

Ethics of the Human Genome Project and Related Research: Who Benefits?

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I. The Human Genome Project: An Overview

A new mother holds her tiny infant for the first time and anxiously takes inventory: two arms, two legs, two hands, two feet, ten fingers, ten toes, forty-six chromosomes, thirty thousand genes, and 3.2 billion base pairs (1). This modern mother is a beneficiary of the Human Genome Project, an international effort launched in 1990 with the goal of sequencing the entire human genome by 2005. On April 14, 2003, the International Human Genome Sequencing Consortium came forward with a press release stating that the project had been completed with 99.99% accuracy two years before the date anticipated, appropriately coinciding with the fiftieth anniversary of James Watson and Francis Crick's discovery of DNA's double-helix structure (3). Researchers worldwide were given a road map of human life through public access over the internet and began to race towards destinations.

Along with the positive aspects of the Human Genome Project's completion came the fears of what could be done with this new information, such as human cloning and eugenically motivated experimentation. To help ease tensions and prevent fear from becoming immediate reality, three percent of the Human Genome Project's annual budget was spent on studying the ethical, legal, and social issues associated with perhaps the greatest biological research undertaking in history (3). Also helping to ease fears over such subjects was the promise of a brighter medical tomorrow through studying genetic diseases, susceptibilities, and pharmacogenomics, or the development of genome specific medications. This brings about perhaps the most challenging ethical issue surrounding the Human Genome Project: what research should be funded, and who should benefit?

One situation this dilemma leads to is that of the haves versus the have-nots. In this scenario, the focus and direction of genetic research involving the newly mapped genome would be driven by those affluent enough to lend monetary support. This approach, while violently out of favor with the doctrine of social welfare, also has the potential to become unclear. If many have-nots share a common problem, a common community, and a virtually common money supply, do the interests of this group enter into those of the haves, or should they simply be denied? Therefore this answer, even completely ignoring its socially Darwinist quality, is incomplete and unacceptable.

A second view on who should benefit from genetic research is that of priority by majority, or devoting nearly all funding towards major disorders while leaving people effected by those which are considered rare to hope for but not be guaranteed consideration of any real value. On the surface the approach of serving the most common need seems justifiable, yet underneath it must be recognized that when a person dies from a rare disorder, it is not insignificant; it is a human life. With no single life being better than any other, no loss can be taken lightly. To disregard them in this manner devalues life and is likened to disregarding victims of shark attacks because they did not die of heart disease, then neglecting to post seasonal beach warnings in order to reduce casualties. A second problem with priority by majority is how to define rare through the

scale used to do so; should be it national, local, ethnic, or gender specific? There are many options, however rare the disorder, and therefore its research priority must be seen as dramatically heightened when it threatens to obliterate an entire culture.

II. The Amish: Beliefs, Culture, and Crisis

During the Reformation of sixteenth-century Europe, many religious reformers split from traditional Protestant and Catholic churches and began to live in accordance with new beliefs. One of the largest of these groups was known as the Anabaptists. They believed in a separation from secular society and baptism as a choice-making adult rather than as an infant. Later known as the Mennonites after fleeing to Switzerland to avoid persecution, this group split into two entities, the Mennonites and the Amish, the latter named for their leader Jacob Aman. The migration of Amish to the United States began in the mid-eighteenth century, and they now number approximately 100,000, living in twenty-two states and part of Canada. As a result of William Penn's "holy experiment" promoting religious tolerance, the largest concentration of Old Order Amish, the most conservative, live as a group of between sixteen and eighteen thousand in Lancaster County, Pennsylvania (2).

