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Attitudes of subjects at risk and their relatives towards genetic counselling in Huntington's chorea

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SUMMARY Ninety-two patients suffering from Huntington's chorea (HC) and their spouses, and 91 subjects with an affected parent and their spouses, living in three counties of industrial South Wales, have been studied regarding their knowledge of their inheritance of the disorder. Particular attention was paid to its influence on their attitudes towards child-bearing, telling their children of the risks, and predictive tests. Only 12% of the patients were known to have received professional advice before completing their families, in contrast to 68% of the sample at risk. It is estimated that 82% of the patients and 60% of the subjects at risk had, or might have, restricted their family size had they known in time. The majority found genetic counselling helpful, but did not necessarily wish to alter their child-bearing plans in consequence. It was clear that information provided by the family alone was usually inadequate and that this applied to the present generation at risk as well as to previous generations. It was concluded that the burden of telling children the risks is too great for most parents and that professional help is needed. The long term impact of genetic counselling on the incidence of the disease is impossible to assess without continued monitoring, but preliminary results are encouraging.[1] Attitudes towards a predictive test reflected much conflict: although 56% overall wished to take one, only 40% of those who were parents wished to know if they were at risk of passing the gene on to their children. Few subjects reported severe social stress on learning of their genetic risks, but about one in four reported experiencing significant anxiety.

Huntington's chorea (HC) is an autosomal dominant disease with onset usually in adult life. While much research has been devoted to the medical and scientific aspects, little is known about the impact of genetic counselling on affected families and particularly on their attitudes towards child-bearing. The patients' organisation Combat[2] and others[3 4] have drawn attention to the desirability of further research in this area. As the new mutation rate is considered low, because no reliable method yet exists of identifying the asymptomatic gene carrier, and as the only treatment is palliative, the possible role of genetic counselling in controlling the disease appears of considerable importance.

Previous large scale surveys in the United States[5] and the United Kingdom[6] assessed the attitudes of Combat members who could be considered a highly motivated and self-selected group. The study to be described drew on an unselected group of patients and adults at high risk in three counties of industrial South Wales. It formed part of a long term prospective study of the disease aimed at evaluating the effectiveness of a programme of systematic genetic counselling which has been described elsewhere.[1 7 8]

The genetic counselling given was non-directive and carried out on two levels. At the initial survey visit general advice on the genetic risks was provided and the subjects' attitude towards and understanding of the disease assessed. More detailed information was given at the clinic visit and specific queries answered. A system of follow-up visits by an experienced nurse, accepted by 74% of the families directly and by 23% indirectly, was instituted to ensure that the information given was understood, to initiate clinic visits for young people reaching adulthood, and to refer for medical advice subjects showing possible signs of the disease. Contraceptive advice, sterilisation, and termination of pregnancy were available where indicated.

The support offered to the subjects also included other medical and social service elements in an

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attempt to provide comprehensive coverage of the family's needs in relation to the illness.

Aims and methods

The aims were to discover how much knowledge of the hereditary implications had been available to the patients, their spouses, and a random sample of children at high risk and their spouses before they had completed their families, from what sources it had been obtained, and how much influence this knowledge had exerted on their child-bearing practices. Attitudes towards the genetic advice received, telling children the risks, and a predictive test were also assessed, together with some of the other effects on the subject.

The patients were ascertained by means of a prevalence survey and the methods used have been described elsewhere.[8 9] However, it proved unrewarding to interview the patients concerning their knowledge of the genetic implications,[10] and although 77 were or had been married, 26 spouses were not available to be included in the survey. Eleven had already died, one was too ill to be interviewed, and six could not be traced. Genetic advice had been deliberately withheld from the remaining eight spouses, all of whom were elderly, four of whom were childless, and several of whom suffered from serious medical conditions. Thus 51 spouses were interviewed, all of whom had had some information about the hereditary nature of HC, though two doubted the diagnosis. Five had not yet been told that HC had been diagnosed in their partners, but two women were extremely suspicious.

