



# Genetic Medicine: Ethical Imperatives & Challenges

## **Genetic Medicine: Ethical Imperatives and Challenges**

James C. Denison, PhD  
President, the Denison Forum on Truth and Culture  
[www.denisonforum.org](http://www.denisonforum.org)

"Without question, man's knowledge of man is undergoing the greatest revolution since Leonardo."<sup>1</sup> This is how Francis Collins describes the breakthrough known as "precision" or "personalized" medicine. Dr. Collins is Director of the National Institutes of Health and former director of the National Human Genome Research Institute. In his introduction to this revolution, he quotes two researchers who state that "there have been few, if any, similar bursts of discovery in the history of medical research."<sup>2</sup>

We are on the cusp of a medical revolution more significant than any that has gone before us. My purpose in this paper is to introduce you to "precision medicine" and outline the ethical imperatives and challenges before us.

### **Introduction to precision medicine**

The human "gene" is the basic biological unit of heredity. You inherited thousands of traits from your parents—everything from eye color to height is influenced by them. Your parents passed these traits to you through your genes.

A "gene" is transmitted via a molecule called deoxyribonucleic acid (DNA). There are 20,000 to 25,000 different genes in human DNA, which is composed of three billion chemical base pairs. The entirety of your genetic information is known as your "genome."

In 1990, the "Human Genome Project" set out to decode these genes. Thirteen years and \$3.8 billion later, the project was completed with a map of the entire human genome. This remarkable project has accelerated greatly the pace of discovery, product development and clinical adoption of precision medicine.

Using this information, researchers are now able to identify the genetic components in 4,000 different diseases. Large-scale studies and sample repositories now link genetic variation to disease and therapy response. And healthcare information technology now exists to support the integration of research and clinical data. As a result, many scientists believe that in a few years, a patient will be able to obtain a full genomic sequence at birth for \$1,000.

This application of genetic science to medicine is known as "precision medicine." It has been defined as "the tailoring of medical treatment to the individual characteristics of

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1 Francis S. Collins, *The Language of Life: DNA and the Revolution in Personalized Medicine* (New York: HarperCollins, 2010) 5.

2 Collins, 70.

each patient." It involves "the ability to classify individuals into subpopulations that are uniquely or disproportionately susceptible to a particular disease or responsive to a specific treatment. Preventive or therapeutic interventions can then be concentrated on those who will benefit, sparing expense and side effects for those who will not."<sup>3</sup>

"Precision" or "personalized" medicine represents a "tectonic shift" in the practice and future of medicine, shifting the focus from reaction to prevention and reducing the overall cost of healthcare.<sup>4</sup>

### **Toward a Kingdom ethic**

In our day, ethical discussion has become secularized. What was once the purview of theologians has become the domain of public policy. This debate typically excludes larger questions about human nature and destiny, seeking the lowest common denominator of public consensus.<sup>5</sup>

As a theologian and minister, I choose to be led by God's revelation in Scripture. While a larger discussion of bioethics in biblical perspective is beyond our purpose, a brief overview may be helpful. I want to suggest a Kingdom ethic as the foundation for our discussion. What do I mean?

Jesus began his ministry with the call to "repent, for the kingdom of heaven is near" (Matthew 4:17). He taught us to "seek first his kingdom and his righteousness" (Matt. 6:33) and instructed us to pray, "your kingdom come, your will be done on earth as it is in heaven" (Matt. 6:10). When he returns, his name will be "King of Kings and Lord of Lords" (Revelation 19:16).

If God is your King, you're sitting in his chair as you read this paper. You're wearing his clothes and breathing his air. Everything that is, is his. He claims dominion and authority over every dimension of life, including the practice of medicine. Our goal should be to make him King of precision medicine, in all its implications.

What would such a Kingdom ethic look like? Here is my view.

First, we are stewards of God's creation. Scripture is clear: "The earth is the Lord's, and everything in it, the world, and all who live in it" (Psalm 24:1, NIV). Earth belongs to God, not to us.

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3 President's Council of Advisors on Science and Technology, *Priorities for Personalized Medicine* (September 2008) 7

(<http://www.whitehouse.gov/administration/eop/ostp/pcast/docsreports/archives>, accessed 15 March 2011).

4 Personalized Medicine Coalition, *The Case for Personalized Medicine* (May 2009) 1.

5 For a development of this theme, see Gilbert Meilaender's excellent *Bioethics: A Primer For Christians*, 2d ed. (Grand Rapids, Michigan: Wm. B. Eerdmans Publishing Co., 2005). His insights have greatly informed this presentation.

