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AN ETHICAL VIEW

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Genomic advances promise to revolutionize the provision of health care in the coming decades. With these advances come new challenges. How can the benefits of genomic progress be provided in a way that is ethically sound and consistent with the values of the Catholic health ministry? Because much genetic testing and counseling is related to reproductive decisions and reproductive medicine, Catholic institutions may have a tendency to avoid offering genetic services. Yet experience shows that genetic services can be provided in ways that are consistent with Catholic values. Indeed, given the increasing importance of genomics, "the critical question for Catholic health care is not so much *whether* we should pursue genomic advances, but, rather, *how* we should pursue them."[1]

The Catholic Health Association has prepared a Catholic vision statement to guide our consideration of genomics. This statement, *Harnessing the Promise of Genomics: A Catholic Vision toward Genomic Advances,* focuses on three foundational principles:

- The *principle of human dignity* requires that we respect every human life, its value and its potential, and that we work to eliminate practices that discriminate among humans on the basis of perceived or actual limitations.
- The *principle of relationality* requires a balance between the needs and desires of an individual, on one hand, and, on the other, the responsibilities and relationships of that individual within the family, the community, and the larger society.
- The *principle of solidarity* reminds us of our obligation to care for those who are most needy, economically, physically, or psychologically, and our responsibility, as we pursue medical and genomic advances, to ensure basic health care for all.Although abstract, these three principles have direct application in concrete situations encountered in genomic medicine. In this article, I will examine their application in two broad areas: (1) genetic testing and screening in relation to reproductive decisions; and (2) screening and testing of children, including newborn screening and presymptomatic genetic testing.

Preventing the Transmission of Genetic Diseases

Prospective parents as well as their obstetric providers hope for the birth of a healthy child. The healing mission supported by the Catholic Church recognizes that preventing a disease or disabling condition is generally preferable to trying to deal with its effects later on. The goal is not perfectionism but the basic health status that is the foundation for normal human flourishing.

The church has long recognized this goal, even in its *Code of Canon Law,* where one of the impediments to valid marriage—blood relationship—was originally based on the belief that the offspring of two closely related individuals were "subject to grave physical and mental weakness."[2] Blood relationship was used to identify couples who were likely to transmit a hereditary defect,

thereby implying that the prevention of such defects was regarded as desirable by the church.

What would the church say about this matter today, when we know much more about genomics, genetic testing, and carrier status? The bishops of Vatican Council II provided guidance in their discussion of "responsible parenthood." They advised couples to consider five criteria when making reproductive decisions:

- The good of the marriage
- The good of the children, both those born and those yet to come
- The couple's financial and other resources
- The couple's spiritual development
- The good of the church and society[3]

Some, if not all, of these criteria are affected by information about genetic conditions and risks for transmitting genetic diseases. The principle of relationality is expressed prominently in three of the criteria: the good of the marriage, the good of all the children, and the good of the larger community. Responsible parenthood mandates consideration of all these relationships when reproductive decisions are being made.

The bishops' instruction also noted that a reproductive decision is a personal and individual decision, to be made by the couple themselves. It is not appropriate for medical professionals to identify the "right" decision, nor for governments to promote eugenic programs. Even moral or religious counselors, although they may raise issues of moral responsibility, must respect the consciences of the individuals who have to make the decision. Respect for human dignity requires that individual conscience, or the joint conscience of a couple, be respected as sacrosanct.

A decision of conscience must, of course, be based on adequate information. Thus it is essential that people at genetic risk have access to genetic counseling and whatever testing is deemed appropriate, given their risk status, and that they be provided with accurate scientific and medical information as well as psychosocial resources. Although it may be more difficult to locate, pastoral counseling from a person knowledgeable about genetics and reproduction in the context of the couple's faith tradition is highly desirable.[4]

For a Catholic, responsible parenthood in relation to genetic risks may lead to a decision *not* to have biological children, or additional biological children, perhaps accompanied by the choice of adoption. Pope Pius XII explicitly supported a couple's decision to prevent the conception of severely diseased offspring through avoiding intercourse during the woman's fertile period "even for the entire duration of the marriage."[5] If carrier testing were done prior to marriage, it could result in a decision not to marry a particular person who is also a carrier. Alternatively, a couple might decide that they are able to accept the prospect of a genetically compromised child, along with the burdens that would bring. Many options are permissible within Catholic teaching, and each couple must make their own decision based on the criteria for responsible parenthood.

