

The Human Genome Project: Examining The Arc's Concerns Regarding the Project's Ethical, Legal, and Social Implications

Following is the text of an address presented by Sharon Davis, Ph.D., on November 12, 1997, at the DOE Human Genome Program Contractor-Grantee Workshop VI ([abstract](#)). Dr. Davis represents The Arc, a 140,000-member national organization on mental retardation. A 2-year project at The Arc is being funded by the DOE Human Genome Program's Ethical, Legal, and Social Issues (ELSI) Program. This project will develop and disseminate educational material for members and leaders of The Arc's 1100 affiliated chapters to inform them about the Human Genome Project and mental retardation and to conduct training through The Arc's existing training vehicles.

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The Arc

I represent The Arc, a national organization on mental retardation whose members are united by a concern for the welfare of people with mental retardation and their families. These members work through 1100 state and local affiliated chapters throughout the United States to develop services and to advocate for full community participation by people with mental retardation. One in ten families is touched by mental retardation in this country, and our members come from all walks of life.

In February 1996, the DOE ELSI Program granted funds to The Arc to conduct an educational program with two major purposes. The first is to make our members aware of this scientific undertaking, the Human Genome Project. The second is to examine the critical issues related to new genetic discoveries affecting people with mental retardation and their families. We want as many people as possible to begin to understand that this research affects all of us. Working with me on this project is Leigh Ann Reynolds, who is here in the audience.

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Mental Retardation

Mental retardation is attributable to any condition that impairs the development of the brain before birth, during birth, or in the childhood years. It is caused not only by the genetic makeup of the individual but also by the possible influences of environmental factors. These factors can range from drug use to childhood diseases to poverty and cultural deprivation. A person is considered to have mental retardation when his intellectual functioning level (intelligence quotient or IQ) is below 70 to 75; significant limitations exist in two or more such adaptive skill areas as self-care, communication, functional academics, and work; and the condition is present from childhood (age 18 or below).

Roughly 90% of people with mental retardation have IQs above 50. In 50% to 75% of these children, the cause is unknown. Most identifiable causes (up to 60%) of severe mental retardation (IQ 50 and below) originate from genetic disorders. Two of the most common genetically transmitted forms of mental retardation are Down syndrome (a chromosomal disorder) and fragile X syndrome (a single-gene disorder). Chromosomal disorders affect about 7 out of 1000 infants. Single-gene disorders affect about 1 in 1500 births. More than 750 genetic disorders have been identified that cause mental retardation.

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The Human Genome Project's Importance to The Arc

When we began our education program, we surveyed The Arc's Board of Directors and learned that only 2 of 24 had heard of the Human Genome Project. While I'm told this is typical of the general public, we believe the leaders and members of The Arc should be more knowledgeable because so many are affected personally. Represented on the board are individuals who have a family member with Down syndrome, fragile X syndrome, Angelman's syndrome, Cri du Chat, and perhaps other genetic disorders. Not only had they not heard of the Human Genome Project, they were generally unaware of the ethical, legal, and social issues affecting families with genetic disorders.

To remedy this general lack of knowledge among our leaders and members, we developed educational materials that have been furnished to our chapters and members on such topics as these:

- Overview of the Human Genome Project,
- Genetic causes of mental retardation,
- Genetic discrimination,
- Genetic testing, screening, and counseling, and
- Genetic privacy.

We also have produced fact sheets on such specific disorders as PKU, Prader-Willi syndrome, and fragile X syndrome. We're still working on others. Interested persons can access materials through our Web site (<http://TheArc.org>).

Workshop presentations at chapter conferences are another part of our educational efforts. We introduce the Human Genome Project using clips from the video developed by the NIH National Human Genome Research Institute. We review the ELSI Program and specific issues we are addressing. Workshop participants have an opportunity to discuss issues and present their views in small groups as they discuss case scenarios designed to elicit a range of perspectives. We conduct pre- and post-testing to gather our members' views on various issues. This information is provided to our Board of Directors to give them a sense of the views of the members they represent. So far, about 500 people have participated in these sessions. Today, I would like to present an overview of issues being addressed by The Arc's leaders and members.

After we review the Human Genome Project's scientific undertaking and the ELSI Program, we tell people that the project deserves our attention because it has a direct impact on individuals and families affected by mental retardation. Researchers are rapidly discovering new genes. We need to consider the impact of such information on society.

