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Genetic Counseling

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Almost ten years ago I published my first paper dealing with genetic counseling.[1] The following words of Christ as reported by St. John were used to point out a Christian attitude toward the handicapped. In replying to the Apostles' question about the man born blind, Christ said, "Neither he nor his parents were guilty; it was so that God's action might declare itself in him" (John 9:3). In successive papers this attitude has been maintained in other words and with other arguments.[2,3,4] There seems to be no question as to the Christian attitude toward the physically and mentally handicapped. Another Christ and a sharer in the Kingdom more certainly than you or I has a definite demand on our love and consideration. An excellent discourse on this subject is Stanley Hauerwas' "The Christian Society and the Weak: A Meditation on the Care of the Retarded."[5]

There is, however, a real difference between the weak whom we have with us and the knowing procreation of children who have a high risk of congenital abnormality. Responsible parenthood is a subject which has received much attention from all quarters and definitely has caught the public imagination centuries after becoming an important consideration for married couples and for individuals contemplating marriage. Responsible parenthood today in many minds is caught up in financial, social and even environmental considerations but there has been an exponential growth in feelings of responsibility concerning the future health of children yet to be conceived.

This concern for health arises from several sources. Certainly every parent would choose to have healthy, mentally alert children rather than handicapped and/or retarded children, since well being in the physical sense is in itself better than non-well being. Almost all parents feel a deep responsibility to give their children the greatest possible chance of being born well (prenatal care, good obstetrical management) and of having the opportunity for optimal development (pediatric care, parental attention, good schooling). Those relatively rare individuals and couples with a high risk of producing children with serious defects do, and quite rightly, feel a responsibility not to thoughtlessly procreate handicapped children. Other reasons somewhat less weighty include the parental self-image, the extra demands on time and money which the handicapped present, and fear of social disapprobation for the family with even one or certainly more than one affected child. With limitation in family size comes a greater demand that the one, two or three children whom a couple is going to have be well. When couples did not control family size with "scientific methods," having a child was considered providential and having a healthy child a great gift. The handicapped child was also providential and accepted and loved as a mysterious gift of God. Today child bearing and Divine Providence seem, in the mind of the couple, to have been separated by the mechanics of control and the will of the controller.

Medical Speciality

In the milieu where all children are wanted and where there is a growing fund of genetic knowledge and public awareness, the medical, or more properly, the paramedical speciality of genetic counseling has emerged. Genetic counseling has three dimensions: diagnosis, education, and decision. The first dimension, diagnosis, belongs to the medical realm and includes the somewhat sophisticated genetic diagnostic techniques of the biochemistry and cytogenetic laboratories and the skilled geneticist. The second, education, might be done by an array of types from medical doctor and/or geneticist through specially trained nurse or social worker to a bachelor degree level genetic educator. The final component, decision, can only be left to the couple. Infringement by any of the aforementioned personnel on the final decision making is, I believe, immoral. That the final decision to continue a pregnancy, to beget a child or to marry this individual will be made in the light of the information acquired during genetic counseling is obvious, but it should also be obvious that this rather narrowly confined information is not the only basis for decision. Should not a couple include the moral, ethical, theological and social dimensions? The genetic counselor should not direct the decision to what he believes this couple should do, regardless of how altruistic he is. To take away the freedom of the couple and to supercede its values and awareness of what is morally acceptable and substitute the values and morality of the counselor is the worse type of elitism.

Three categories of patients are referred to our service:

1. Those with a diagnosed condition in themselves, their children, or in a family member. The question is specific: "What is the chance that my/our child will have . . . ?"

2. Those who have an undiagnosed or undiagnosable condition in the family, such as repeated miscarriage, retardation of unknown etiology, etc. Included in this group would be those patients or relatives of patients who have had cytogenetic studies or in whom such studies are indicated. Often genetic studies in these individuals lead to a diagnosis of the condition under question.

3. A new category has been generated by popular writing in Sunday supplements, women's magazines and the media in general. This group is comprised of couples contemplating parenthood who want to be sure that they do not have a greater than usual chance of problems with their as yet, unconceived children. They have no particular question in mind, at least at the outset. This category is least productive of results beyond the normal risk figures for all newborns and consumes almost as much time as individuals in either of the previous two categories. We fear that this group may grow in numbers and will necessitate some modification in our procedure.

In each case all pertinent medical data is collected. Diagnosis where available is verified. The couples then are brought to the office and a personal medical history is taken and as complete a pedigree as possible, at least through second cousins. Pedigree studies are essential in order to establish the hereditary pattern of the condition in question and also to uncover any other genetic disease in the family which may be present. Often this information leads to further studies, cytogenetic or biochemical, and lends insight into an as yet undiagnosed condition which has led to definite diagnosis. While pedigree studies are essential for giving the risk figures for the disease in question and for other congenital abnormalities, it also serves another function which is essential to the counseling function.