Attitudes toward Disabled People

People living with genetic or hereditary disabilities sometimes object to efforts to prevent the transmission of genetic diseases. In their view, such efforts imply that the lives of affected individuals are not of value—that those people should not have come into existence.Efforts to reduce the transmission of genetic diseases may affect disabled people in two ways: Such efforts may seem to deny their right to have their own biological children, and they may reinforce negative attitudes toward the disabled.

Regarding their own reproductive decisions, adults with genetic diseases or disabilities have the same rights and responsibilities as everyone else. Having experienced a particular condition, they may be in a good position to weigh the suffering—as well as the satisfaction—that an affected child would experience. Different people will arrive at different conclusions, just as nondisabled persons do. On the public television documentary, *Short Stories,* two married couples with achondroplasia (dwarfism) comment on their offspring. One couple says, "There should always be people like us in the world"; and the other couple, whose two sons are unaffected, says, "We're glad that we are the last generation that will be this way."[6]

Interest in the prevention of genetic diseases may appear to negate the value of people who have these conditions, but it need not. As with AIDS, we must separate the disease (which we aim to prevent) from the person who already has it. Our society has surely progressed in its understanding of the needs of people with disabilities. While much remains to be done, legal requirements and changing societal attitudes have

resulted in accommodations and resources that make it possible for disabled people to live full and productive lives. The principle of solidarity with the disadvantaged places a special responsibility on the Catholic health care ministry and the church to act as advocates for the needs of disabled persons.

Prenatal Diagnosis

Often a health care provider will choose not to offer prenatal genetic diagnosis on the assumption that the only reason for such diagnosis is termination of pregnancy if the results are positive. This belief is mistaken because some couples wish to have advance warning of a disease or disabling condition so that they can make preparations. Preparations might include concrete plans such as delivery at a site equipped to care for the newborn, as well as the parents' own psychological and emotional readiness.It would not be appropriate for a provider to condition prenatal diagnosis on a commitment by the couple concerning how they intend to use the resulting information. Experienced professionals in the field know that people often change their minds as to their intentions after they have received the results of a diagnosis. As the director of a genetic counseling program at a Catholic hospital expressed it, "We are not thought police." The director's program provides morally permissible diagnostic procedures and information but does not try to coerce the couple concerning how they will use the resulting information.

Prenatal testing services offered by Catholic providers can offer a safe context for potential parents who may feel pressure from other providers to terminate pregnancy in response to positive test results. Although most certified genetic counselors act as neutral providers of information, as is consistent with their code of ethics, some obstetricians take a more assertive position in relation to pregnancies they feel should not continue. Genetic services as part of the Catholic health ministry can act as a counterbalance to such pressures.

Before choosing to have prenatal diagnosis through amniocentesis, however, a couple needs to understand that the procedure itself presents some risk to the pregnancy. If their purpose in requesting the procedure is solely for reassurance, then they will want to examine whether this benefit outweighs the risk. In any case, the risk of miscarriage or other harm to the fetus is not acceptable unless there is a specific reason for testing, such as familial history or advanced maternal age. For appropriate patients, the availability of noninvasive screening for chromosomal disorders in the late first trimester offers a great advantage in avoiding the risks of amniocentesis.

