the possible test results) on the child's placement. The legal situation regarding consent to testing in the adoption process has not been clarified judicially.

Discussion

The two central questions to be answered, or at least considered, are: What is good clinical practice with regard to predictive or carrier testing in childhood? and Who has the responsibility or the right to arrange predictive testing or carrier testing in childhood: the families, the individual persons (the children as future adults), adoption agencies, or doctors?

Unfortunately, there is a dearth of firm evidence on which to base policies. Various blends of ethics, anecdote, and prejudice are likely to shape people's attitudes, including our own. While there is real concern about the abrogation of a child's future autonomy as an adult, and about the loss of confidentiality entailed in childhood testing, we do not know whether harm actually results from such testing (but see below). There are concerns that the parents' attitudes to the child may change, and that unfavourable parental expectations about a child's future mental, physical, and emotional development could be self-fulfilling. These factors could result in low self-esteem in the child, but there is little evidence that genetic tests produce such damage in practice. This may be because such testing does not cause harm, or because such testing in the past has been limited in scope, and the resulting harm has not come to light because evidence of it has not been sought. We have had to arrive at our responses to these questions in the absence of adequate empirical data, relying upon our various experiences and upon our concern to maintain the generally accepted principles of medical ethics.

Table 1 Possible advantages and disadvantages of predictive testing in childhood

<i>Advantages</i>	
(1)	Relieves anxiety about possible early signs of the disorder.
(2)	Family uncertainty about the future is reduced.
(3)	More accurate genetic counselling becomes possible.
(4)	The child's attitude towards reproduction in adulthood will be more responsible.
(5)	Children who might benefit from genetic counselling in the future will be identified.
(6)	Practical planning for education and career, housing, and family finances becomes possible.
(7)	Parental expectations of the child's behaviour become altered.
<i>Disadvantages</i>	
(1)	Removes the child's right to decide whether or not to be tested in adulthood.
(2)	Parental expectations of the child's future reproductive behaviour become altered.
(3)	Damages the child's sense of self-esteem.
(4)	Generates unwarranted anxiety about possible early signs, before any genuine manifestation of the disorder.
(5)	Leads to future difficulties in obtaining life insurance.
(6)	*Rarely leads to clarification of the genetic status of other family members.

* This is not so much a disadvantage as a lack of advantage, but was included as a disadvantage for the purposes of the questionnaire.

Predictive testing (where there is no direct health benefit to the child). "What is good practice with regard to predictive testing in childhood?"

The working party has arrived at a consensus view that there should be a general presumption against such testing, although we recognise that circumstances may arise when testing could be appropriate. We now set out the reasoning that has led us to this common understanding.

There is the precedent of the decision made by those performing predictive testing for HD, a consensus reached on the basis of the supposed likely effects of such testing and not from the experience of harmful effects (the consensus was reached before such testing of children was ever undertaken). In families with HD, persons may be singled out for no rational reason as being destined to develop the disorder. Such family "preselection" of children as being likely to develop HD is recognised as having potentially profound adverse effects on the emotional development of the unfortunately labelled child.²⁶ These effects could be even more severe if medical evidence supported, and perhaps directed, the collusive family dynamic that has selected one child as the "victim".

Adults undergoing predictive testing for HD have extensive counselling, in which they are required to confront the effects that the test is likely to have on their whole life. Insurance (of life and health), home ownership, career, marriage, and reproductive plans may all be deeply affected by the results of the testing. Only some 10 to 15% of at risk adults choose to accept such testing.⁵⁶ It may be difficult for parents to appreciate the reluctance to undergo testing that is felt by many adults at risk of HD, and which their at risk child may also come to experience. A sensitive exploration of the issues will often help parents to appreciate the limitations of their perspective, and how their feelings may differ from those of their child in the future; this may lead them to reconsider their request for testing. However, where the family has already "preselected" the child, as being affected or unaffected, they may continue to press for testing. In these circumstances, it could be particularly inappropriate to perform the test.

There is little evidence as to ill effects resulting from predictive testing in childhood for Huntington's disease or for other disorders, although the fundamental principles involved are much the same for any late onset condition, especially for other neurodegenerative disorders. Only limited evidence is available concerning the Swedish newborn screening programme for alpha-1-antitrypsin deficiency, which was discontinued because of severe adverse psychosocial consequences.²⁷ However, it is clear that predictive testing in childhood for late onset disorders, even in the context of a high risk family situation, can raise as many problems and as much anxiety as is generated by continuing anxiety about the child's genetic status, and the knowledge that a child will develop such a disorder may, at least in some family contexts, cause worse problems than continued uncertainty. At present, we have no

means of identifying those families that would be helped by having uncertainty resolved by genetic tests and distinguishing them from other families in which the results of testing would be harmful. Indeed, there is not even any agreed means of deciding whether or not a family might have been helped or harmed by such an intervention, and what time scale should be considered in coming to a judgement on this.