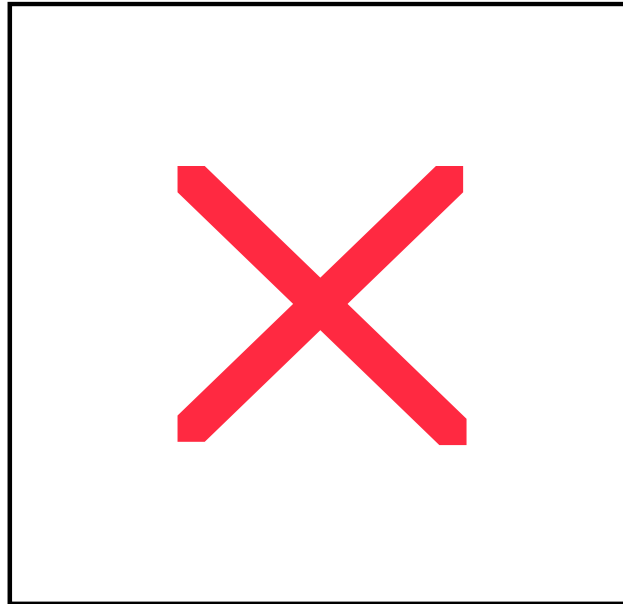
The Old Order Amish of Lancaster County are a group descended from roughly two hundred Swiss immigrants seeking tolerance for their beliefs. They are a self-sustaining community who do not reject modern technology but simply choose to examine advances closely before deciding on acceptance. If a technology is not necessary or could cause division within a community, such as cars or electricity being affordable by some and not others, they choose avoidance. In relation to medical care and attention, the Amish are not opposed to modern technology. They prefer to use midwives for birthing and community knowledge for minor illnesses, but they allow outside care for major diseases and complications. This is because of the Amish system of monetary assistance, sometimes known as Amish AID, which functions as community insurance. A family pays a certain amount for a certain period of time as dictated by local requirements, and they are provided for should they need funding for a hospital stay or medication (2).

The Amish pay all the taxes of regular citizens except for the Social Security tax, because they do not collect welfare, Medicaid, or Social Security as it would go against their belief of caring for their own. Amish children do not attend public school but rather receive private Amish schooling in one-room schoolhouses up to the eighth grade when they go to work full time on the family farm. In the home, the Old Order Amish speak a dialect of German known as Pennsylvania Dutch, and during worship services the language used is high German. English is only used when communicating with the outside world. Because of the harsh lifestyle, the Amish avoidance of active evangelism, and the outstanding language barrier, there are few to no converts. Marriage outside of the community is forbidden in all cases; should a member wish to marry an outsider he or she must leave the community on their own and is never allowed to return, even to visit family. This has resulted in the collective gene pool of the community remaining closed for over twelve generations, and it is still drawing, almost three hundred years later, from the original two hundred genomes of their founders (4).

As a result of their closed gene pools, genetic mutations, which were present in the initial population of settlers but were quite dormant, are now becoming common

illnesses among newer generations. Predictably these disorders include a host of dwarfism and mental retardation, but they also include an unusually high rate of dangerous and rare metabolic disorders. The rate at which these disorders effect the Amish population in comparison to the rest of the United States is alarming. An example of this is Ellis van Creveld (EVC) Syndrome, an inherited gene mutation that causes disproportionate dwarfism, postaxial polydactyly, ectodermal dysplasia, a small chest, and a high frequency of congenital heart defects (5). This syndrome effects one in sixty thousand live births in the general population, but among the Amish it is seen in five per every one thousand live births, affecting up to thirteen percent of the community (Fig.1). Fifty percent of those with the syndrome will die in infancy (4).

The most deadly and most common metabolic disorder among the Old Order Amish is known as Glutaric Aciduria Type I (GA-I), which stems from a rare mutation that both parents must posses in order to be inherited by a child. Children with the disorder appear healthy but may suffer severe brain damage if they catch even a simple



cold due to a lack of an enzyme that breaks down glutaric acid. When a healthy individual becomes ill, the body begins to break down proteins in muscle tissue, which are converted into metabolites such as glutaric acid. These are then processed into water and carbon dioxide. Without the enzyme that breaks down the acid in GA-I patients, the

Figure 1. Ellis van Creveld Syndrome (EVC) population effects

chemical builds up in the bloodstream and causes acidosis. It is theorized that this chemical abundance targets a region of the brain also associated with Huntington's disease and when left untreated causes brain damage,