Guilt Present

Having had an affected child or having a hereditary disease condition in the family is often guilt producing. In order to counsel, one must be able to estimate what the psychological factors are in this individual or couple. Family information including various ills, reproductive performance, retardation, premarital pregnancies are all highly personal and often guarded family secrets. The way in which such information is given and the reaction of the partner is most revealing. It is possible to acquire a fair estimate of where this couple is in its marriage and who is the dominant figure. One can also gain some idea of individual and combined strengths just by listening to the way the pair talks about its families and how much each knows about the other. Pedigree taking is time consuming and could be done by trained ancillary personnel so as to ascertain all scientific information necessary. However, it is during this time that the close personal contact with the couple is established. Sensitivity to the problem as presented, assurance of concern, acceptance of the couple, are all established along with a relaxed, loving atmosphere. For these latter reasons I have continued to do all the interview work myself and have also continued to avail myself of the privilege of wearing a Roman collar. My identification as a priest gives a definite advantage because patients expect sympathy and understanding and more than just a professional interest. Much of the rapport which a physician must work to establish is more easily attained. Somewhat surprisingly this seems to develop into a greater advantage when the clients are not Catholic than when they are.

After all medical and pedigree information and all laboratory reports are available, the risk figures are presented. This includes education about the disease(s) and their range of reaction, penetrance and expression, and instruction about statistics. The amount of time and the method of relaying this information will be somewhat dependent on the couple's pre-education and what it is able to hear. This may sound strange but from the pedigree interview one should be aware of which partner has been accepting the responsibility as well as the prejudgements brought to counseling. Often one partner has already made a decision and the couple is coming to genetics counseling so that the other may be convinced by the size of the risk. A counselor should, of course, avoid being used as an instrument by one or the other partner. It may seem that the premade decision is not to risk procreation again on the part of one member but, almost as often, there is the desire on the part of one individual to ignore the risk.

Human Drama

The very human drama that this situation encompasses should be recognized by the counselor. While he is only able to treat this couple at one point in time, he must still recognize the psychological habits which are already present and the dominance relationship already established. Dominance is not, of course, always what it may seem on the surface. The male is still looked to as the dominant figure and most often thinks he is, while in actuality the wife may control. Certainly in the area of procreation the wife is most often the decider even where there is clear dominance of the male in other areas.

Often guilt is the greatest handicap to overcome in presenting information. In cases where inheritance is dominant or in sex-linked conditions, one member of the couple does have the responsibility of being the sole bearer of the gene causing the condition. Sex-linked conditions are most difficult. The conditions which are presented to the counselor are generally those with rather drastic physical effects, e.g., duchenes muscular dystrophy and hemophilia. Secondly, the already built-in maternal feeling of responsibility and the feeling of failing to be an adequate female is compounded in this situation where the wife alone is the carrier. One need only attend a meeting of the local hemophilia society to recognize the enduring pain expressed by these mothers. Cases of dominance are generally somewhat easier to handle since the question of procreation is only being raised because the health of the bearer of the gene is sufficiently adequate to allow marriage and the physical possibility of reproduction. Many of the diseases which fit this category have a rather wide range of reaction and often this range is already well known because of the varying expression found in the family. Decisions are often made based on the degree of expression shown by the bearer of the gene or the effect in his or her immediate family. The more debilitating the effect, as directly experienced, the less likely are individuals to elect to procreate. Also, in dominant disease with variable expression, the genetic milieu in which the gene finds itself has an effect on its ultimate expression so that there is major responsibility on the part of one party, but the second parent also provides one-half of the background in which that gene will express itself. This fact allows for some sharing of responsibility.

In those cases of recessive disease or polygenic disease, sharing of responsibility is not a problem. It is interesting that we seldom see the more common recessive diseases. Probably these couples are receiving counseling from their primary physician. Tay-Sachs disease, cystic fibrosis, etc., have a rather clear inheritance pattern and most couples are exposed to other parents of such children and/or to organizations founded to help those afflicted and their families. Inquiries about recessive disease is more often made by siblings of affected individuals and questions about the ability of identifying carriers are frequent. A large segment of our population is present with conditions where the inheritance pattern is not clear but where there exists a genetic component to the condition. Thus, conditions such as spina bifida, hare lip and cleft palate, schizophrenia and others have a recurrence risk dependent on the frequency of affected individuals in pedigrees. These risk values are arrived at by empiric means and offer our best estimate for a population of couples like those presenting. An understanding of empiric risk and the values contained are essential.

Positive Attitude

Risk values must be presented in a positive as well as a negative fashion. A recurrence risk of five percent for some condition sounds pessimistic; a ninety-five percent chance of having a child normal for that character sounds very optimistic. Again it is necessary to know what a couple is trying to hear and what it will block. One husband who had a hare lip and cleft palate reasonably well repaired surgically was very anxious that his children not suffer as he had suffered. The wife, on the other hand, expressed her feelings that a risk of as high as fifty percent would be encouraging and even decisive in her estimation. Obviously, this couple needed to become more sensitive to the feelings of each other.