Given the dearth of evidence, it is the ethical consequences of childhood testing (loss of adult autonomy and confidentiality, and the possibility of causing harm to the developing child), together with the limited empirical evidence that is available, that has led us to advocate this cautious policy, erring towards a presumption of non-maleficence ("primum non nocere – the first goal is to cause no harm").

Furthermore, there are now good legal reasons for professionals to make decisions in this area primarily on the basis of the long term best interests of the child; failure to do so could lead to professionals later being sued for acting against the interests of the child, even if this was at the request of the parents.

Despite our judgement that it is wise to avoid predictive tests in childhood, we recognise that experience and information may accumulate over time, and the apparent disadvantages of predictive testing for HD in childhood may not apply in practice to all other late onset disorders, or even to Huntington's disease in every situation and for all time. The situation will certainly change when effective interventions become available for these conditions.

If the child stands to gain some health benefit from predictive testing, then it is obviously good practice to make the testing available. However, predictive testing carried out to satisfy the curiosity of a medical practitioner (except in the context of certain types of research, see below), or to relieve parental anxiety, would not seem to us in general to be appropriate. There may be circumstances where such testing is justified, but we expect these circumstances to be unusual. In our judgement, the possible emotional harm to the child, the abrogation of their autonomy and the breaching of their confidentiality, will generally outweigh the possible benefits that we can see.

An additional caution about the use of predictive tests in childhood must be noted: gene testing is not a substitute for good clinical assessment. This issue has been raised previously in the context of HD, where the occurrence of juvenile onset cases is infrequent but well recognised. If an at risk child develops clinical features compatible with the early stages of juvenile HD, then some alternative explanation for the suspicious clinical features may still be more likely to be correct, because the early signs of HD can be gradual and non-specific. In such circumstances, performing a predictive genetic test (not "presymptomatic" in the usual sense of the word) may be considered, but it is unlikely to be helpful.³²⁸ Showing that a child has a mutation in the HD gene does not indicate that the clinical features are

caused by HD, and may discourage the further diagnostic efforts needed to arrive at the correct explanation. If the child's symptoms are caused by another disorder, then an unfavourable predictive test has been carried out on a minor without their consent.

Similar scenarios can arise with myotonic dystrophy and other mendelian disorders. Identifying children as being at increased risk of developing one of the common multifactorial diseases (cardiovascular disease, hypertension, diabetes melitus, cancers, ...) has the same drawbacks as predictive testing for late onset monogenic diseases unless there is some demonstrably effective therapy, or unless the testing is carried out as part of a therapeutic trial in which the allocation of treatments depends upon identifying the subjects with increased risk at the outset.

Carrier testing

The arguments here are less clear, and no consensus exists among health professionals. The working party has again arrived at a consensus view that carrier tests in childhood should generally be deferred. The arguments for and against testing can be (and indeed already often are) discussed openly in genetic counselling sessions with the parents of children who might be carriers of a genetic disorder. In most cases, this will rapidly lead to a consensus that testing can be deferred without any penalty, to allow the child eventually to participate in the decision as an autonomous person. We present the contrary arguments and the reasons for our view in the next six sections.

It may be better for a child to know about their carrier status in childhood: this may then be accepted as a simple matter of fact, without the emotional problems that could arise from a later disclosure

This argument would only be valid if the one alternative to testing in childhood were to be silence on the issue within the family. We would like to emphasise that a decision not to test a child for carrier status is not intended to discourage a family from talking about the possible carrier status of the child. In practice, talk about such matters may actually be easier without firm knowledge; it is not difficult to explain to a child that the decision as to when and whether to test is being left for them to decide once they are older. This explicit granting of control to the child may actually be helpful in enhancing their self-esteem, and in allowing them to come to terms with possibly unfavourable results once adult.

We recognise that 50% of "possible" carrier children would be shown not to be carriers by testing in childhood, and that life might be simplified for them by childhood testing. However, it is our judgement that the possible benefits experienced by these children must be balanced against the possible harm done by identifying carriers in childhood. Such harm might be especially likely in a family where some children are shown to be carriers, and

others are shown not to be. There may also be families in which being identified in childhood as not a carrier could be emotionally disadvantageous.

Testing their child may decrease the level of anxiety of the parents simply by giving them this knowledge

But it may well increase their anxiety if the child is shown to be a carrier, and the testing may need to be repeated in any case, as techniques and methods improve.

There is no direct evidence to indicate that carrier testing in childhood is harmful

Nor is there good evidence of benefit, and some evidence has accumulated suggesting that knowledge of carrier status can have adverse effects, which might be magnified if the carrier (or the non-carrier) is identified involuntarily when young, and is labelled in this way by the whole family unit. We have arrived at our judgement on the basis of the currently available information, and recognised that further research is needed to examine these issues.

