THE CENTER FOR BIOETHICS AND HUMAN DIGNITY

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# DIGNITY

THE NEWSLETTER OF THE CENTER FOR BIOETHICS AND HUMAN DIGNITY

*Christian perspectives on bioethical challenges such as end-of-life care, euthanasia, genetic and reproductive technologies, and the changing face of health care.*

## REFLECTIONS FROM THE DIRECTOR OF THE NATIONAL HUMAN GENOME RESEARCH INSTITUTE

Francis S. Collins, M.D., Ph.D. *(Bethesda, MD)*

The time is ripe for a serious discussion about the pathway that genetic science is leading us down. It is a pathway that I as a physician and as a Christian have a great deal of hope for because of its promise for alleviating human suffering – which is surely one of our strongest mandates. However, it is also a pathway which poses certain troubling risks. Such risks are real possibilities that we must attempt to address effectively. When we have done so, we can move this exciting field forward in a way that maximizes the benefits and minimizes the risks. In studying the genes that we carry, we are trying to learn the "parts list" for human biology. Historically and philosophically, this is profound. Our human biological instruction book allows us to do all the biological activities we carry out from the time that we are single-celled embryos until the end of our lives. It is an exciting notion that we now have this instruction book in front of us and are able to read it, even if we don't understand it very well.

To understand hereditary factors, we must understand the wonderful molecule called DNA, the double helical structure of which Watson and Crick figured out some forty-seven years ago. It is a very elegant system of encoding information. What a privilege it is for a physician-scientist to do research that uncovers something about our creation and gives us a glimpse into the elegant way God thinks! DNA is certainly a remarkable way of coding information in a very efficient, elegant and digital fashion, allowing us to carry around an enormous amount of information in a very modest space.

In each individual's genetic code, there are three billion base pairs, where each base can be either adenine (A), cytosine (C), guanine (G) or thymine (T). A always pairs with T and G always pairs with C. The four possible choices for each one of these three billion positions yields a huge potential coding capacity. In light of all the things that we have to do as human beings three billion base pairs may not seem like quite enough, but this number must be sufficient. Astonishingly, much of our DNA doesn't have an obvious function, as an estimated 70% or more does not contain genes and just seems to be along for the ride. However, we will never know which part of the genome is the functioning part without studying all of it, and this justifies the broad, all-encompassing approach to the Human Genome Project.

Reading out the sequence of the human genetic code – a feat which has been nearly accomplished – is not, of course, the end of the story. This is really just the end of the beginning. The interesting and challenging part is figuring out what it all means. These three billion letters seem to make up somewhere in the neighborhood of thirty thousand genes, each gene being a packet of information that conveys a certain instruction. The fact that we are not even sure what the actual number of genes is, even though we have most of the sequence in front of us, reveals just how hard it is to stare at page after page of A, C, G and T and figure out what it is telling us. If we printed out the whole sequence and stacked the pages on top of each other, these three billion letters would

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## PALLIATIVE SEDATION: MAY WE SLEEP BEFORE WE DIE?

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Sedation is a clinically important therapeutic intervention in the imminently dying patient. As the patient with an advanced, irreversible illness nears the end of life, symptoms accumulate that are progressively more difficult to manage and that may become unresponsive to standard medical interventions. The most common of these intractable symptoms are pain, agitated delirium, dyspnea and existential or psychological distress. Although sedation is a risk-laden procedure, it is sometimes necessary and maintains the physician's twin obligations to benefit patients and to "do no harm" when practiced by trained, dedicated clinicians.

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be as high as the Washington monument. So we have a very large book written in a language that we don't yet understand very well.