The second group was obtained by drawing a random sample of 100 persons, out of a total population of 236 living in the study area of Gwent, Mid, and South Glamorgan, identified as having an affected parent and being between the ages of 18 and 49 years on 1 January 1977. Nine subjects were excluded: three could not be traced as two had been adopted from an early age and one had run away from home. Six subjects were found to be ignorant of the genetic implications. Two of these were single women, over child-bearing age, who suffered from severe disability. A single man, in his forties, who was considered to be showing early signs of HC, denied all knowledge of the 'family complaint' (though reminded by his brother at the time of the interview). The others, two women and one man, had lost touch with the affected parent at an early age and had no knowledge that he/she had developed HC. Thus, only 91 subjects were able to be questioned; four refused to be interviewed personally but were given a copy of the questionnaire and allowed relatives to speak for them.

Although 77 of the high risk group were or had been married, only 39 spouses were able to be interviewed as, in 30 cases, the subjects refused permission. It is thought that many of these preferred their partners not to be fully cognisant of the risks they ran, particularly where marital disharmony existed. Of the remaining eight spouses, four could not be traced, one was abroad, one had died, and two had received no genetic advice.

The interviews took place during 1977 to 1980, by prior appointment and usually at home, though a few preferred to come to the hospital or to meet the interviewer elsewhere. Geographical mobility did not prove to be an insuperable problem. Three women had moved out of the study area but were able to be visited personally, either in their new home or when they returned to the study area on holiday. Others had moved within the study area, but were able to be traced, several only after persistent enquiries taking up many hours of time. Initial reactions could be hostile, but no subject refused totally to participate in the survey. A patient and sympathetic approach was needed, which acknowledged the subject's anxiety, but stressed the complete confidentiality of the answers and the positive value of the research being undertaken.

Population studied

The social characteristics of the patient group, which consisted of 35 males and 57 females, have already been outlined.[9] The spouses consisted of 27 husbands and 24 wives. The original children at risk sample contained 51 males and 49 females, of whom 47 males and 44 females were interviewed. Table 1 sets out the distribution of their ages, as at 1 January 1977, and year of birth. Five (5%) were in their teens, and 12 (13%) were aged 45 to 49 years, with the largest number, 26 (29%), aged between 25 and 29 years.

Table 2 sets out the civil state of the sample at risk: 14 (15%) were single and 77 (85%) were or had been married, four for the second time.

TABLE 1 *Ages of children at risk by 5-year categories and sex.*

Age (at 1.1.77)	*Children at risk*			
	Males	*Females*	*Total*	*%*
Less than 20 years	2	3	5	5
20–24	6	3	9	10
25–29	11	15	26	29
30–34	6	7	13	14
35–39	5	7	12	13
40–44	8	6	14	15
45–49	9	3	12	13
Total	47	44	91	99

TABLE 1a *Year of birth of children at risk by 5-year categories and sex.*

Year of birth	*Children at risk*		
	Males	*Females*	*Total*
1927–1929	4	2	6
1930–1934	9	5	14
1935–1939	7	6	13
1940–1944	5	9	14
1945–1949	10	10	20
1950–1954	9	9	18
1955–1958	3	3	6
Total	47	44	91

The number of children born to the total patient group, the spouses of patients surveyed, and the subjects at risk, is shown in table 3. In the patient group six children are counted twice, so the true figure is 185. Half the children at risk had had no children as yet or one child only. Ten illegitimate births are known to have occurred in the patient group and four in the children at risk sample. It is possible that the number of births is under-reported, but it is thought that this number is likely to be very small.

The social class distribution is set out in table 4. The majority in both groups were employed as manual workers; among the spouses of patients only nine heads of households and 22 of the group at risk were engaged in non-manual work. Two men at risk held university degrees; none in the patient group did so. Only two women at risk, a nurse and a primary school teacher, had undergone further training after leaving school. The majority had married spouses from a similar background. No subjects actually stated that they had had educational ambitions which were thwarted by the effects of illness in the family, but several women had had their attendance at school interrupted by the demands of the choreic parent.