Genesis says that "the Lord God took the man and put him in the Garden of Eden to work it and take care of it" (Genesis 2:15). "Work" translates the Hebrew *abad*, which means to nurture or sustain. "Take care of" translates *shamar*, which means to protect, preserve, or guard. When we misuse the skies and soil, rivers and oceans he made, we violate the stewardship he has entrusted to us.

At the same time, our planet was created to serve us. God made it to meet our physical needs (Gen. 1:29-30; 9:1-3). By his design, our lives are sustained by its resources. We have a spiritual obligation to develop and utilize these resources in ways that honor God and his creation.

Such stewardship includes our bodies, gifts of his creative grace. For Christians, our bodies are the temple of the Holy Spirit and reflect on our Owner and Resident (Gen. 1:27; 1 Cor. 3:16; Rom 12:1-2). We are to extend this care to the physical lives of others.

Two: care for human life begins at conception. This is a very large debate, the complexities of which are beyond the scope of this paper, but I understand Scripture to teach that we are created by God at our conception. David said to the Lord, "You created my inmost being; you knit me together in my mother's womb" (Psalm 139:13). The Lord told Jeremiah, "Before I formed you in the womb I knew you, before you were born I set you apart; I appointed you as a prophet to the nations" (Jeremiah 1:5). We belong to our Creator and King from the moment we are conceived.

Three: we are required to care for the neediest members of society. To me, this obligation begins with the preborn, who are the most innocent and helpless of us, and extends to the diseased, the elderly and the infirm. They are all creations of our King and residents of his realm.

Four: we should seek *shalom* for all. Biblical "peace" is more than the absence of conflict—it is the presence of righteousness in our relationship with God, others, and ourselves. Our Father wants our best, and calls us into a divine-human partnership by which we are to serve him and one another.

### **Precision medicine before conception**

Now, let's survey the ethical issues raised by precision medicine, along a chronological spectrum that begins before conception and ends with death. Given a Kingdom ethic that calls us to be good stewards of the King's creation, we have a moral responsibility to use these techniques to enhance the health of our bodies and world. But we must avoid those practices that violate Kingdom principles.

We begin with the positives: Genetic screening can tell prospective parents if they are carriers of mutant genes. In the future, parents can be tested with regard to genetic possibilities for their potential children. We can already do this for Down syndrome, cystic fibrosis, spina bifida, sickle cell disease and other disorders. We will one day be able to do it for thousands of potential diseases.

Carrier screening then enables prospective parents to make reproductive decisions based on possible disease outcomes for their children. For example, the Jewish population is especially at risk for Tay-Sachs, an inherited disease that usually results in death by the

age of four. In response, a community of Orthodox Jews in New York City developed a program called *Dor Yeshorim* ("the generation of the righteous") to discourage marriage of young people who are found to be carriers of this disorder. The program has greatly reduced the incidence of Tay-Sachs within the community and has now been extended to screen for cystic fibrosis and Gaucher's disease as well.

You can see the enormous potential of this technology for eliminating genetically inherited diseases. But let's also consider some other implications that are more ambiguous.

Imagine potential mates being chosen for their genetic capacities and reproductive potential. Will genetic information be analyzed before couples marry, or even date?

Is this the future of human progress, a kind of eugenics? Will pressure be brought increasingly to bear on couples to produce children in this way? Will parents face lawsuits from children whose lives could have been prevented or diseases corrected by such techniques?

What are the implications for health care providers? Will physicians and hospitals be pressured to provide such services? Will they face litigation if they do not, or if their test results prove to be incorrect? How do they balance medical paternalism, doing what they know to be best for the patient, with the freedom of parents to bear children as they choose?

Will this technique be used as a kind of evolution in which only the healthiest and most intelligent genes are transmitted? Will reproduction one day be taken from the bedroom and confined to the laboratory? This is not the stuff of science fiction movies any longer, but a very real possibility in the next five to ten years.

### **Precision medicine for conception**

As difficult as the ethics of precision medicine may be before conception, they become far more complicated when applied to conception itself.

We begin with assisted reproduction. One out of every 1,000 pregnancies in America involves a disease that could have been predicted by carrier screening. "Assisted reproduction" is the umbrella term for a variety of medical techniques intended to produce a successful pregnancy:

- Artificial insemination: the use of donor sperm to inseminate the woman.
- *In vitro* fertilization: sperm and ovum are combined in the laboratory, then implanted in the woman's uterus.
- Pre-implantation genetic diagnosis (PGD): fertilized embryos are tested genetically for defects before being implanted.