The lack of current treatments and emerging gene therapy research raise ethical dilemmas regarding such areas as privacy, fairness, and discrimination. We point out that we are ALL affected. We need to educate ourselves and speak out on the issues that will eventually, either directly or indirectly, affect us all.

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The Arc's Issues

Even though there is a wide range of issues important to families, we decided to focus on these three topics:

- Insurance and employment discrimination based on a person's genes.
- Decisions involving genetic testing, screening, and counseling (including prenatal and newborn testing).
- Ethical issues involving genetic therapies that may eventually "cure" mental retardation.

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Genetic Discrimination

The first issue we address is genetic discrimination. Discrimination based on the presence of a disability has always been an issue of great concern to The Arc. People with mental retardation have long been discriminated against in both

insurance and employment. Now a new, less-known type of discrimination is emerging due to the increasing use of genetic tests that can reveal the presence of defective genes. This new phenomenon is called genetic discrimination.

Genetic discrimination occurs when people are treated differently because they have a disease gene or genes, even when they show no symptoms of disease. Men and women who are carriers for a genetic condition may also be discriminated against because of their potential to have a child with a genetic condition.

An example of genetic discrimination was reported in *Newsweek* last December. A pediatrician who suspected that a 2-year-old boy (David) had fragile X syndrome ordered lab tests that confirmed his suspicion. The family spent several years learning to deal with David's condition. At age 6, David visited a neurologist who scribbled "fragile X" on a health insurance company claim form. The company responded promptly by canceling coverage for the entire family of six. There is no medical treatment for fragile X, and none of David's siblings had been diagnosed with the condition.

The *Washington Post* cited a case in which a pregnant woman whose fetus tested positive for cystic fibrosis was told that her health maintenance organization (HMO) would be willing to cover the cost of an abortion but would not cover the infant under the family's medical policy if she elected to carry the pregnancy to term.

The denial of health insurance based on a person's genes is one of the most common forms of discrimination. Insurance companies gather and use medical information to predict the risk of illness and death. They then determine which individuals and groups to insure and at what price.

Genetic discrimination has been documented in other settings as well. A recent study identified cases in which healthy people were fired from jobs, treated differently in school, or barred from adopting a child because they carried genes that could potentially result in disease or disability.

Legislation Banning Discrimination. The Americans with Disabilities Act (ADA) offers some protection to those currently affected or predisposed to a genetic disorder. ADA does not, however, cover the insurance industry as long as its decisions on coverage are based on experience that demonstrates people's risk of illness, disability, or death.

As the number of genetic tests increases, discrimination will increase. That's why legislation banning discrimination has been proposed. The Arc's Board of Directors passed a resolution last year calling for such legislation. The U.S. Congress made a beginning step to deter genetic discrimination when it passed the Federal Health Insurance and Accountability Act of 1996. This law limits the use of preexisting-condition exclusions by health insurers and specifically mentions "genetic information." Other legislation is being considered by the U.S. Congress.

A number of states also have laws banning genetic discrimination in health insurance. Some states also are examining discrimination in employment and passing laws banning such discrimination based on genetic information. Much of this legislation, however, has serious deficiencies.

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Genetic Privacy

Legislators are beginning to address issues of genetic privacy. Medical records are not private. If you have ever applied for individual life, health, or disability insurance, information about you may be in the Medical Information Bureau (MIB), a computerized database of medical and some nonmedical information. Originally created to prevent consumer fraud, MIB holds information on 10 million to 20 million Americans. If an insurance applicant has a condition pertinent to health or longevity, such information must be provided by insurance companies to MIB. Nonmedical information that could affect insurability, such as an adverse driving record or participation in hazardous sports, also can be reported.

You must give consent to MIB to establish your record or for an insurance company to search your MIB record. If you don't give consent, the insurance company may automatically deny coverage. Next time you fill out an insurance application, examine it carefully. I recently received a letter urging me to apply for mortgage insurance, and sure

enough, if I signed the application, I was giving permission for the company to access my MIB file. An individual in one of our recent workshops informed the group that he was denied mortgage insurance because of diabetes, a condition listed in his MIB file. As this database grows, some people are concerned that corporations, agencies, and others will have access to our medical records and genetic information just as they do to our credit histories.

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Genetic Testing, Screening, and Counseling

Issues related to genetic testing, screening, and counseling are another area of concern to The Arc. Thanks to the Human Genome Project and the identification of new genes, specific laboratory tests are being developed that can determine whether or not an individual is at high risk for a particular genetic condition.