TABLE 2 *Civil state of children at risk by sex.*

Civil state	*Male*	*Female*	*Total*	*%*
Married	32	35	67	74
Single	11	3	14	15
Divorced	2	4	6	7
Separated	2	1	3	3
Widowed	0	1	1	1
Total	47	44	91	100

TABLE 3 *Number of children born to patients, spouses of patients, and children at risk.*

No of children	*Patient group*		*Spouses*		*Children at risk*	
	(n=92)	*(%)*	*(n=51)*	*(%)*	*(n=91)*	*(%)*
0	22	24	5	10	31	34
1	16	17	11	22	15	16
2	28	30	17	33	21	23
3	8	9	7	14	12	13
4	8	9	4	8	7	8
5	3	3	2	4	4	4
6	3	3	3	6	1	1
7	2	2	2	4	0	0
8	2	2	0	0	0	0
No of children born	191		124		147	

TABLE 4 *Distribution of patients, spouses of patients, and children at risk by social class.*

Social class	*Patients*	*%*	*Spouses of patients*	*%*	*Children at risk*	*%*
1	1	1	1	2	1	1
2	8	9	8	16	13	14
3	49	53	22	43	42	46
4	12	13	9	18	18	20
5	18	20	11	22	17	19
Never worked	4	4	0		0	
Totals	92	100	51	101	91	100

Three subjects at risk, one man and two women (excluding the man who could not be interviewed), were considered to be affected but had not yet had the diagnosis confirmed. Two of these were still employed.

Source and extent of genetic counselling

It was estimated that before the prevalence survey, which was begun in 1974 and completed by 1978, 61 patients (66%) had either had no knowledge of the genetic implications of HC or knowledge from the family only.[7] The reasons for this were sometimes very hard to discover, but appeared to relate to such factors as: severing of family links at an early age; the diagnosis in the previous generation not being made in life; deliberate concealment by the family; and a reluctance on the part of the medical profession to reveal the diagnosis and its implications, a problem already noted by Caro[11] and Barette and Marsden.[6] As a result of the prevalence survey many of these families learned fully about the hereditary nature of the disease for the first time.

The first source and all sources of genetic advice for the two groups are set out in table 5. As it was the practice of the survey team (once the diagnosis had been established) to ask a responsible family member to inform other members at risk before making further contact, the family is noted to be the biggest purveyor of information. However, it emerged clearly that this information is likely to be inadequate: the extent of the risk was rarely made explicit, nor was the nature of the transmission clearly understood. Advice from professional sources could also be inaccurate. Very many subjects commented that not until the survey team visited did they fully understand the genetic implications. Of the group who had not been approached directly by the survey team, 28 (68%) showed a lack of understanding and nine believed it was inevitable that they, or their children, would develop HC.

TABLE 5 *Sources of genetic advice for spouses of patients and children at risk.*

Source	First source				All sources			
	Spouses of patients (n=51)	%	*Children at risk (n=91)*	%	*Spouses of patients (n=51)*	%	*Children at risk (n=91)*	%
Family	16	31	66	73	19	37	70	77
Hospital personnel	17	33	7	8	19	37	19	21
General practitioner	7	14	6	7	10	20	15	16
Survey team	9	18	5	5	50	54	50	55
Media	2	4	5	5	13	25	22	24
Other	0	0	2	2	0	0	2	2

Information provided by the 'media' was generally perceived as helpful and was responsible for seven families seeking further advice. Five of these only knew the name of the disease from seeing it on a parent's death certificate.

'Other' source referred to two subjects who learned the nature of their parents' illness from casual remarks by a visitor, who happened to be a nurse.

There is some evidence from other studies[12 13] that genetic advice is often poorly understood and remembered and members of these HC families showed a clear need to discuss the information several times and in different ways.[6]

Influence of genetic advice on attitudes towards child-bearing

Only 12 of the spouses of patients had received information about the complaint before their families were complete and 11 said that they had been influenced. Eight of these had already delayed having children or further children because of the symptoms noted, and knowledge of the diagnosis confirmed their decision. Four spouses were married to patients who had been unaffected at the time of counselling: two decided to 'risk' one child each; one decided to limit his family to two; and the other would have had his chosen family size, but his wife only conceived once.

By contrast 62 (68%) of the group at risk had received genetic advice, directly or indirectly, from a professional source before their families were complete, and 29 (32%) had only learned of the hereditary implications after completing their families. Of the group whose families were incomplete, 48 were married and 14 were single. Table 6 sets out the numbers of children who had already been born to these 48 married couples at the time of the study, divided into those who said they had been influenced and those who said they had not been influenced.