Each of these techniques can be enhanced medically by genetic testing. Soon it will be possible for donor sperm to be selected for their genetic traits. Think of athletes or scholars selling their sperm or eggs to banks.

Some 100,000 embryos created by IVF but not implanted are currently in frozen animation around the country. If you believe that life begins at conception, as I do, then you must conclude that each of these lives was created and then frozen.

With pre-implantation genetic diagnosis, parents may soon be able to select not only the gender of their child, but his or her intelligence, singing voice and athletic abilities. How many lives will be created in this way in the future? How many embryos will be frozen?

How will these developments affect the stem cell debate? Stem cells are unspecialized cells that can be promoted to become any of the 210 different kinds of tissues in the human body. Embryonic stem cells are especially pluripotent, capable of becoming anything needed. But to obtain them, we must kill the embryo.

How is precision medicine relevant to this debate? On the positive side, stem cells could greatly enhance regenerative medicine. We can transplant stem cells today into patients suffering from conditions such as Parkinson's disease, diabetes, or spinal cord injury. What if we could genetically engineer cells to fight an entire spectrum of diseases?

Will embryonic research labs produce millions of embryos for such use? Will we see embryos cloned to produce stem cells for transplant, or for research to study a specific disease? Where is the line between sexual and asexual reproduction?

Abortion permits the destruction of a fetus; cloned embryos for research purposes would require such death. At what point would such reproduction be halted? Fourteen days? After organs have developed and can be used for transplantation?

How do these advances relate to organ donation? More than 100,000 Americans are currently on waiting lists for a donated organ. Organ transplantation was revolutionized in 1972 by the discovery of cyclosporine, a powerful immunosuppressive drug. However, survival rates for those who receive organs still vary, from 90% for kidney transplants to less than 50% for heart-lung procedures. One very real application of genetic medicine is our ability to engineer life for donor purposes.

Will laboratories create embryos designed to produce stem cells as needed for specific issues? Will the parents of a child with leukemia be able to order stem cells engineered to that child's needs? Will parents be able to conceive children through IVF that will produce organs genetically engineered for transplant? Will we see a day when a couple, or a mother, could sell such organs on the open market? The National Organ Transplantation Act of 1984 prohibits the sale of organs for transplant. How will this law be enforced?

Consent has been necessary for organ donation in the past, either of the subject or of the immediate family. What of embryos, especially those conceived for purposes of research or organ creation?

The movie *Gattaca* envisions a world in which life begins in two ways: *in utero*, through natural means, and *in vitro*, through genetic engineering. Those who are engineered become superior members of society; those who are not are called "in-valids," and perform custodial work and other duties. Is this our future?

### **Precision medicine after conception**

Today you can send a swab of your saliva to "23andMe," a company that performs genetic testing on DNA samples. For a fee of \$99, you will receive in return a report detailing your risk factors for up to 97 diseases.

In the future, we will be able to diagnose thousands of diseases based on their genetic markers. And we will be able to do so immediately after conception, *in utero*. Doctors will be able to tell parents if their unborn child has a tendency to obesity or diabetes, so they can regulate diet and exercise. And we can start regimens *in utero* or immediately after birth as needed.

Currently, newborn babies are blood tested for a variety of inheritable diseases. Among them is PKU, an enzyme disorder that leads to severe retardation. If a PKU infant is placed immediately on a diet very low in phenylalanine, such an outcome can be prevented.

However, such capacities bring enormous ethical challenges. Will advances in neonatal genetic testing motivate even more abortions? For instance, testing for fragile X syndrome, the second most common cause of mental retardation, could lead to even more elective terminations of pregnancy.

If a fetus is found to have physical problems that can be affected by the mother's health, will she be required to behave in ways that will not prove harmful to the child? Will the child be able to sue her if she does not? Will health care providers face litigation if they do not offer such diagnostic and treatment services, or if they do not produce the desired results? When parents can discover nearly everything about their unborn child, will abortion rates skyrocket?

And consider the life of the individual vs. the good of the whole. If genetic testing reveals that a baby would place a great strain on the economic and health care systems of the community, will there be increased pressure on the parents to abort?

### **Precision medicine after birth**

Precision medicine offers at least three very exciting possibilities for the treatment of disease after birth.

The first relates to medication. The fifth-leading cause of death in America is adverse reaction to medication. With the advances made by precision medicine, we can eventually eliminate this issue, as we target the specific genetic nature of the disease and prescribe medicines appropriate to it.

And we will be able to help patients continue with their regimen. After two years, patients with a genetic diagnosis have shown more than 86 percent adherence to their treatment regimen, compared to 38 percent prior to testing.