An essential part of the instructional phase of genetic counseling is the presentation of options by which a particular disease might be avoided while allowing for some measure of procreation. Most methodologies are morally unacceptable to many, including myself, but cannot be ignored or kept secret if they are legally and medically available. The primary methods are at present selective abortion and artificial insemination. (Other methodologies such as artificial inovulation, embryo transplantation and extrauterine gestation may be closer to reality than most of us care to recognize.)

Methods which the genetic counselor considers immoral may not be passed over in silence or counselled against, just as methods or approaches he considers to be most proper or moral should not be urged upon the couple. Patients are referred and come seeking genetic counseling and not necessarily moral counseling. Unless they ask for the counselor's moral opinion and advice, he has no obligation or even right to force this upon them. I hold this opinion for several re ns.

1. A couple requesting inf nation should not have inform on withheld.

2. The greatest virtue is prac ed in choosing what is morally ght when one is aware that this is ree choice rather than doing the c ect thing because there was no her alternative.

3. Many and perhaps most netic counselors believe that sel ive abortion and artificial insemi ion are morally correct procedur especially in the effort to avoi roducing seriously handicappe hildren. They do recommend ese procedures. The best one can pe to do at present is to neu lize this influence by urging a fai resentation of information wi out persuasion.

4. Witholding information f n a couple possibly carrying a D n's child or a child with Tay-Sach disease, etc., can well lead to a d vastating legal action as demonst ted in a recent rubella case.

Decision Making

The most important and nsitive part of genetic counselin begins after the scientific inform tion has been completed. Interest ngly this segment is the one most ften omitted. I have seen excellent medical workups, brilliant diagnosis and more than adequate information giving, followed by nothing, or even worse, by the question, "What do you intend to do?" — and this before a group of residents and students. Far worse, in my estimation, is the situation in which the genetic counselor maps out an approach such as giving a positive response to attempting another pregnancy but encouraging amniocentesis with subsequent abortion depending on the findings. Couples often will accept and want this kind of management. It is easy to let the physician take the responsibility of decision making. However, this approach takes away the freedom and the responsibility which can only reside in this individual or this couple. To the question, "What do you think we should do?" or "What would you do if you were us?" the genetic counselor should give no answer, nor, as far as possible, any indication of an answer.

What is needed in the professional situation of genetic couseling is some indication of how decisions should be made, the gravity of the decision and the very positive effect the making of a proper decision arrived at in the proper way will have. A couple involved in counseling is generally aware of the importance of this decision and how it will change their whole married life. Sympathy and understanding and a concern on the part of the counselor are vital. Insight gained through pedigree studies now becomes essential; rapport with the couple is vital. At least two related processes must be understood for a couple to reach a decision about its reproductive future which will enhance its marriage rather than cause deterioration.

First the partners must take the time to communicate what effect having a child with a specific problem would have on themselves, on each other and on their life together. This may be very theoretical and certainly they may react quite differently than they foresaw, but an intelligent, concerned individual must make decisions based on the best present estimates. This means, of course, looking at values, needs, desires and capabilities. How much does this woman need to carry and deliver a child; what has been their experience in the past? The sensitivity called for by each partner is almost heroic. Often important factors in the decision are impossible to put into words and need to be communicated and understood by other means. This is not an exercise which can be accomplished in your office or in a day or a week. Only this couple can decide because only this couple has access to the data. Often others should be brought into the process and here one would hope a trusted moral counselor would have tremendous effect.

Secondly, the decision reached must be a fully mutual decision. This should follow from the former exercise but does not necessarily. Often the needs or desires of one member will so influence the other that he or she will go along with the partner. This is generally a very loving decision made with high motivation but still not sufficient. If he or she decides to accept the risk because of the many positive factors to be obtained, the opposite member cannot just go along. If a couple takes the heightened risk of having a gravely defective child and such a child is born, the burden and the guilt will eventually fall on the individual who dominated in the decision. This will cause added hardship on the child and place a grave strain on the marriage. Going along is not really the loving thing to do. The decision reached must be mutual, accepted with full responsibility by each individual.

The atmosphere in which the counseling ends is positive. The importance of the decision to be made is in almost every case obvious to the couple and the need to make the best decision possible is equally obvious. Whatever decision that is made, the life of this couple will be changed. If the decision is well made they should have a closer, happier marriage than many who are never forced to engage in this type of exercise.

I am told that many marriages with genetic problems end in divorce. Reproductive incompatibility seems to be the ultimate confirmation of incompatibility my experience only one cou has been divorced and the c itive factors in this case were n ally genetic. This result may be e to the biased population seen ut I hope and pray that the ap ach taken has been a help towar olid-ifying the marriage relat ship rather than one that ulti tely causes dissension.

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