Problems may arise with carrier testing because of stigmatisation or the fear of stigmatisation, even in an area where the carrier frequency is high.²⁹ Adult carriers of Tay-Sachs disease have been shown to view their future health with less optimism than others³⁰; it is not clear how this would affect those carriers identified in childhood. An earlier study has shown that 19% of Tay-Sachs carriers were worried about their carrier status at follow up some years later,³¹ and 10% of sickle cell and thalassaemia carriers in Rochester, New York are unable to state that being a carrier will not damage their health.³² We do not know whether children identified as carriers will have the same degree of misunderstanding, or less, or more. The wider issues of social stigma have also caused concern even in the recent past, particularly in relation to sickle cell carrier screening in the USA.¹³³³

Some evidence is emerging of the distress caused by carrier testing for cystic fibrosis in the healthy adult sibs of affected subjects.³⁴ The limited experience with population screening for cystic fibrosis in Britain indicates that many adults, even some of those related to known carriers,³⁵ decline the offer of carrier screening tests. Uptake is influenced predominantly by how the test is offered,³⁶³⁷ so public enthusiasm for the tests may be substantially less than the enthusiasm of the health or health research professionals, and the future adult enthusiasm of the present child cannot be taken for granted.

Carrier testing in childhood avoids urgent carrier testing in young teenage pregnancies (often concealed until late pregnancy)

Sexual activity, however, is not a reason for genetic testing, and it should not be presupposed that such teenagers will all want carrier testing and possible prenatal diagnosis,

calling for a decision about a possible termination of pregnancy (in fact, could some teenage pregnancies in at risk families be concealed so as to avoid such tests?) A number of alternative practical approaches can be adopted. First, the question of their possible genetic risk can be put to the adolescent within the family setting or by their family doctor, and referral for genetic counselling can be arranged if and when they choose. To push such counselling or testing upon an adolescent (or anyone) is unacceptable, and it is likely to generate more problems than it "solves". Furthermore, adolescents who may be carriers of a genetic disorder, like other adolescents, should be given proper information about human reproduction and contraception.

Testing in childhood ensures that the testing is at least performed, and may absolve the doctor of his responsibility in this regard

However, imparting the information to the family does not ensure that the correct information will be given to the child in an appropriate fashion and at an appropriate age. If parents in practice find it difficult to impart the facts of sexual intercourse and human reproduction to their children, will they find it easier to discuss genetic disease? Anecdotes reported to us suggest not.

When carrier tests are carried out in childhood, we believe that there is still a responsibility on the medical profession to ensure that the future adult is offered genetic counselling at an appropriate age. This is both to ensure that the adults have the opportunity to discuss the issues relevant to themselves, and because the facts given to the parents, in connection with the particular disease prognosis or diagnostic testing, may no longer apply because of advances in medical knowledge.

Carrying out carrier tests in childhood, then, does not absolve the doctor of responsibility for ensuring that the future adult is offered genetic counselling at an appropriate age. While the initial scenario may work well in some families, it must be remembered that many families do not conform to the stereotype nuclear family structure. In a social context with parental fluidity and serial step parenting, and because of the emotional responses of guilt, blame, responsibility, and denial in relation to their genetic disease in many families, it is not possible to rely upon the effectiveness and sensitivity of intergenerational communication. In addition, to set up testing of children in this way runs counter to a core principle of clinical genetic practice, by separating the testing from the counselling by a gap of years. Finally, the carrier testing methods may change, so that further analysis may be appropriate in any case once the child is adult and beginning to plan a family.

If testing is not performed in childhood, how may a doctor fulfil his responsibility to offer testing to all relevant persons in the family?

There are several possible ways of ensuring that

a child is offered testing at an appropriate age. These include: (1) a clear transfer of responsibility from doctor to family, so that the onus of offering genetic counselling and testing to the child (once adolescent or adult) lies with them. This may be the only reasonable option if the family is planning to emigrate; (2) a clear transfer of responsibility to the child's family practitioner, who can prompt a genetics referral when the child is old enough to be directly interested in the issues; (3) continuing involvement with the family in the form of an active genetic register, with regular, long term follow up to keep in touch with family members even when they move area.³⁸⁻⁴¹

In general, we would favour the third option, of involvement on an active follow up register. However, it is clear that this is the most expensive option, because the resources required to establish and maintain such registers are considerable. We must also emphasise that testing in childhood does not in itself remove the physician's responsibility to ensure that the child is offered counselling when older.

Summary

The above arguments tend towards the view that carrier testing for genetic disease in childhood may not be the most appropriate approach, at least if practical alternatives can be devised and established. Inadvertent carrier testing (from research or from prenatal diagnoses that indicate an unaffected, but carrier fetus) should be avoided wherever possible. Research is needed to ascertain the psychosocial consequences, the adverse effects or the benefits, of carrier testing in childhood.

