50/50 The Dilemma of Genetic Counselling in Huntington's Disease

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WHAT IS HUNTINGTON'S DISEASE?

Huntington's Disease is a hereditary disease which is passed from generation to generation through auto-somal dominant transmission. The transmission is unrelated to the sex of the child and each child of an affected parent has a 50% chance of inheriting the disease or not inheriting the disease. This disease never skips a generation but for those who do not inherit the offending gene (S), then the disease can no longer be transmitted to future generations. The disease usually manifests itself in the late 30's and early 40's years of age, that is, towards the end of the reproductive life of the patient. The average length of time from onset to death is about 15 years and death is usually due to pneumonia or heart disease in a debilitated person.

Manifested

The disease is manifested in two major ways. It could show itself in intellectual deterioration in which the patient's memory begins to be impaired. There may be degrees of irritability and loss of attention and concentration in the early stages. Abnormal jerky involuntary movements develop and the patient frequently falls or drops articles. As the disease progresses, there is slurring of speech, a jerky and incoordinated gait, facial grimacing and difficulty in swallowing. Invariably the patient will deteriorate, lose weight and progress towards death. At present there is no specific treatment for Huntington's Disease although pharmacological means are available to act as palliative measures for the symptoms of abnormal movements and some of the behavioural symptoms associated with Huntington's Disease. The major management thrusts of Huntington's Disease lie in the social and emotional support that professionals can give to the patients and their families. Genetic counselling as well as general counselling are part and parcel of the management of the patients and their families.

GENERAL COUNSELLING:

The general counselling of patients and their families, can be for convenience sake, divided into several stages.

1) Pre-symptomatic counselling — frequently people at risk of Huntington's Disease present to the clinic seeking confirmation that they have or have not the disease. Frequently this is motivated by family pressure and also by considerable anxiety about the "at-riskness". The diagnosis of Huntington's Disease is at present basically a clinical one and in the clinical situation the patient could be readily assured that at that point of time he may not be suffering from Huntington's Disease. The anxieties, fears, apprehensions and the significance in practical terms of this sword of Damocles hanging over these people, require some working through. It is necessary at this stage to assess the degree of family cohesion and the degree of mutual support between the person at risk and the spouse and other members of the family. In the future, if this person develops Huntington's Disease, then the strength of the family will have to be called upon and enhanced to support the affected patient.

2) Apart from counselling in emotional and psychological aspects, frequently counselling in terms of the practical management of their lives, must be instituted at this stage. Such practical matters as life assurance, superannuation, health insurance covers and other legal and social arrangements must be considered.

3) Frequently, the question of whether the children should or should not be told demands a great deal of soul searching. Very often the decision to tell or withhold information from young children is dependent on the degree of maturity of the children and the personal strengths and weaknesses of the parents concerned. There is no hard and fast rule in this aspect of counselling.

4) Material is beginning to become available to assist parents to learn as much as they can of accurate and up-to-date information about the disease so that they may pass on information and counsel their children accordingly.

   In addition to the written literature that is available from the Huntington's Disease Clinic, a copy of the A.B.C. documentary, "Something in the Family", is also available from the Clinic for showing to groups of parents who have not yet seen this videotape and forms the basis for group discussion to facilitate counselling in this situation.