More and more people will be able to know whether or not they will develop a disorder or possibly pass a disease gene on to their children. How will having this information impact society? Do most of us want to know about future genetic information that could change our lives? For example, if you could find out today that you have an 80% to 90% chance of developing a rare and untreatable form of colon cancer in the future, would you want to know?

Other issues of concern include the following: Must a physician offer prenatal genetic screening to all pregnant women or risk medical malpractice liability if he doesn't? Should a woman have a right to refuse prenatal screening? What if she's already had a child with a serious genetic condition? One of our workshop participants reported that her 3-year-old son's serious genetic condition had already cost more than \$1 million, paid for by the state. She expressed her intention to have another child and her strong belief that she would refuse prenatal testing. What about testing infants and children for genetic conditions when there is no treatment available?

Positive and Negative Consequences of Testing. Genetic testing can have both positive and negative consequences. On the positive side, testing can help people make more informed decisions about their future. Another advantage of testing is that it provides an opportunity to seek genetic counseling so that the risk of passing on a disease gene is reduced.

On the other hand, anxiety runs high when individuals are confronted with a positive test result confirming a genetic condition. Are people prepared to deal with such news? Also, testing does not provide clear-cut answers. A positive result does not guarantee and a negative result cannot completely rule out the possibility that a person will become ill or be affected by the condition. Environment and other factors play a role. Another problem is that physicians may misinterpret the results of genetic tests now available.

If positive test results are not kept private, the individual risks the possibility of losing health insurance and employment for herself, the children, and other family members.

This concern was expressed to me at a recent meeting by a woman who is president of a local chapter of The Arc in Massachusetts. She learned recently that her two older brothers, now in their 60s, have fragile X syndrome, a genetic condition that causes mental retardation. Her two grown sons are unaffected. She could be a carrier, however, and so might they. There's now a simple test for fragile X, but she has read The Arc's educational materials and is concerned about genetic discrimination. She doesn't want any positive test results on her record or her sons', so she has decided to take the fragile X test and pay for it herself. If she is not a carrier, her sons won't have to make a decision regarding testing. Furthermore, her insurance company won't know the results.

Testing of Children. One major area of concern regarding genetic testing is the testing of children. Parents often want to have their children tested to plan for the future or reduce the anxiety of not knowing. Yet, receiving positive test results may actually restrict the child's future in many ways. Children suffer the possibility of being stigmatized for their condition, with a resulting loss of self-esteem. They may also face discrimination by family, employers, insurance companies, educational institutions, and others. What rights does a child have to agree to or refuse testing?

There are currently no universal standards among physicians for testing children. Health professionals usually recommend that testing be conducted only when there is a clear benefit to the child. Family members, on the other

hand, may feel testing should be considered if it can benefit others. For example, a member of our project's advisory committee had a daughter with severe disabilities but no diagnosis. He had his daughter tested because his sister, who wanted to have a child, wished to rule out a genetic condition in the family.

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Gene Therapy

The third issue area we're discussing as an organization is the use of gene therapy, which potentially could cure some genetically caused mental retardation. Gene therapy is an experimental treatment in which normal genes are introduced into the body's cells to correct or modify the cell's function.

We inform our members that, although gene therapy holds great promise for treating some genetic diseases and conditions that have not been treatable previously, no one has ever been "cured" of their condition by gene therapy. Much more scientific research is needed for gene therapy to have a significant impact on the treatment of genetic conditions.

At this time, the most promising use of gene therapy for mental retardation is to treat single-gene conditions involving enzyme defects. Most of these defects cause significant disability that cannot be treated satisfactorily by such other approaches as modifying the diet or supplying the enzyme. Some examples include adrenoleukodystrophy, galactosemia, and the urea cycle disorders.

At some time in the future, it may also become possible to apply gene therapy to fragile X syndrome. Scientists, however, say that more knowledge is needed about the function and control of the fragile X gene before such studies are undertaken.

Gene Therapy and Ethical Issues. Gene therapy also raises some ethical questions related to the quality of life of people with diseases and conditions. We asked our members if The Arc should be advocating the development of gene therapies that can "cure" people with mental retardation. Some people say that disabilities are not diseases and, therefore, do not need to be cured or repaired. They feel that people with conditions such as mental retardation can cope with their condition and lead meaningful lives, not in spite of their disability but with it. Yet, The Arc always has supported efforts to prevent mental retardation. Should we, as an organization, support gene therapy that could one day cure mental retardation?