Unsought information in research and clinical practice

It is possible for unsought information to be generated about a family in the course of research, for which consent has not been obtained, and which is unlikely to be of use in the research. This is particularly likely to occur when blood samples have been collected on all available family members as part of a preliminary gene localisation (linkage) study, when these samples are stored in a research unit, and when tests are carried out on the stored research samples some years later once a specific test has been developed. This amounts to genetic testing without counselling or consent, and is likely to arise when samples have been collected indiscriminately from all available family members, and when the samples are tested wholesale in a manner for which consent was not obtained from the family (because the test was then not available). This can generate difficult practical and ethical problems, but these can often be avoided by careful advance planning, attention to detail in research consent procedures, and a clear separation of research work from service results and clinical records. This area has been discussed by Harper,⁴² and guidelines have been proposed. A specific effort on the part of clinical and laboratory staff will often be required to ensure that information is not gen-

erated on family members which has not been requested by them; once such unsought information is available, it may be difficult to prevent its being passed to the family, perhaps inadvertently and possibly resulting in serious family distress.

Predictive testing carried out as part of a research study into the consequences of early diagnosis may well be legitimate, but attention should be paid to the guidelines of the Medical Research Council Working Party on Research on Children⁴³ and the British Paediatric Association's Guidelines for the Ethical Conduct of Medical Research Involving Children.⁴⁴

Unsought information may also result from the genetic testing of children in standard clinical practice, and we report one scenario that illustrates some of the problems that can result. In a family in which some members carried a balanced chromosomal rearrangement, the children were tested cytogenetically when very young, and at least one child was found to carry the rearrangement in balanced form. The family subsequently attended for counselling because they did not know how to give their teenage daughter her (positive) tests results; it was decided to offer her a test once she expressed interest, as if she had never been tested in early childhood, rather than tell her that she had in fact been tested before and that her parents had known her test result for years.

Who has the right or responsibility to arrange for the genetic testing of children?

PARENTS

There is no clear consensus on this issue among British professionals. One legal view from North America would hold that parents are likely to have the right to the full disclosure of any genetic information about the child to which they want access,⁴⁵ although this has not yet been tested in the courts. It has also been suggested that geneticists may have an obligation to carry out such testing, and it has been emphasised that each child's case must be considered individually; no general policy would be defensible.⁴⁶ The legal situation in this country is different, and to our minds more satisfactory, in that any decisions must be made on the basis of the best interests of the child, although again no blanket policy would be defensible. Given that the genetic test results will have no medical management implications for the child, and that the uptake of carrier and late onset predictive tests by adults in Britain is low, the arguments in favour of parental rights to demand this information are weak, although the parents may have the right to demand alternative means of ensuring that testing and counselling are offered when the child is older.

ADOPTION AGENCIES

Adoption agencies are recognised by paediatricians as having a legitimate interest in the genetic status of prospective adoptive children. This view is reasonable, in so far as the result

of such testing will influence the likelihood of a child being accepted by suitable adoptive parents. However, it may be better for a child to be adopted by parents whose willingness to adopt is not dependent upon the results of predictive or carrier genetic tests.

MEDICAL PRACTITIONERS

Where testing is to the possible medical advantage of the child, it is clearly the duty of medical practitioners to ensure that it is carried out. Where testing may be of interest to the future adult, for health reasons or to permit informed reproductive decision making, the offer of counselling (and possible testing) should be made once the person is mature or in early adult life; this may require the establishment of an active genetic register.

With respect to carrier testing, or to predictive testing for a late onset disorder, the medical profession needs not assume any obligation to initiate such tests. When such testing of children is requested by others, however, doctors have a right to refuse to carry it out if they consider that it may cause harm to the child or is not in the child's best interests.

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Appendix 1. Results of the questionnaire study

Molecular genetics laboratories

We sent questionnaires to 35 of the laboratories listed in the Clinical Molecular Genetics Society handbook, targeting those ones that we thought most likely to be involved in diagnostic work and including all Regional NHS laboratories. We received replies from 16 laboratories, of which half did not carry out testing that would fall within our definition as being potentially problematical.

Some laboratories carry out the genetic testing of young children to determine their disease status, and generate carrier status information incidentally. This information is not then usually withheld from the family. Several laboratories carry out predictive testing for disorders such as familial adenomatous polyposis, myotonic dystrophy, and adult polycystic kidney disease. Testing is also carried out for some X linked disorders that can affect females (OCT deficiency, fragile X mental retardation). In these conditions there is a distinction to be drawn between testing for a child who may be clinically affected and one in whom evidence of carrier status is being sought. Six of the laboratories regularly test young children specifically to determine their carrier status for recessive conditions such as cystic fibrosis or sex linked muscular dystrophy (Duchenne or Becker). Presymptomatic diagnosis of Leber's optic atrophy has been carried out in a young girl. Two centres specifically stated that it was their policy not to carry out any such predictive or carrier tests on children.