The discussion of possible fetal therapy in response to a positive prenatal diagnosis should be approached with caution. There are few interventions that can remedy genetic or other conditions during the fetal period. Maternal-fetal surgery for myelomeningocele has been offered at several centers, but it has not been shown that results are any better than when surgery is done shortly after birth. Currently, this procedure is under clinical trials and should be offered only as part of a trial.[7] Gene therapy research during the fetal period has not yet been attempted. Since gene therapy has had only limited success with born children, it would be premature to propose any extension of the research to fetuses.[8] In almost all cases, it would be inappropriate to suggest to potential parents that there are efficacious intrauterine therapies for problems diagnosed in the fetus.

Few interventions can remedy genetic conditions during the fetal period.

Expanded Carrier Screening

In 1997 a consensus conference at the National Institutes of Health recommended that cystic fibrosis (CF) carrier screening be expanded to all couples planning a pregnancy or seeking prenatal care. (Previously, only individuals with a family history of CF or reproductive partners of individuals with CF were recommended for screening.) This expansion of genetic screening represented the first large-scale application of genomic information to a broad population.

In order to prepare its members to implement the new recommendation, the American College of Obstetricians and Gynecologists (ACOG) prepared laboratory and clinical guidelines in collaboration with the American College of Medical Genetics (ACMG). In October 2001, ACOG and ACMG issued these guidelines together with patient brochures, sample consent forms, and letters for informing patients of various test results.[9]

If, having been screened during an already-established pregnancy, a couple found they were both CF carriers, they would have to decide

whether to proceed with prenatal diagnosis for a possibly affected fetus. For a couple at the stage of planning a pregnancy, positive test results would allow them to consider other options. For example, they could decide not to take the risk involved in becoming pregnant. They could also consider preimplantation genetic diagnosis (PGD) in conjunction with in vitro fertilization. This process, which involves the discard of affected embryos, is not morally acceptable under Catholic teaching and could not be offered by a Catholic facility. Although the couple contemplating PGD should be made aware of Catholic objections, the choice, as with other decisions, is theirs to make. Since PGD involves the discard of embryos at approximately two to three days after fertilization, some have argued that it is morally preferable to termination of an established pregnancy.

Does screening make sense for couples considering marriage?

Any genetic testing or screening program should encourage people who test positive to inform their relatives that they are also at risk. The ACOG clinical guidelines for CF screening include a sample letter that a patient could send to family members and relatives. Even though it is important that individuals who test positive inform relatives, experience shows that many people are reluctant to be the bearer of bad news; others do not feel confident that they are communicating the correct information. ACOG's sample letter accomplishes three goals: It implies that family notification is normal or expected, it ensures that the information is accurate, and it makes notification physically (if not psychologically) easy.

Premarriage Screening

If CF screening is now recommended for couples planning a pregnancy, could it also be offered to couples as part of premarriage preparation?

In the 1980s, a student of mine who had returned to college after raising her family became very interested in genetics. She was concerned that Catholic couples had little assistance in making decisions within the context of church teaching. She prepared material on genetics to be used within the premarriage sessions required for marrying in a Catholic church. The material included an informational packet and a family history form to be completed by the prospective bride and groom. The aim was not to provide professional genetic counseling, but, rather, to encourage couples to discuss what they would do if certain types of situations arose. Did they agree or disagree, for example, on the acceptability of abortion in case of fetal defect? The family history might also identify couples who could benefit from professional genetic counseling, possibly leading to genetic testing for a specific condition. My student worked hard at promoting her project in Catholic parishes, but received no encouragement whatsoever.

Does premarriage screening make any sense? One community, the orthodox Ashkenazi Jewish community, has had two decades of experience with a premarital screening program, *Dor Yeshorim*. Young people are screened for several genetic diseases prevalent in this community. The screening for carrier status is anonymous, with identification only by ID number and birthday. When two young people think they may be interested in eventual marriage, their identifiers are checked and they are told whether or not they are maritally compatible.

This program works because the community is highly coherent and young people are guided in their choice of spouse by their parents and rabbis. Some years ago, Fr. Robert Baumiller, SJ, of Xavier University, Cincinnati, attempted to implement a research trial of CF premarriage screening in a Catholic population, but found little interest. However, if an individual knows of genetic risk factors in his or her family, then premarriage screening of this individual and a potential partner could lead to very fruitful discussions. If the two persons found that they disagreed significantly on how they would handle potential reproductive decisions, they might want to reconsider their marriage plans.

