

**An**

**Average Man's "Discovery"**

**of the Human Genome**

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“Scientists marked what they called a milestone on a truly unprecedented voyage of discovery, as ambitious as landing on the moon or building the atomic bomb.”<sup>1</sup> The words blared from the television as I just entered the house from a hard day’s work. My wife, Annie, was sitting on the sofa watching “World News Tonight,” as was her daily ritual. She averted her eyes from television towards the door upon hearing the sound of my return.

I asked, “What’s this great discovery Peter Jennings is blabbing on about?”

Annie replied, “Oh, some thing called the ‘human genome.’ Francis Collins, the director of the National Human Genome experiment announced that they’re celebrating the revelation of the first draft of the book of life.”

“The book of life? Oh please, tell her to spare the American public this poetic mumbo-jumbo. There’s *always* a new discovery, something that promises to revolutionize the world. Who cares about the human genome? Those darn scientists! They think they’re so important.”

The news report continued, “The map is simple looking, but it represents the chemical code that controls how the human body is built and how it operates. It is profound. The human genome is also long. If it were printed in the newspaper, it is a 3.2 billion letter sentence that would take 100,000 pages.”<sup>1</sup>

“Oh great, they just discovered we’re a big, fat sentence! How exciting!” I remarked sarcastically.

Annie interjected, “C’mon, honey. I really think this map will seriously make a difference. They’re talking about pinpointing genes that cause cancer or disease. That means they will be able to give genetic tests and determine whether a person has that gene. If we know how these genes affect us, there’s a great deal that can be done.”

The news report went on, “This map marks the beginning of an era of discovery that will affect the lives of every human being.”<sup>1</sup>

“Oh, give me a break!” I exclaimed as I clicked the television off.

I was never really a fanatic of science. Or, more accurately, I abhorred admitting that I did not fully understand it or its implications. My high school biology teacher once told me I was hopeless and that I was mentally incapable of grasping even the most basic of scientific ideas. My response to that kind of constructive criticism was to give up. As a result, I have doubted and largely ignored any sort of “great scientific achievement” they mention on the news. However, that night felt a little different. Annie was convinced of the significance of this “human genome” thing. Was I wrong to discount it so quickly? I somehow felt haunted by my skepticism. What impact did this new information have? Would it make things better? Would it make things worse? These questions continued to tease me as time went by. . .

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“Tonight is ‘Guess That Theme’ night,” Annie said as she led me to the dining room table one night. She continued, “Tonight, monsieur, we will be dining upon honey glazed baby carrots, sauteed baby corn, and some juicy barbecued baby back ribs.”

I piped up excitedly, “Ooh! Ooh! I get it! Fingerfood Night!!”

Annie cocked her head to the side and replied, “Well, ummm, not exactly...”

“Um, um, um...is this some sort of new diet? Like no carbs?”

“Hmm, let me try again. We’re having baby carrots, BABY corn, and BA-BY ribs.”

“Baby?”

“Yes!”

“Wait, I don’t get it...baby? Baby?! BABY!! We’re having a baby?!” Annie nodded vigorously.

I screamed, “We’re having a baby!!!!!!!!”

Annie and I hugged each other and leaped with joy. Then Annie said, “I just found out today at Dr. Peterson’s office. I’m so thrilled! Oh, and she suggested that we go see a genetic counselor. She referred us to a very highly recommended...”

“Genetic counselor?”

“Yes. Dr. Peterson said that many primary care physicians such as herself weren’t best prepared to offer any genetic counseling, what with the new developments in genetic technology and all.”<sup>2</sup>

“Why exactly do we need a genetic counselor?”

“Well, according to Dr. Peterson, newborn screening for genetic disease is a relatively routine pediatric practice. And now, the Human Genome Project is opening a wealth of genetic information at an incredible rate.<sup>2</sup> Please, honey, I think it’ll be very worthwhile to meet him at least. And besides, we won’t be seeing him until later in the pregnancy.”

Two months later, we were scheduled to meet with the genetic counselor. I was curious as to what he would tell us. I did not put too much stock in the whole genetic counseling thing. Still, it would be interesting to see what “science” could tell us about our baby.

When we met the genetic counselor, he was not the awkward, geeky, scientist I was expecting. Instead, he was warm and friendly. I could tell that his kind manner appealed to Annie. He sat us down and began explaining his role. He began by describing the Human Genome Project and how with the mapping of the genome, incredible advances have been made.

Knowledge of the genetic basis of disease and the number of genetic tests available had increased considerably.

“Technologies for studying gene expression, which allow analysis of transcription of as many as 10,000 genes in one experiment, make it possible to investigate the differences between normal tissue types and the variations in that expression pattern during disease.<sup>3</sup> Tests for genetic variations linked to some 400 diseases now exist, and is still on the rise.”<sup>4</sup>

Was this the same Human Genome I had heard on the news?

The counselor continued, “There are various forms of genetic testing and it is important to note the differences. One is testing someone who is symptomatic, another is testing for a 100% penetrant gene that has not yet expressed itself, and yet another is testing to determine increased susceptibility to a particular disease.<sup>2</sup> In the latter two cases, a positive test result does not necessarily predict whether an individual will be affected by that disease, and it cannot predict at what age the disease will onset or how extreme the disease will be.”