Cytogenetics laboratories

Ten of the 23 regional or subregional cytogenetics laboratories to which questionnaires were sent provided us with detailed information about their testing of children, with particular emphasis on the determination of their carrier status for familial chromosomal rearrangements. Several other laboratories responded with general statements and observations. The picture that emerges is that most laboratories examine samples from several children each year in order to determine whether or not they carry a familial chromosomal rearrangement. However, several reasons were given to account for some of these tests that do not necessarily apply in all situations. First, some tests are carried out at birth to confirm a prenatal test result. Second, the results of the test may be used to assist with the interpretation of test results in other family members. However, there is probably still a substantial number of children tested where these considerations do not apply, and where the questions raised by our working party do need to be addressed.

It was put to us that cytogenetics laboratories generally accept samples referred from paediatricians, geneticists, or other clinicians in good faith, without questioning the ethical or clinical judgements involved; it is primarily with the clinicians that these issues need to be explored. The same argument could be used by molecular laboratory staff. Whether or not this argument is an adequate response to these issues may deserve wider discussion.

Clinicians and genetic co-workers (nurses, counsellors, etc)

The questionnaire was circulated to members of several professional groups, including members of the Clinical Genetics Society (550, including some paediatricians and genetic co-workers), members of the Genetic Nurses and Social Workers Association (73), consultant members of the British Paediatric Association (990), members of the British Paediatric Cardiac Association (100), members of the British Society for Haematology (780), the Society for Endocrinology (1000), the British Paediatric Neurology Association (20 non-BPA members), Association of British Neurologists (500), the British Association of Paediatric Surgeons (87), general surgeons with an interest in familial adenomatous polyposis (44), and to selected ophthalmologists (11). Respondents are shown in table 2. Because of the importance of obtaining responses from a representative group of paediatricians, and because of the low response rate from paediatricians as a whole, a second copy of the questionnaire was distributed to the non-respondents among a randomly selected group of 10% of consultant paediatricians.

Table 2 Clinical questionnaire

Specialties of responding clinicians	Responses	Questionnaires sent
Consultant clinical geneticists	32 (58%)	55
Genetic co-workers	17 (23%)	73
Consultant paediatricians	337 (34%)	990
Consultant paediatrician members of the British Paediatric Association		
Other members of CGS (Clinical Genetics Society)	14	470
"10%" group of paediatricians (of all consultant members of British Paediatric Association)	50 (42%)	118
"FAP" surgeons	14 (32%)	44
Paediatric surgeons	14 (16%)	87
Ophthalmologists	7 (64%)	11
Haematologists (45 adult, 6 paediatric)	51	
Other physicians	30	

The proportion of respondents for each professional group is not precise because of some overlap of memberships between different groups (for example, CGS and the Genetic Nurses and Social Workers Association; CGS and the British Paediatric Association).

The low response rate from "other" CGS members appears to have resulted from the reluctance of non-consultant clinical geneticists to reply to a questionnaire that required them to list "their" cases in which childhood testing for genetic disorders was proposed; such cases, it was felt, would be best reported by the appropriate consultant so as to avoid a duplication of reports.

PREDICTIVE TESTING

Only 184 of the 512 respondents (36%) had received requests to carry out predictive testing of 902 children. In addition, some respondents were involved in newborn screening programmes for haemoglobinopathies and other disorders. Many of the tests discussed were potentially of health benefit to the child (for example, hyperlipidaemias, medium chain acyl-CoA dehydrogenase deficiency, family cancer syndromes, haemoglobinopathies, cystic fibrosis, alpha-1-antitrypsin deficiency) or were for disorders likely to become manifest in childhood.

(adrenoleucodystrophy, Becker or Duchenne muscular dystrophy). Requests for predictive testing were declined or deferred by a number of persons, principally for neurodegenerative disorders (HD, Charcot-Marie-Tooth disease, myotonic dystrophy) and also for retinitis pigmentosa (RP), autosomal dominant polycystic kidney disease (APKD), and familial adenomatous polyposis coli (FAP). Some requests for the predictive testing of children for myotonic dystrophy, RP, APKD, and hypertrophic obstructive cardiomyopathy (HOCM) were accepted and carried out.

Only a few of these requests originated from outside the nuclear family, typically from adoption agencies (preplacement).

CARRIER TESTING

A greater proportion of respondents (41%) received requests to determine the carrier status of healthy children for inherited

Table 3 Differences in attitudes between professional groups

A "Do you agree that the following have the right to request that genetic testing (predictive or carrier status) be performed on a child, even if the result will have no direct health benefit for the child?"