In the influenced group, seven of those who had no children said that they will continue to have none and 11 decided to have no further children. Five had children or further children but most restricted their family size to one or two (three of these births were said to be 'accidental'). Four were uncertain about further child-bearing but said their family size would be limited.

In the uninfluenced group, eight had children after receiving genetic advice and now said their families were complete. Ten were actively planning children (three wives were pregnant) and three were uncertain;

TABLE 6 *Number of children already born to the married children at risk, whose families were incomplete at the time of genetic counselling, divided into 'influenced' (A), and 'not influenced' (B) categories.*

No of children	*A*	*B*	*Totals*
0	9	6	15
1	7	5	12
2	9	4	13
3	1	3	4
4	1	1	2
5	0	2	2
Total	27	21	48
Total No of children	32	36	
Mean family size	1·19	1·71	

TABLE 7 *Number of children born to couples at risk who had completed their families before (A) and after (B) knowing about the hereditary nature of HC, and to those who were childless and under the age of 30 years at the time of counselling (C).*

No of children	*Couples at risk*		
	A (n=29)	*B (n=31)*	*C (n=25)*
0	3	7	16
1	2	8	4
2	8	10	3
3	8	2	2
4	5	2	0
5	2	2	0
6	1	0	0
No of children born	78	52	16
Mean	2·68	1·67	0·64

however, most of these said they did not plan to have large families for economic reasons.

Thus, at the time of the interview, a further 31 couples said they had completed their families, making 60 in all, and table 7 sets out the distribution of births into the two groups, those who had received counselling after and before completion of their families. Three couples were childless for reasons unconnected with HC. Table 8 sets out the distribution of births into the two groups combined, and the 17 subjects who intended to have further children or were uncertain.

It was estimated that 25 of the married subjects had been counselled under the age of 30 years, before they had started their families, though their spouses may not have been. The fertility of this group is set out in table 7. Six are actively trying to have children or further children and six are uncertain. A few have embarked on a second relationship and wish to make their new partners 'the gift of a child'. Others want a child of a particular sex. Thirteen have completed their families.

The 'influenced' group contained three subjects who planned to marry: two men said they would probably have only small sized families and one woman, who was marrying her cousin, also at high risk, would have no children. One single pregnant woman did not plan further children. Two men were waiting to see if they developed symptoms before they decided on marriage and parenthood and one man (in his thirties) was uncertain whether to marry at all. He was the only subject who stated that his girlfriend broke off the relationship when told about HC.

The uninfluenced group contained four men who definitely planned to marry and have children, but thought their size of family would be determined by economic factors and not the genetic implications. One single woman with one child planned to have no more and two men (both aged over 35 years) planned to remain single, apparently for reasons other than the risks they ran.

Twenty-nine couples at risk had completed their families before knowing about HC. If they had known earlier 12 (41%) said they would have restricted their family size in consequence (two spouses did not agree and would have wanted the same sized family). Eight (28%) would not have altered their family size (including two men who had no children because they had married wives who already had large families) and nine (31%) were uncertain, but thought that they would probably have restricted their family size. Table 8 sets out the number of children born to this group. All but two subjects had only known about the hereditary implications after the age of 29 years (table 9) and it is estimated that, had they known earlier, approximately 28 births might have been prevented.

Thus, in the group at risk overall, 34 persons (37%) said that they had been influenced and 21 persons (23%) that they could have been. The number of spouses of patients who said they would have restricted their family size had they known earlier was 31 (61%) and, as has been noted, 11 spouses (22%) knew before their families were complete and altered their child-bearing plans in consequence. Thus 42 (82%) of the spouses of patients were, or said they would have been, influenced compared with 55 (60%) of the group at risk.

The Stern and Eldridge US study[5] found that 86% of their respondents would modify their family size if at risk for HC and that "desire for limitation was greatest among those affected but lowest among young adults at high risk", confirmed by this study.