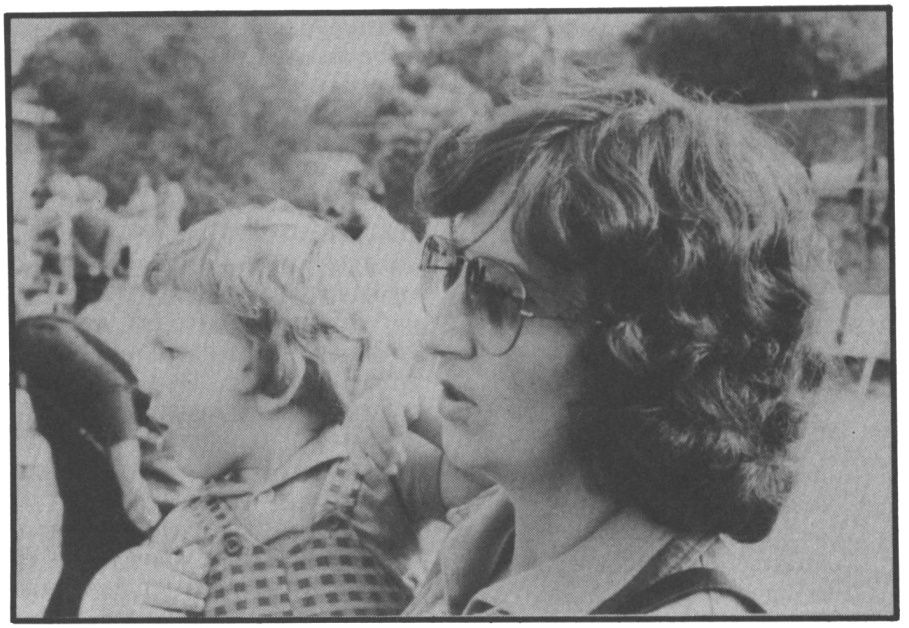
GENETIC COUNSELLING

The principle we adopt in genetic counselling of Huntington's Disease families is that of a non-directive approach. The basis on which decision making in child bearing has to be assessed first. It is absolutely necessary to discover what factual information a couple possess and what interpretation they place on these data before any step in counselling can take place. The couple must have a clear understanding of the known facts of the disease and the known facts relating to the transmission and risk factors involved. The couple must be allowed to express all their fears and apprehensions and anxieties regarding these facts. Any interpretation of the facts from the couple must be considered as genuine and must be dealt with appropriately. Without a proper provision of known facts of the disease and understanding of the proper interpretation of these genetic and social factors, counselling cannot really proceed without creating misunderstanding, misinterpretation; and decisions could be made which are not appropriate to the couple concerned. Questions frequently asked of us relate to the nature of genetic transmission, the possibility of a cure for the disease, a possibility of successful treatment being available within a generation and the possibilities of options available. The possible availability of predictive tests is a question to understand that at present there is no cure for the disease and there are no reliable predictive tests for people at risk. Treatment is available but only palliative and not curative. The symptoms of abnormal movements and certain psychological disturbances relating to intellectual impairment could be treated pharmacologically. Emotional disturbances associated with the disease and its implications can also be successfully handled by appropriate supportive psychotherapy. There is no place for raising hopes regarding the future of the disease although one must remain optimistic that the day will come when more specific treatment or even a cure will be available; and certainly the day could come in our generation that predictive tests could become available to define those who will develop the disease from those who will not develop the disease.

Necessary

It is also necessary to assess the philosophic and personal determinants of the couple. The personal philosophy and expectations of what life has to offer to these couples, must play a very important part in the situation of genetic counselling. The strengths of these couples must be clearly elicited so that they may be utilised in their decision making process. Their weaknesses must also be defined and steps taken so that these weaknesses do not interfere with proper decision making. Keeping in mind that the decision to have children is primarily an emotional decision rather than a scientific or rational act and that this decision is subject to all kinds of pressures from relatives, peers and society at large, then it is necessary that the couples being counselled be made aware of this fact. Each couple must be made aware of such forces playing upon them, whether these forces are co-ercing them to have children, or not to have children. The next step in the counselling would involve the couple expressing their view point regarding child-bearing. In this situation it is necessary to create an atmosphere of acceptance so that each couple will be able to discuss it freely without fear of "judgement". Frequently we encourage the couple to conduct a dialogue between themselves to toss the "pros" and "cons" to each other with the implicit atmosphere that decision making in the counselling situation is not necessary. It is given to understand by these couples that the decision does not have to be made immediately and that the decision making process must be gradual, with a great deal of thought and discussion between themselves. The genetic counsellor must remain impartial and neutral in this situation.