We ask this question because in some groups, people with disabilities say they don't want to be cured. A segment of deaf people refers to itself as the deaf culture and prefers to have children who are deaf. Some people with physical disabilities say that calls for cures demean them as individuals. These people are very critical of Christopher Reeve, who advocates funding to cure spinal cord injuries. They say he hasn't accepted his disability. Others have difficulty with the idea of no longer being disabled.

This is what Ted Kennedy, Jr., said in a recent interview: "I think of myself as a person with a disability. In fact, going through the 2 years of chemotherapy and the cancer, losing my leg--it was probably the defining event of my life. It shaped who I am. And, you know, if someone offered me a pill today that would somehow magically grow my leg back, I would say no."

We asked members of The Arc what they believe about curing mental retardation if it were possible. Some 90% participating in our workshops across the country said advocating for a cure did not devalue those with the condition. About 85% agree that The Arc should advocate for more funding for research to cure genetic conditions. This information will help guide our Board of Directors in its decision-making regarding these issues.

Arguments in Favor of Gene Therapy. The major argument in favor of gene therapy is based on its potential for treating individuals severely affected by their condition. A perfect example is Lesch-Nyhan disease, which is characterized by communication deficits, writhing movements, and involuntary self-injurious behavior. Males who have this disorder have to be restrained constantly to prevent them from inflicting severe damage on themselves. Most have their teeth removed to keep from biting their lips off. If we have a new medical technology that will cure this

condition, don't we have an obligation to use it?

Arguments Against Gene Therapy. A number of arguments are offered against gene therapy, including the concern about the potential for harmful abuse if we don't distinguish between good and bad uses of gene therapy. The eugenics movement of the 1920s to the 1940s found people with mental retardation being involuntarily sterilized, along with others considered less desirable. Another concern is that in mental retardation gene-therapy research, many candidates are likely to be children who are too young or too disabled to understand the ramifications of the treatment. Finally, gene therapy is very expensive and may never be sufficiently cost-effective to merit high social priority. Opponents say that if those who can afford gene therapy are the only ones to receive it, the distribution of desirable biological traits will widen the differences among various socioeconomic groups.

Genetic Enhancement. Another relevant issue is genetic enhancement, using gene therapy to supply a characteristic that a parent might want in a child but which does not involve the treatment or prevention of disease. Such physical characteristics as height, weight, and enhanced cognitive abilities are common areas mentioned.

What about cognitive enhancement for children with mild mental retardation? If cognitive enhancement could increase the IQ of such a child to allow functioning in the normal range, should it be considered? Currently, it is not considered acceptable to use gene therapy to treat conditions that are not diseases. Is mild mental retardation a disease that needs to be cured or a condition or trait that does not justify a need for cure? This is a topic we haven't addressed as an organization yet. We have raised it as an issue in our forthcoming report on ethical issues related to gene therapy. Even though such a cure does not exist yet and may never exist, we believe we should be involved proactively in discussing genetic enhancement before any policy is enacted governing use.

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Conclusions

The Arc's goal is to create an adequate level of awareness among its members on the issues I've outlined, so that our voices can be heard before harmful policy is created. There are differences of opinion on many of the issues we have addressed and general agreement on others.

We conclude by telling our members that genetic research is changing the world in which we live. As families potentially affected by the knowledge gained from research, we must become involved by being educated and expressing our views when appropriate. Even though ethical, legal, and social issues have been debated widely by scientists, ethicists, and others, few voices have been heard from families directly affected by a genetic condition. We urge our members to keep informed about the issues so they also can make informed personal choices regarding testing, participation in research, and gene therapy if it becomes available in the future.

We are also reaching out beyond our membership. The American Association of University Affiliated Programs for People with Developmental Disabilities is disseminating our materials to its member university-affiliated programs and research centers for mental retardation and developmental disabilities. The director of the Frank Porter Graham Child Development Center and Mental Retardation Research Center in Chapel Hill, North Carolina, who wrote the fragile X syndrome report for us, is sponsoring a conference for early childhood educators and is distributing all our reports to attendees. These professionals in the field often work closely with families and seek to collaborate with us in our common mission.

Finally, The Arc will continue to advocate for understanding and acceptance of the condition of mental retardation and fair treatment for citizens with mental retardation. The potential for cure for some must not make us less accepting of those living with the condition.

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