Ten percent of spouses of Welsh patients would have had the same sized family even if they had known earlier. This figure is higher than the 2% in the US study[5] and the 6% in the UK study[6] who would not have been influenced even if they had known their partners carried the gene. This attitude

TABLE 8 *Number of children already born to married couples at risk, divided into those who had completed their families (A) and those who had not completed their families (B) at time of study.*

No of children	*A*	*B*	*Totals*
0	10	8	18
1	10	4	14
2	18	3	21
3	10	2	12
4	7	0	7
5	4	0	4
6	1	0	1
Total	60	17	77
Total No of children	130	16	
Mean family size	2·17	0·94	

TABLE 9 *Age of knowing the hereditary implications for children at risk who had completed their families before knowing about HC, by sex and 5-year categories.*

Age	*Female*	*Male*	*Totals*
>20	0	0	0
20–24	1	0	1
25–29	0	1	1
30–34	5	2	7
35–39	4	3	7
40–44	5	3	8
45–49	1	4	5
Total	16	13	29

was often justified by remarks such as "Children are worth the risk" and "There will be a cure by the time they grow up".

Both Pearson[3] and Wexler [14] have discussed the fact that many HC families continue to bear children in full knowledge of the genetic risks. Pearson considers that the key to this phenomenon lies in the way subjects handle the anxiety provoked and comments "Perhaps the human drive for procreation springs from a well so deep that it will always override and take control of human intellect, bending all reason and argument into rationalization through the processes of projection and denial". Wexler stresses "the symbolic and magical significance of the child as an insurance of the parents' continuing health". All these reactions were apparent in this study. Subjects could believe that they had knowledge, specific to their family, which enabled them to be certain that they did not carry the gene (though their sibs might) and thus they could have children without fear, for example, "Only women in my family get it, so I, being a man, will not". Others said that they were glad that their parents had borne them, and could not 'punish' their parents by not having children themselves. There was also a strong feeling in some families that they had suffered enough, without having to endure the further deprivation of not having children.

Use of birth control methods

Five female patients had already been sterilised, one more sterilisation was planned, and one patient had had a hysterectomy. Three wives of patients had been sterilised and one wife had had a hysterectomy. One male patient and two husbands of patients had had vasectomies and one more vasectomy was planned. Two pregnancies were known to have been terminated: one woman, before the survey, was apparently refused a termination because of a lack of family history. No further births were expected in this group and none subsequently has occurred.

In the children at risk group, 10 subjects and three spouses had undergone vasectomies and two more were planned in a subject and a spouse. Ten subjects and two spouses had been sterilised and two subjects had had hysterectomies. Thus, overall 22 subjects and five spouses were not able to bear further children: 23 operations were said to be directly attributable to the knowledge of HC. Three pregnancies were known to have been terminated. Twenty-five subjects were using artifical methods of contraception, four were trusting to other methods, and one woman failed to reply to the question. Twenty-two subjects were not using any form of birth control because they were considered either to be over child-bearing age, or to be naturally infertile, or not to have a partner.

Attitudes towards genetic advice

In general attitudes towards genetic advice were positive (table 10). Few (8%) wished definitely not to have known at all, but a significant number (26%) of children at risk and their spouses were uncertain. Spouses of children at risk recorded 16 discordant answers. Single persons at risk recorded the highest percentage (57%) of those who either did not want to know at all or were uncertain.

Wanting to know earlier could be for several different reasons: the most important ones seemed to be the opportunity to make an informed decision about family size and the feeling that the subject's relationship with the patient would have been improved had it been known that the latter was ill. Some reasons given were unexpected. A few wives who had violent husbands wished they had had no children, because without them they could have left their husbands when their behaviour deteriorated. "You can't do it when you have children. No one wants you".

Subjects who were glad that they had received genetic advice at the age they did could hold two opposing attitudes. Some subjects were glad that they had known in time to restrict their family size. Others were glad that they had known after they had had their children and thus were spared the dilemma of making a decision. They might not have had their children but "could not imagine their lives without them".

Not wishing to know at all and being uncertain could reflect resentment and also a high level of anxiety. Some subjects at risk commented that they could not bear the thought of their families seeing them in the grip of HC. Others felt caught in a conflict, exemplified by the wife of a patient who said that she felt she should have known earlier for the sake of her children. On the other hand, had she known her husband was ill, she might have felt obliged to stay with him and she could not imagine how she could have endured his violent behaviour.