	Affirmative replies (% of respondents in each professional groups)			
	Parents	Extended family	Adoption agencies	Medical practitioner
Geneticists (n=46)	57*	6	28	21
Co-workers (n=17)	29	0	12	12
Paediatricians (n=337)	75	10	55	42
"10% Paeds" (n=50)	72	16	48	40
Haematologists (n=51)	76	14	37	61
All (n=512)	71	11	47	41

* Of the 27 geneticists (59%) who thought that parents should be able to arrange for genetic testing of their child(ren), 18 (39% of all the geneticists) would permit this for certain disorders only.

B "It is up to the family to decide whether or not their child(ren) should be tested for genetic disorders (for predictive or carrier tests)".

	Agree (%)
Geneticists	46
Co-workers	35
Paediatricians	79
"10% Paeds"	78
Haematologists	84
All	74

Table 4 Responses to supplementary questionnaire

"Would you test a 5 year old child whose parents wanted to know the genetic status for the following disorder(s)?"

Disorders	Geneticists (n=49)		Paediatricians (n=260)	
	Yes	No	Yes	No
Predictive testing				
Adult polycystic kidney disease	27	17	198	29
Becker muscular dystrophy	30	15	189	38
Charcot-Marie-Tooth disease	11	28	154	46
Facioscapulohumeral muscular dystrophy	8	30	156	50
Huntington's disease	2	47	100	89
Hyperlipidaemias	40	7	223	16
Hypertrophic obstructive cardiomyopathy	37	8	201	13
Leber's optic atrophy	20	18	152	40
Marfan syndrome	43	4	199	24
Multiple endocrine neoplasia	32	9	160	22
Myotonic dystrophy	19	26	182	42
Neurofibromatosis I	30	13	207	29
Polyposis coli	28	16	214	14
Prion protein dementia	0	42	66	48
Retinitis pigmentosa	20	22	177	35
Spinal muscular atrophy	16	23	163	45
von Hippel-Lindau disease	36	8	143	22
Unaffected carrier status testing				
Alpha-1-antitrypsin deficiency	19	27	132	92
Cystic fibrosis	22	27	170	74
Haematological disease	22	28	155	74
Spinal muscular atrophy	17	30	130	98
Tay-Sachs disease	20	30	142	88
Adrenoleucodystrophy	12	32	132	74
Becker muscular dystrophy	11	33	136	82
Choroideraemia	11	32	75	81
Duchenne muscular dystrophy	14	32	163	75
G6PD deficiency	13	32	139	90
Haemophilia (A and B)	13	33	159	74
Immune defects	12	31	139	79
Norrie's disease	12	30	81	77
Retinitis pigmentosa	11	32	130	78
(Balanced) chromosomal translocation	27	20	146	83

disorders. As in predictive testing, most of these professionals were clinical geneticists, paediatricians, or haematologists. The bulk of such testing involved familial chromosomal rearrangements, Duchenne and Becker dystrophies, cystic fibrosis, and the haemoglobinopathies; almost all of these tests were carried out, but some respondents who discouraged such tests did not report the number of tests requested. Carrier status tests for X linked visual disorders, coagulopathies, and fragile X mental retardation were also carried out regularly.

A number of requests for carrier testing were declined or deferred, for example, for CF, DMD, chromosome translocations, etc, although only a minority of respondents (8%) had a firm age limit below which they did not carry out such testing, and the age limit varied from 1 to 18 years. Of the 276 respondents who had received no requests, 28 discourage such tests in childhood, 123 refer all such requests elsewhere, and 130 had never been asked. Only a few requests were received from outside the family. The numbers of children on whom testing was performed or requested was reported as 1706 children annually. Although this is probably only a proportion of the tests carried out, it is clear that a substantial number of children are being tested for their carrier status.

Attitudes

PREDICTIVE TESTS

Predictive tests in childhood were thought to be justified under some circumstances by 66% of respondents on the grounds of a health benefit to the subject, while 16% of the respondents thought that such testing was not justified. Those who considered such testing to be helpful gave many examples of disorders where this could be so. These disorders included the hyperlipidaemias, FAP, Wilms' tumour, retinoblastoma, and Marfan syndrome. Some respondents stated that they would only carry out such testing if they had available a useful intervention (for example, treatment or surveillance for complications). A minority of respondents wished to offer testing for disorders where there are no clearly established useful interventions, including HD, HOCM, Leber's optic atrophy, and myotonic dystrophy.

Attitudes to predictive genetic testing in childhood were explored further in very general terms. Respondents were asked to indicate the strength of their agreement or disagreement with the set of statements about genetic testing (table 1). The seven points given as possible advantages of predictive testing in childhood were all regarded positively by a large majority of respondents, although not so many supported the notion that the child's attitude to reproduction as an adult would be "more responsible" as a result of testing in childhood. On this question, most geneticists and all fieldworkers expressed disagreement, whereas a majority of paediatricians and others supported this judgement. A majority thought that testing in childhood would also permit more accurate genetic counselling of other family members.