I interjected, “Well, is there any reason why we would want our child to have a genetic test if the results give no definitive results?”

“Not all tests give such results. For instance, retinoblastoma is a genetic disease that is almost 100% penetrant. While most genes associated with genetic tests identify only increased susceptibility, 100% penetrance indicates that having the gene means ultimately having the disease. Even with genetic tests that can only identify susceptibility as opposed to penetrance, there are medical benefits in the form of preventative measures that may be available: lifestyle modifications, diet, drug therapy, etc.<sup>2</sup> With retinoblastoma, there are such treatments, but with other diseases like Huntington’s Disease, there is neither prevention nor cure.”

Annie, clearly more knowledgeable than I, asked, “But there are so many genetic diseases. How do we know what to test?”

“Well, in the least, your child will be genetically screened for certain diseases regardless of your consent. It sounds harsh but it’s current policy unless you live in Maryland.<sup>2</sup> Genetic screening refers to testing whole populations for diseases. For instance, in the United States, there is almost universal screening for phenylketonuria, hemoglobinopathies, and galactosemia...”<sup>5</sup>

I had never heard of any of these.

“...In order for a disease to fall under that category of genetic screening, there are several criteria that the Institute of Medicine recommends be fulfilled: 1) that there is a clear indication of benefit to the newborn, 2) that a system is in place to confirm the diagnosis, 3) that treatment and follow-up are available for affected newborns, 4) that the condition is frequent and severe enough to be a public health concern, 5) that the condition causes a known spectrum of symptoms, and 6) that the screening test is simple and reliable, with relatively low false-positive and false-negative rates.”<sup>6</sup>

“So should our child be tested for anything else then?”

“If you decide to, your child could be tested for any particular condition in which there is a strong family history.”<sup>3</sup>

I was getting a little concerned. “What happens if that disease has no preventative treatment?”

“That is a very prevalent concern in which there are disadvantages and advantages. Perhaps through knowing the results of a particular test, the individual becomes allowed to incorporate positive or negative status as part of his or her self-concept and allowed to better plan

his or her life.<sup>2</sup> The drawback in that lies in the possibility that the disease might never express itself. But, regardless of whether the disease is onset, it is important to note that there are many social and psychological repercussions in getting tested. We ought to discuss these before proceeding.”

The counselor explained that there was risk of discrimination because of the genetic abnormality even if it was not 100% penetrant. There were also issues of privacy of genetic information and who has a right to it. He said, “As a society, we are only in the early stages of understanding the relationship of genotype-phenotype correlation and the interaction of various genotypes with the environment. But this has not stopped different institutions from using this information in a discriminatory manner.”<sup>7,8</sup>

He continued to explain that other risks included stigmatization associated with the genetic abnormality and survivor guilt on the part of the parents or unaffected siblings.<sup>2</sup> There was also the risk of parents viewing their child as ill or even doomed and, as a result, bonding poorly or otherwise neglecting the child.<sup>2</sup> Was this guy suggesting that we could possibly not be as loving of parents as we could be if our child was not predisposed for disease?

“What do most parents do when they receive a positive test result?” I asked worriedly. “Well, it really depends on whether there is a form of treatment out there for the disease. For the most part, it is extremely difficult to quantify human reaction.”<sup>2</sup>

I left the counselor’s office feeling completely overwhelmed. The abundance of information and the onslaught of implications swirled in my head incessantly. Suddenly, everything somehow became an issue of genetics. I recalled that my Uncle Harold told me the other day that he had applied for a job at Rockwell International to do some assembly-line work

but was rejected because of a disability he did not have.<sup>9</sup> Rockwell's reasoning was that he had failed a nerve conduction test that detects predisposition for carpal tunnel syndrome. He, along with seventy-one others who had their job offers withdrawn, were suing Rockwell. It was the first time I had ever heard of someone not being hired because there was the *possibility* they would develop a disease or disorder later. Can a predisposition really be considered a disability?

I began thinking about how I had been looking for a new job. Would a potential employer want to screen me for possible disabilities? What if they asked me to take a genetic test? What if they found that I was predisposed to having a certain genetic disease, just like the counselor was explaining? Will they not hire me because of that? Would the fact that I was competent and hard-working compensate for a disease that may never manifest itself? My horrific conclusion was that perhaps it would not. More than anything, companies would be concerned about lowering health care and worker compensation costs.<sup>10</sup>

What if I wanted to buy my own health coverage? Would insurance companies reject me based upon genetic factors that predict future illness without causing substantial present symptoms?<sup>11</sup> What happens to my child if she gets tested? Will she be discriminated genetically for the rest of her life? As a parent, will I love her less and distance myself from her like the counselor had mentioned? Will I feel guilty for bringing her into such a life? Will my child feel guilty for being abnormal or for being a burden?

It was not until the visit with the counselor that I realized how pervasive and important the mapping the human genome actually was. It was science with a genuine impact. It was not simply just a big, long sentence to which humans could be reduced. There were serious implications to the abundance of knowledge that that one sentence carried. While science did not

have the answers to everything, it brought me closer to making a better decision as to what was best for me, my family, and especially my unborn child. The human genome was not as intangible as it seemed. In fact, its reach spans every human being including me, average American.

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