Two subjects at risk felt certain that, had they known earlier, they would not have married at all. A few spouses doubted if they would have married their partners and a few children at risk doubted if their partners would have married them.

Most persons (57%) perceived genetic counselling as helpful (table 11). However, desire to make an informed decision about family size was by no means the only reason why subjects wished to know the truth (as has already been indicated) nor necessarily the only reason when they did. Other factors could

TABLE 10 *When spouses of patients, children at risk (by civil state), and spouses of children at risk wished to receive genetic advice.*

	Spouses of patients	%	*Children at risk*				*Spouses of children at risk*	%	*Totals*	%
			Married	%	*Single*	%				
Earlier	38	74	26	34	2	14	14	36	80	44
Same age	4	8	30	39	4	29	10	26	48	27
Later	0	0	0	0	0	0	0	0	0	0
Not at all	5	10	4	5	3	21	3	7	15	8
Uncertain	4	8	17	22	5	36	12	31	38	21
Total	51		77		14		39		181	

TABLE 11 *Attitudes towards genetic counselling of spouses of patients, children at risk, and their spouses.*

	Spouses of patients	%	*Children at risk*	%	*Spouses of children at risk*	%	*Totals*	%
Helpful	27	53	55	61	21	54	103	57
Not helpful	6	12	15	16	7	18	28	16
Worrying	14	27	14	15	9	23	37	20
Uncertain	4	8	7	8	2	5	13	7
Total	51		91		39		181	

be felt to be as important or more important, and persons who were glad to have been informed of their genetic risks did not necessarily modify their family size in consequence. Subjects often stated that "they had a right to know these things" and welcomed the opportunity to participate in a research project which gave them the opportunity to "fight" HC. They were also often relieved to hear of a Unit specialising in HC which could offer help.

Reference was often made to the way in which genetic counselling was given. Those who had a positive attitude were relieved to have the opportunity to "talk the issues through". A strongly directive approach, particularly if it was non-supportive, was almost universally resented. One woman (in her forties) was apparently told by a consultant physician that her children should not have children, though there was no evidence that she was affected. She felt that no consideration had been given to her feelings of shock and dismay and no help offered in caring for her affected mother. She determined to avoid doctors in future and to tell her children nothing.

A single girl described her experience of being told the nature of the mother's illness (by a consultant physician) as "feeling as though a bomb had fallen on top of me". She was convinced that she would develop the disease very shortly. She was also told, apparently, not to have children which was her dearest wish. It is perhaps not surprising that she became pregnant 3 months later. As noted by Stern and Eldridge[5] a supportive approach was clearly preferred.

Should children be told the risks?

Of those questioned, 127 (70%) felt that children should be told the risks (table 12). The spouses of patients were the most certain and the spouses of children at risk were the least certain. Nine subjects at risk were uncertain because they had no children and "it all depends on the child". Although there was also a widespread feeling that parents are the best people to inform their children, because "other people might frighten them", only 17 (37%) of the spouses of patients who were parents had actually told their children, but 32 (70%) said their children knew. Of the children at risk seven (31%) who were parents had told their children, but 21 (38%) were said to know (table 13).

There was a widespread feeling that children should not be told until they were "old enough to understand" and that often this will not be until they are contemplating marriage. However, in 14 families (26%) the children were said to have been told before the age of 19 years (table 14) because their parents believed it was best to "grow up naturally" with this information. The children at risk, numbering 21, who had themselves been counselled in their teens felt the same. "It doesn't come as such a shock", and none of these wished to know at any other age. This attitude may be indicative of a supportive and confident parental relationship. However, several others, who had been told at a later age, commented

TABLE 12 *Attitude of spouses of patients, children at risk, and their spouses towards whether children should be told the genetic risks of HC.*

	Spouses of patients	%	*Children at risk*	%	*Spouses of children at risk*	%	*Totals*	%
Yes	38	74	66	72	23	59	127	70
No	1	3	6	7	6	15	13	7
Uncertain	12	23	19	21	10	26	41	23
Total	51		91		39		181	

TABLE 13 *Whether the spouses of patients and the children at risk had told their children about the genetic risks of HC.*