RIGHT TO REQUEST GENETIC TESTING

The next question, concerning who had the right to request the genetic testing of a child, showed marked differences in attitude between the members of different professional groups (table 3). Because the response rate to the questionnaire was poor (only 58% of consultant geneticists and 34% of consultant paediatricians), it is not clear how representative the respondents are of their professional groups. However, the attitudes of the group of 10% of consultant paediatricians who were given a second mailing if they had failed to respond to the first, were very similar to those of the other responding paediatricians. It is therefore reasonable to interpret the questionnaire results as if the respondents' views are representative of their professional groups.

A majority of genetics co-workers stated that not even parents should have the right to request the genetic testing of their child. While clinical geneticists were divided on this issue, they thought that this right should apply to testing for certain disorders only. In contrast, a majority of respondents overall (364=71%), and of paediatricians and haematologists in particular, thought that parents should have the right to request the genetic testing of their child. This was confirmed by the responses to a separate question (table 3), suggesting that the "right to request" was generally interpreted as a right to have the test performed. A very substantial majority of all groups thought that social services (unless having parental responsibility), legal authorities, the extended family, and the education authorities should not have this right. Some generalisations concerning differences between professional groups can be made. Most haematologists who responded thought that medical practitioners but not adoption agencies should have this right. Paediatricians were divided on the question of doctors' rights and of adoption agencies' rights, but were more in favour of adoption agencies' rights than were other groups; clinical geneticists and fieldworkers would restrict the rights of both these groups of professionals.

POTENTIAL DISADVANTAGES OF TESTING IN CHILDHOOD

Turning to the potential disadvantages of carrying out tests in childhood, a majority also agreed with the statements that such tests limited the child's future autonomy, could lead to difficulties obtaining life insurance, and could have an adverse effect upon parental expectations of the child's future reproductive behaviour (table 1). Additional potential disadvantages that were put forward included future difficulty in obtaining training and employment, and the disturbance of the

parent-child relationship leading to impaired "bonding" and the possible rejection of the child by the parent.

Despite the lack of firm evidence on which to base their views, and the broad range of opinion found within each professional group, we did find significant differences of opinion between professional groups. Paediatricians were evenly divided as to whether or not the child's sense of self-esteem could be damaged by testing, whereas a small majority of geneticists and a large majority of fieldworkers thought that this could be an important disadvantage of such testing. A majority of all respondents also considered that a diagnosis in childhood could result in excessive anxiety about possible early signs of the disorder, just as a majority of respondents also considered that it could lead to the resolution of such anxiety.

The questions we had identified as issues to be considered were recognised as potential issues, and additional distinct issues were not identified. There was some evidence that respondents were more confident of recognising potential advantages of testing than they were of recognising the potential disadvantages (fewer responses to the statements of potential advantages of testing indicated neutrality, and substantially more responses indicated a strength of feeling).

One set of respondents thought that children might adjust to unwelcome genetic information more readily in middle childhood than in adolescence. Others expounded the opposite view, that such a process could be harmful or misleading. A large majority of all groups thought that children should be consulted on these questions once they are sufficiently mature.

There were several reasons for considering the process as potentially harmful. The disorder may be variable, so that the previous experience in the family merely causes confusion. Children may be singled out to their own detriment or that of their sibs. Healthy children may be "medicalised" when they are still very impressionable. The test results may be given an excessive emotional weight in some families, generating disproportionate levels of anxiety. Some respondents thought that, by performing the test only at the specific request of the (now adult) persons, one is allowing them a degree of control that may be helpful in their coming to terms with unwelcome information.

Supplementary questionnaire

Some months later a second questionnaire was distributed to members of the Clinical Genetics Society and to consultant members of the British Paediatric Association. This asked whether or not the respondent would be willing to arrange genetic testing of a healthy 5 year old child for a number of different disorders (predictive tests for some dominant and sex linked disorders; carrier status tests for some autosomal recessive and sex linked disorders and for balanced chromosomal translocations). It was hoped that differences in the acceptability of

testing for different disorders might allow inferences to be drawn about the factors that influence professional attitudes.

INTERPRETATION OF SUPPLEMENTARY QUESTIONNAIRE RESULTS

Responses were received from 260 paediatricians, 49 geneticists, seven co-workers, and eight haematologists — 324 in all. The same trends appear in these data as in the longer, original questionnaire: paediatricians were generally much more willing for genetic tests to be performed in childhood than either geneticists or co-workers. Fewer firm opinions were expressed concerning the more uncommon disorders. There was little to suggest that the availability of an early treatment or surveillance for complications for a disorder made any difference to the readiness to test for it.