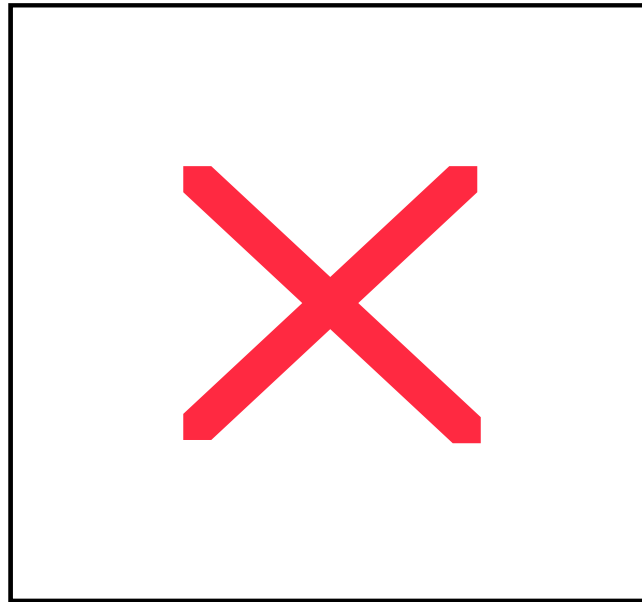


Figure 2. Glutaric Aciduria Type I population effects

paralysis, and seizures (4). The whole process can occur in a matter of hours. The frequency of GA-I in the general population is estimated to be one in forty thousand live births (6). Within the Old Order Amish community, the frequency is estimated to be one in two hundred live births, with one in ten Amish being carriers (Fig. 2).

While there is research devoted to both EVC Syndrome and GA-I and effective but exhaustive therapies for GA-I are available, these diseases receive less attention compared to other diseases such as cancer and Alzheimer's Disease. Those who are focusing on these rare conditions must divide their time between research and clinical treatment of patients, for example Lancaster County's Dr. Holmes Morton, who first began diagnosing the conditions among the Amish communities (4). The Amish, even as a whole, cannot fund large-scale research even if it would mean saving their culture. This leads to a dependence on public funding and a need for focus on disorders which threaten to erase entire groups of people, no matter the rarity in which the diseases occur in the general population.

III. The Big Picture and An Answer

One argument against this would be that the Amish are making a conscious choice not to integrate their genetics and continue to practice their way of life. It can be said that though screening is available, two carrier individuals still choose to reproduce and therefore choose at the same time to be excluded from the focus of finding cures. This viewpoint, however, is absurd. If this stance is allowed to be made, the ease with which saying diseases that effect mostly African Americans, those of Jewish decent, or one gender should not receive any more priority would increase. The scale may not be the same, but the fundamental concept is. It is not acceptable to ignore the needs of a particular group based on lifestyle or beliefs, forcing a choice between convictions and self-preservation, while acknowledging the staggering loss of life within these groups. If it becomes so, then the real threat posed by the completion of the Human Genome Project is not that of cloning or engineering better babies, but is effectively a genetic holocaust, wiping out whole groups of people. While the Amish do choose their lifestyle, this does not exclude them from the benefits of research, just as religious groups such as Jehovah's Witnesses choose not to vote but still benefit from laws enacted by policy makers.

The Human Genome Project is beautifully that--Human. It is not called the Protestant Genome Project, the African American or Caucasian Genome Project, or the Specific Geographic Location Genome Project. The ethical solution is in the name and must not be ignored. Our society, though quickly advancing, has a social responsibility to protect and preserve culture and life. To say that certain groups dwindling in number do not deserve attention is to place these groups of people lower than animals, as our society protects endangered species and has animal rights organizations. In light of this observation, denying special attention to groups of people on the basis of their culture being "slow" or "backwards" seems highly hypocritical and deplorable to the highest degree. The issue comes down to this solution and this warning: we must all benefit or we will all lose. We have achieved a massive scientific milestone in decoding our collective life, and the ethics must also progress in order to prevent dangerous biases in the use of this knowledge from leading to the demise of many, thereby destroying the resource from which it was drawn.

References

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