Second, conventional treatments will be enhanced greatly. For example, there are five types of colon cancer and at least 10 different categories of breast cancer; what works for one does not necessarily work for another.

Third, genetic treatments are in the future. We may be able to substitute healthy genes for disease-causing genes, treating the cancer or other disease in new and much more effective ways.

However, ethical dilemmas exist here as well. When genetic testing reveals diseases that will begin later in life, how are we to respond? Gaucher's Disease Type I is typically diagnosed at age 28. How are we to handle such information when it becomes known at birth? If I knew that I would develop Lou Gehrig's Disease in three years, leading to a terrible death and horrible burdens for my family, how might I respond? Will we see suicides and assisted suicides escalate?

Will medical advances make euthanasia easier to experience and practice? Will there be pressure brought to bear by families or society? In China, which enforced one-child policies for decades, will euthanasia become a reality based on genetic predictors?

### **General ethical issues**

Here are other ethical issues raised more generally by precision medicine. The first is *eugenics*. Will precision medicine make possible the creation of an "evolved" species?

Plato wanted the most intelligent children to be separated from family and raised as philosopher-rulers. Will we be able to select for such dominant leaders? Will precision medicine do the work of natural selection? Will it be used to increase the intelligence, capacity, and health of our species? If natural selection "improves" every species, will genetic testing improve ours? We believe medicine to be God's gift to us in stewarding his creation. Where is the balance between working with God and becoming God?

*Personal ethics vs. communal law*: to what degree are these issues the subject of the state? Many in the pro-choice camp oppose abortion personally, but do not want the state making such decisions for the mother. Will this thinking prevail with precision medicine? Where and how?

*Privacy issues*: who owns your tissue? Who should have access to your genotype results? How should that information be used? How does this issue relate to insurance, employability, and reproductive decisions?

*Discrimination*: Will genetic testing lead to discrimination by employers and insurers? In May of 2008, President George W. Bush signed a bill which makes it illegal for employers and insurers to discriminate on the basis of genetic information. The Genetic Information Non-Discrimination Act (GINA) will encourage the participation of patients in genetic testing and the use of genetic information in medical records. Will this be enforced fully? How will it be expanded to respond to new advances?

*Medical purpose*: How do we integrate precision medicine within the larger sphere of medical ethics? We want a "seamless garment" strategy vs. a piecemeal, random approach.

Aristotle argued that everything exists from four causes: the material itself, its form (its pattern or shape), its efficient cause (the agent molding it), and its final cause (its

purpose). For example, clay is molded by the potter into a bowl, which exists as a vessel for eating. We must know #4, the purpose, before we can use #1, the material, effectively. So with precision medicine—what is our purpose? Is it the cessation of illness, the enhancement of life, or the evolution of the species? The answer to this critical question will determine the techniques we authorize.

*Access:* Who has access to genetic testing? At present, Medicare does not cover tests "except as explicitly authorized by statute." Outdated current procedural terminology (CPT) codes must be updated in light of genetic testing. Services provided to interpret genetic tests are often not reimbursable. However, Congress has taken steps in recent years to reform Medicare reimbursement policies in light of the value of molecular diagnostic tests. And several large private insurers have instituted progressive coverage policies for certain genetic tests.

*Clinical decision support systems:* What results should be disclosed to the patient? To family, employer, insurer? In what manner?

*Health care provider issues:* How will faith-based health care providers respond to the opportunities and challenges of precision medicine? Will abortion prohibitions be revised? Will embryonic stem cell research be more difficult to resist, especially as enormous sums of money are involved? Will individual practitioners be able to follow their moral principles when they conflict with their employer or patient?

## **Conclusion**

Winston Churchill said in 1942: "This is not the end. It is not even the beginning of the end. But it is, perhaps, the end of the beginning." This is where we are with precision medicine.

The human story suggests that these challenges will increase, not decrease, in the years ahead. Human history teaches two incontrovertible truths in this regard. One: we will do nearly anything to keep loved ones alive. Even those who are most strongly committed to biblical ethics are challenged to compromise their beliefs when life is at stake. Two: if someone can make money doing something, someone will. It is vital that we build an ethical framework now, before temptations and pressures increase in coming years.

If we view all of life as part of God's creation and Kingdom, we will make ethical decisions based on his revealed truth and will for our lives. We will protect life from conception to death. We will enhance health but not engineer it. We will serve society, not seek to evolve it. We will be stewards of creation, not its master.

Solomon claimed that "the fear of the Lord is the beginning of wisdom" (Proverbs 9:10). With precision medicine, how wise will we be?