	Have you told your children?						*Do your children know?*					
	Spouses of patients (n=46)	%	*Children at risk (n=55)*	%	*Totals (n=101)*	%	*Spouses of patients (n=46)*	%	*Children at risk (n=55)*	%	*Total (n=101)*	%
Yes	17	37	17	31	34	34	32	70	21	38	53	52
No	29	63	38	69	67	66	14	30	34	62	48	48

TABLE 14 *Age of knowing about the hereditary implications of HC for children of patients and subjects at risk.*

Age of child	*Spouses of patients*		*Subjects at risk*		*Totals*	%
	No of families	%	*No of families*	%		
>14 years	8	25	4	19	12	23
15–19	5	16	9	43	14	26
20+	8	25	1	5	9	17
Varied	9	28	4	19	13	25
Uncertain what age	2	6	3	14	5	9
Total	32	100	21	100	53	100

that in their teens, they "would not have taken in the information". One man was glad that he only knew after finishing his apprenticeship. Had he known before, he would have thought to himself "what's the use?".

Although such a large number of parents felt children should be told the risks, there could be many reservations underlying these answers. Many of the group at risk did not intend telling their children the full story unless they themselves became affected and had told them, specifically, not to worry. A few subjects said their children must know their risks as they "see the disease all around them, but I am not going to discuss the matter with them and frighten them".

It was clear that many parents at risk intended to perpetuate the pattern of providing their children with either no information or inadequate information. The burden of telling children the risks was too great for most people and professional help was needed. Barette and Marsden[6] came to the same conclusion.

Predictive tests

Attitudes towards a predictive test could be ambivalent: 51 (56%) of the group at risk overall would wish to take one if it existed, compared with 22 (40%) of those who were parents, who wished to know if they were at risk of passing on the disease to their children (table 15).

Positive answers were often given with a considerable amount of misgiving. Many subjects said they would dread being told they carried the gene but thought they should submit to it for their children's sake and in order to make financial provision for their family. Others who believed that they had found a way of "fighting off" HC wanted to know if their methods had worked. A few wanted the status of an 'ill' person, apparently hoping that the test would be positive, in order to receive better treatment from their families and the community at large. Several subjects appeared quite confident that a test would show that they did not carry the gene. One or two others found the uncertainty in which they lived

TABLE 15 *Attitudes of total group and those who were parents only of children at risk towards predictive tests.*

	Do you want a predictive test?						*Do you want to know if you are at risk of passing the disease on to your children?*					
	Total group (n=91)						*Parents only (n=55)*					
	Male	*%*	*Female*	*%*	*Totals*	*%*	*Male*	*%*	*Female*	*%*	*Totals*	*%*
Yes	27	57	24	55	51	56	10	38	12	41	22	40
No	8	17	7	16	15	16	8	31	7	24	15	27
Don't know	12	26	13	30	25	27	8	31	10	34	18	33
Total	47		44		91		26		29		55	

quite intolerable and felt that even knowing they would develop the disease would be preferable.

Those who did not want to know were often worried about the effect of HC, should they develop it, on their families. A typical comment was "I don't want to know because, if I thought I was going the same way as my mother, I'd kill myself. I couldn't bear my family to see me like that".

Those who were uncertain felt in a dilemma. "I feel I should have a positive test because of my children, but if I am going to develop the disease, I don't want to know. What a burden to place on my husband".

In the two studies which assessed the attitudes of Combat members, the proportions of those wishing to take a predictive test were higher, 77% of respondents with an affected parent in the US study[5] and 80% of the sample in the UK.[6] In Australia, Teltscher and Polgar,[15] taking a random sample from their Huntington's Disease Register, reported that 84% of their subjects were found to have a positive attitude towards such tests.

In a recent review Thomas[16] states that the medical profession is deeply divided on the issue of whether a reliable predictive test should be used before an effective treatment is developed. On present evidence there would be a significant demand from subjects at risk and a heavy burden would be placed on medical staff who would have the responsibility for informing subjects of the results, should it be used. The experience of this study indicates that there would also be considerable need for counselling, both before and after testing. It is suggested that pilot studies will be needed to determine the range and extent of supporting services required and to evolve generally acceptable guidelines.