Respondents fell into two broad groups, that in which most tests were approved, and that in which most would be refused. The most important factor influencing responses seems to have been the respondents' attitudes to such testing in general, and the nature of the various disorders seems to have had relatively little influence. Most respondents answered similarly for all the diseases for which they stated a view (although the proportion of questions answered with a "Don't know" did vary between persons). Only with Huntington's disease and prion dementia was there a suggestion of a widespread unwillingness to carry out predictive testing on a young child. Even with these conditions, it was striking that many paediatricians expressed a readiness to carry out presymptomatic predictive testing. The few paediatric haematologists surveyed expressed uncertainty about testing for most disorders, and were divided in their willingness to test children to determine their carrier status for haematological disorders. The further analysis of these responses, and the exploration of the reasons for this pattern of results, will need to be studied as a future piece of research, and is beyond the scope of this report.

Appendix 3 Attitudes of family and patient support

Comments on the replies from the group which favoured a policy of not performing predictive or carrier tests in childhood included remarks such as "at least wait until child able to give informed consent", and "once of an age to be sexually active". This group included all six replies from adults with spinal muscular atrophy. Those opposed to the principle of predictive or carrier testing in childhood generally favoured the right of health professionals to make decisions about whether or not to proceed with tests requested by parents. Those in favour of carrying out such tests generally opposed this type of professional control, regarding it as excessively paternalistic.

Remarks written in opposition to the principle of genetic testing in childhood included:

"Testing will destroy the innocence of childhood."

"Good counselling — yes; testing — no."

"If there is some preventive measure ... then tests should be done, but otherwise no."

"I don't think it should be done just for the parents' peace of mind..."

"We feel it is not the parents' right, but the 'child's' right in adulthood. Doctors should be able to refuse the tests in order to protect the child."

"I wanted my son tested to see if he was a carrier of PKU... but after long discussions my husband and I decided it would serve no purpose other than labelling him... Other more serious disorders would need a lot more thought but I think the same decisions should be made."

"It is important, I think, for a child to grow up knowing that there is a chance that he/she is a carrier for something—but not until she/he is at the older age when they are thinking of partners and producing offspring should it be necessary to do any testing..."

"Predictive testing would be just another example of projected unethical genetic cleansing."

"If as a result of trying to wipe out the disease, there were fewer remaining, then society may become less tolerant."

Remarks written by those in general support of childhood testing included:

"A parent's job is hard enough. If there is the possibility that their child will become seriously ill at whatever age, then parents should have the opportunity to either allay their fears and the stress on the family or alternatively to make provision for the support — moral, emotional, financial — of their child. A parent should have the right to ask for testing but (should) not be "pressured" in any way. The decision to test or not should be with the parents."

"Doctors should have no say in the matter."

"I think it (predictive testing) is essential, and that persons with a potential disorder in later life should be made aware so that they can plan for their futures, and be prepared (for) what to expect."

"This issue (carrier testing in childhood) is simpler (than predictive testing). I feel that testing of children in this case would be not only right, but vital. An informed decision on reproduction could be made."

There were two further comments that provide interesting perspectives:

"Decisions on carrier testing should be left to the child when they are old enough... GPs should ensure that such children fully understand the risks before the decision to start a family. Support and counselling are needed and cannot be left to parents."

"As with all matters relevant to a child, the fundamental criterion has to be what is in the best interests of the child. This may be a decision capable of being made by the parents or doctors, but not necessarily either."

Appendix 2

Table 5 Prospective study of testing in molecular genetics laboratories 1992

Genetics tests	Performed	Age range	Deferred
Alpha-1-antitrypsin deficiency (carrier test)	1	9y	0
BCR	13	Infancy-14y	2
BMD predictive	4	1-12y	0
CF carrier	68	Infancy-15y	9
DMD carrier	5	8-15y	4
Fabry's carrier	1	3y	0
FAP	22	Infancy-11y	0
FSH dystrophy	1	10y	0
Fragile X carrier	10	4-14y	1
Fragile X NTM	5	4-14y	0
Haemoglobinopathy carrier	10	1-14y	5
Haemophilia A carrier	8	3-14y	0
HMSN I	1	Birth	1
HOCM predictive	0		1 (age 4)
Huntington's disease	0		2 (ages 5, 14)
Kennedy's disease (predictive)	0		1 (infant)
Myotonic dystrophy	13	Infancy-14y	7
Spinal muscular atrophy (SMA), intermediate	1	3mth	0
SMA 3	1	10y	0

Carrier and predictive tests for adrenoleucodystrophy, Wilson's disease, and a few other conditions were deferred for a variety of reasons, sometimes including technical as well as ethical and counselling factors.

BCR = balanced chromosomal rearrangement

BMD = Becker muscular dystrophy

CF = cystic fibrosis

DMD = Duchenne muscular dystrophy

FAP = familial adenomatous polyposis coli

FSH = facioscapulohumeral

HMSN = hereditary motor sensory neuropathy

NTM = normal transmitting male