The discussion at the same time does not enforce any preconceived values on the couple. It is absolutely necessary to keep in mind that the decision must be made by the couples concerned without pressure being exerted from the counsellor. It is also necessary for the counsellor to specifically support whatever decision that the couple would make. It has been our practice to inform the couple that whatever decision they come to, there is no right or wrong in the situation, only that each decision must be seen to be appropriate, having taken consideration of all circumstances and all forces that impinge upon their lives. The decision they make, as far as we are concerned, is a correct one for them at that particular time and that the clinic is quite prepared to accept and support whatever decisions they make. This promise of continuing support we see as very important in the counselling situation. The agony of decision making in this 50-50 risk situation is tremendous and it is absolutely imperative that counsellors must be available and seen to be available to back the decision which ever way they turn. It can be anticipated that with either decision there will be problems in the future and the counsellor must be aware of this and be available to support the couple in their decision.



A 50/50 risk.

ALTERNATIVES TO HAVING CHILDREN

It is necessary to keep in mind the available alternatives to having children. At present for a female at risk, the only possible alternatives are adoption and fostering. The list of parents awaiting children for adoption is extremely lengthy and the waiting period can be very long. With a history of Huntington's Disease in the family, it is unlikely that these couples will be provided with children for adoption. It may be possible that they could foster children until such a time as the risk period is largely over or when the disease occurs. If the husband is at risk, then artificial insemination is at present an option available. However, if the wife is at risk then this option no longer applies.

THE FUTURE

What then of the future in the genetic counselling of couples at risk? It is very likely that within the next two decades some predictive tests would be available for a better prediction of "at-riskness" than currently exists. The most promising predictive tests would seem to lie in the direction of linkage study of genetic markers. The possibility of identifying abnormality in a chromosome at present is not high but with the development of further technology, this method of prediction may be possible. The third area of predictive tests research is in the area of fibroblast cell cultures. This method of study is in its early stages and the possibility of this being a predictive test has yet to be substantiated. Should a pre-symptomatic detection method be available, the whole field of genetic counselling in Huntington's Disease would change dramatically. It may be possible to make pre-natal predictions in fetuses. If this is possible then intra-uterine prediction and subsequent offer of therapeutic abortion would be available to the couple concerned with the possibility of a later pregnancy producing a fetus free of the Huntington's disease risk. For those people whose test is negative they will be able to continue to have children in the usual way, freed from this Sword of Damocles hanging over their heads.

The availability of such options would greatly change the nature of the dilemma of genetic counselling in Huntington's Disease.

JOURNALISM AWARD

ANNOUNCEMENT AT MELBOURNE DINNER

The Editor of Australian Child and Family Welfare, Mr Cliff Picton, announced the First Annual Award to Journalism at a Journal Dinner held at the 'Naughty Nineties' Music Hall on October 17th, 1978.

Representatives of Melbourne daily papers and the Journalists' Association were present at the function.

The competition aims to acknowledge the work of Journalists who contribute to the community awareness of the needs of children and families in Australia.

The Competition

1. A cash prize of $750 is offered to that journalist, who, in the opinion of the judges, achieves the highest standard of understanding and journalistic ability on an issue relevant to the welfare of children and families in Australia.
2. Contributions may include both news and feature stories dealing with such things as maltreatment, parent effectiveness, welfare services, or any other issues which pertain to the well-being of children and families.
3. The entry must have been written and published within Australia but does not include articles which appear in professional journals.
4. The award is open only to members of the Australian Journalists Association, but each entry must be the work of a single author.

The Rules

1. All entries must have been published during the year July 1, 1978 — June 30, 1979.
2. The closing date for all entries is August 31st, 1979.
3. Accompanying each entry there should be —
   (a) One tear sheet of the material in published form.
   (b) A statement of approximately 100 words indicating the purpose and circumstance under which the article was written.
   (c) A signed declaration of the article's originality.
4. There exists a limit of two entries per author.
5. Awards will be announced in October 1979 and will be presented at the Annual Dinner of Australian Child and Family Welfare.
6. Entries will only be returned after judging if requested.

The Judging

Entries will be judged on the basis of sensitivity to the subject matter, significance, quality, accuracy, and community impact.

The judging panel will include representatives of both the A.J.A. and the Australian Child and Family Welfare journal and an independent appointment.

The judges' decision will be final.