The experience of being at risk

Published reports on the way subjects at risk handle their anxiety are sparse. On the basis of in-depth interviews with 34 subjects at risk, Wexler[14] has written that the years in anticipation (of developing Huntington's disease) can be "years of dread, of silent apprehension, of noisy emotional disarray, or of intense productivity". Pearson,[3] on the basis of an intimate knowledge of a small self-selected group of 13 persons, and a good reported knowledge of 21 others, considered that 40% of his subjects at one time or another manifested an anxiety neurosis which could be reactivated at crisis points, and that spouses were similarly affected. In this study, 20 (23%) were considered to be affected by anxiety to a significant degree. All subjects, except one woman who had received genetic counselling in the previous 6 months, had known about their high risk status for at least 2 years and a few others for much longer.

Eight subjects (three men and five women) reported experiencing continuous severe anxiety but only one man had been admitted to hospital for depression and was receiving psychiatric treatment. Seven of these attributed their anxieties primarily to their experiences with the choreic parent and the knowledge that they were at risk exacerbated their symptoms. Several described their marriages as unstable and two men drank heavily. The remaining woman had been unaware of the family complaint until the diagnosis was made in her sib.

Ten subjects (eight women and two men) rated themselves as suffering from relatively mild chronic anxiety. They considered they were managing to lead relatively normal lives but became easily depressed and often looked for signs and symptoms in themselves. Supportive spouses were of immense importance to them.

Two men were rated as anxious by their families but not by themselves. One man had suffered a broken marriage and was said to be moody, periodically violent, and to drink heavily. The other was a solitary person, unable to keep employment.

The remaining 71 subjects, which included the three probably affected persons, reported intermittent worry, in varying degrees, or no worry at all. They felt that their lives were not being significantly affected and that, if worry was felt, they were able to keep it at bay by various devices, based on their experience of HC in their family. Many led very active lives, thus proving to themselves and others that they were not showing signs of HC. A few considered that they lived more positively as a

result of knowing that they were at risk. Others believed in the avoidance of stress and worry, even to the extent of studiously avoiding affected relatives. One grossly obese woman, though suffering from a serious heart condition, refused to diet because her affected mother was very thin.

Only one spouse reported feeling continuously depressed since hearing about the risk to her husband, but was not receiving any medical treatment. Six spouses (four wives and two husbands) were receiving long term psychiatric treatment for anxiety and depression but none attributed their symptoms to their knowledge of HC.

General discussion

Our survey of HC in South Wales was responsible for enlightening many HC families about the genetic implications of the disease. The long term impact of this knowledge on their lives, and particularly on their attitudes towards child-bearing, is not easy to assess at this stage. To allow most subjects at risk to make an informed decision about family size, genetic counselling must be offered early, when they are in their teens and twenties, and when the majority are asymptomatic. It has been suggested[6] that those who are influenced sufficiently to limit their family size may be the very persons who do not carry the gene. The very small evidence from this study does not seem to support this view. It is also possible that, with the progression of the disease, the choreic may 'carelessly' produce more children than originally intended: early identification and the provision of birth control methods, where acceptable, may reduce this hazard.

Carter and Evans[17] and Carter *et al*[18] have shown that when informed of their risk before starting their families, the fertility of children with an affected parent is greatly reduced. A previous study, carried out by the authors, revealed that 52 adults at high risk said that they intended to have fewer children as a result of counselling, compared to 25 who stated that they had not been so influenced.[1] While these surveys give hope, only continued monitoring can test the hypothesis that a programme of systematic genetic counselling of all adults at high risk may result in a reduction of the incidence of HC. However, it seems clear that long term follow-up and family support are essential ingredients of such a programme if it is to prove successful.

Although many parents were anxious to shield their children from the knowledge of their genetic risks, it was clear that a significant number of subjects wished to know and positive benefits, as well as negative results, could ensue. While a lack of self-confidence was a common feeling among the group at risk, surprisingly few strongly adverse reactions were attributed to genetic counselling, provided it was supportive, and these were mainly reported by persons already made vulnerable by their past experience of the disease. The question of how best to counsel children at risk, particularly adolescents, awaits detailed studies.

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