Newborn Screening

Newborn screening for genetic disorders may appear to be ethically unproblematic. However, recent recommendations for national newborn screening standards have raised concerns in relation to risk-benefit issues that have ethical implications.

In 1999 the Newborn Screening Task Force of the American Academy of Pediatrics (AAP) asked the U.S. Health Resources and Services Administration (HRSA) to address the need for uniform national newborn screening systems and policies.

HRSA turned to the ACMG, which recommended in early 2005 that all newborns be screened for 29 conditions, most of them rare. Recommendations were based on agreed-upon criteria, including the availability of a sensitive and specific test that could be used 24 to 48 hours after birth and evidence that "there are demonstrated benefits of early detection, timely intervention and efficacious treatment of the condition being tested."[10]

The last criterion has caused the most controversy. Critics argue that, except for a handful of the conditions identified, no one knows whether helpful treatments exist, or even whether the child involved will ever show signs of serious disease. Thus a child who tested positive might be subjected to treatments that were of no value or possibly even harmful. Jeffrey R. Botkin, MD, chair of the AAP's ethics committee, noted that many of the conditions "are not well understood, the spectrum of the disease is not well understood, it is uncertain how efficacious the treatments are, and it is uncertain how well people can tolerate the treatments."[11]

Supporters of the recommendations include the March of Dimes Foundation, which believes that it is important to obtain equity for all infants by eliminating disparities among states concerning which conditions are screened for.[12] On March 8, 2005, the *Federal Register* announced a 60-day comment period on the proposed recommendations, giving pediatricians, parents, and advocacy groups the opportunity to be heard before final recommendations were issued.[13]

Presymptomatic Testing of Children and Adolescents

A family that is at risk for a particular genetic condition might be interested in having a child tested for the genetic mutation underlying the condition. If the test is performed to make or to confirm a medical diagnosis, it will fall under general principles governing medical care. But if the test involves a late-onset disease, such as Huntington's disease or Alzheimer's disease, or a predisposition to a disease such as breast or colon cancer, then the decision will be more problematic.

Parents may request presymptomatic testing for a variety of reasons: to gain peace of mind, to provide a basis for educational or career decisions, to allow for early preventive or therapeutic interventions, or to act as a guide for eventual marital and reproductive decisions, among others. When children become aware of genetic conditions in their family and extended family, they too may become concerned and, as a result, express an interest in testing. Adolescents in particular may want to make their own decisions about being tested.[14]

The AAP opposes predictive or predispositional genetic testing of children unless there is evidence for anticipated benefit to the child. The academy's Committee on Bioethics noted that many at-risk adults choose not to have such testing. That being the case, the committee said, the testing should not be imposed on a child who is unable to make the choice for him or herself, and who cannot undo the test once results have been obtained and communicated.[15]

A joint report from the American Society of Human Genetics and the ACMG states that direct and timely medical benefit to the child should be the primary justification for genetic testing in children and adolescents. However, they acknowledge that substantial psychosocial benefits to the competent adolescent may also be a justification for testing. But "if the medical or psychosocial benefits of a genetic test will not accrue until adulthood, as in the case of carrier status or adult-onset disease, genetic testing generally should be deferred."[16] Testing specifically for carrier status is mainly directed to marital and reproductive decisions. In some situations—for example, the *Dor Yeshorim* program mentioned above—genetic screening is offered to older high school students who may be competent but are not yet adults. Such carrier screening is rare in the United States. The AAP recommends that it be offered only to adolescents who are pregnant or planning a pregnancy, and who are fully informed of risks and benefits. In the case of an unmarried adolescent, parental involvement in the process is desirable. However, the laws of most states protect the confidentiality of adolescent health care related to pregnancy. ■

Critics argue that, in most cases, no one knows whether helpful treatments exist.

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