One of the ways in which we are making headway is by looking at human variation. Human beings are quite similar in terms of genetics. If we compared any two people's DNA sequences base by base, we might encounter a difference about every one thousand letters (where one person may have a C and the other may have a T). The other 999 letters would be the same. The two people's DNA would thus be 99.9% identical, regardless of ethnicity or population background. Nevertheless, that 0.1% would still mean that there are many variations between these people's genomes. Most of these variations probably fall in parts of the genome that aren't doing very much, so these differences don't have consequences. The numbers of common variants that actually affect function may be as small as a couple of hundred thousand, many of which have rather mild effects. On the other hand, a few of these will fall into those vulnerable parts of the genome that can put people at risk for diabetes or Alzheimer's disease. Those are the ones that we are most interested in uncovering.



How are discoveries about the human genome going to affect medical care? Information gained from the Human Genome Project will help us develop diagnostic tools to detect whether a given person has a genetic predisposition for a particular disease. We already have such tests for breast cancer, colon cancer and Alzheimer's disease. This list is going to grow quickly. However, is the availability of such tests necessarily a good thing, particularly if there is as yet no intervention to treat the disease in question? On the other hand, if an intervention is already available for a certain disease, then knowledge gained from genetic testing might be very desirable because it would allow us to do something to decrease our chances of getting a terrible disease. We are already getting close to this level of knowledge for some conditions such as colon cancer. Diagnostics are most useful if they are associated with a preventive medical strategy that people can undertake to reduce their risk.

What people are most excited about, however, is our potential ability not only to make predictions but also to actually discover cures, so that if people fall through the prevention safety net, something will still be there to catch them. Gene therapy gets much attention in this regard, because it is radically different from other more traditional approaches to medicine. In its simplest form, gene therapy involves transferring a normal copy of the gene that is not working to the tissue where its action is needed. For cystic fibrosis, for example, transferring a normal copy of the gene to the lungs of an affected young person might be expected to provide considerable benefit if the gene could be transferred efficiently to a high proportion of the cells in the lung and if it was properly regulated. Unfortunately, this approach has not yet shown much in the way of concrete clinical benefits and is not free of risks. The death of Jesse Gelsinger during a 1999 gene therapy trial reverberated through the scientific community and the public at large. It was the end of our innocence about gene therapy. I believe that we should definitely continue to pursue gene therapy at the basic science level with maximum intensity because it does have a lot of promise; however, we also have to be realistic in recognizing that some of the scenarios portraying gene therapy as a panacea have been ahead of actual reality.

Certain questions have to be addressed if we are going to see a beneficial outcome of genetic research for the health of all. We have to be sure that misuses of this whole set of advances do not eclipse the benefits. If we are not vigilant, something that should have been a wonderful revolution will be turned into something harmful. There are a number of potential worries here, and it would benefit all of us involved in this discussion to keep Proverbs 19:2 in mind: "It is not good to have zeal without knowledge, nor to be hasty and miss the way." Will we prevent people's genes from being used against them? When is the right time for a genetic test to leave the research lab and move into clinical practice? Will medicine and science take the initiative in this decision, or will it be driven solely by the marketplace? Is the general public ready to incorporate genetic information into its medical care? The prospects for genetic medicine are complicated by the fact that access to health care is not universal. Zeal for doing God's will and for a good outcome need to be combined with a clear commitment to understanding all of the intricacies of these issues.

Given the complex concerns raised by genetic research, some have asked why we are doing this at all. The New Testament book of Matthew serves as a powerful reminder of how much time Christ spent healing people in His very short time on this earth: "Jesus went through all the towns and villages, teaching in their synagogues, preaching the good news of the kingdom and healing every disease and sickness" (Matthew 9:35). Perhaps because they are called to be Christ-like, Christians feel a particular responsibility for reaching out and healing the sick. That is one of the reasons why studying this aspect of our biology and trying to apply it medically is not merely a good idea, but a moral necessity. It is an ethical requirement of us. If we can develop the ability to heal, if genetic research holds out hope and promise and can prevent suffering in our fellow human beings, then we have to do it. However, we must also shoulder the responsibility of making sure that these powerful tools are used for good purposes and not for unethical ones. ■