

The Human Genome as Metaphor

Howard F. Stein, PhD

Ever since high school physics class, I have been fascinated by Wilson Cloud chambers, in which cosmic rays are detected by their tracks in water vapor. The interstellar visitor itself passes unseen; it can only be inferred from its trail. Until now, genes have behaved much like cosmic rays, ever present but for the most part both silent and invisible. This will soon come to an abrupt end as a result of the discoveries of the Human Genome Project, as described by Dr Lori Whittaker in this issue of the Journal.¹ The Human Genome Project is both science and metaphor. It will create a track in dense social vapor. It will confront us with our own assumptions about what it is to be human, to be ill, to be perfect, to be monster, and to be wise. The Human Genome Project is culture-saturated science.

Are We Wise Enough to Reinvent Ourselves?

From an evolutionary viewpoint, genetic variety and variability ("hybrid vigor," as it was once called) have enhanced the long-term adaptability of the human species to an ever-changing physical and social environment. Genetic manipulation of specific cultural traits might well lead to evolutionary dead ends, even though—from the viewpoint of a particular society's aesthetic or political preferences—it may be beneficial for a time. Are we prescient and omniscient enough to know what genes we will need to survive in the future? Perhaps those that will have the greatest value in the future are those that we are quick to brand unredeemably deleterious in the present.

Americans value, and thrive on, short-term solutions: 10-day antibiotic regimens, quarterly financial reports, 4-year presidential terms. When we extol "results" and "outcomes," we usually mean something we can measure immediately. If it is so difficult for us to commit to long-range planning in these more tangible areas, it is

unlikely that we will be able to plan within the scope of man's history, much less that of evolutionary biology. Yet, this is precisely what will be required as we implement the Human Genome Project through genetic counseling. Genetic technology will allow (demand) us to act in ways unprecedented in human history. It will allow us literally to re-invent ourselves, to give us once again a new start, a new frontier.

It is very American to want to tinker with, to fix, to improve everything. "Futility" is so heated an issue in biomedical ethics because it is attitudinally un-American. Only technological success, our yardstick of progress, will do. But *should* we do all that we *can* do? Within the American results-oriented ethic, genetics could become yet another industrial and corporate "product line." Margin of profit rather than impact on human history might become genetic engineering's social measure.

All human decision-making, including clinical and scientific, is ambivalence ridden; not ruled entirely by reason, not based solely on reality, and not protective invariably of enlightened self-interests.²⁻⁴ We know both more and far less than we realize. Will there be room for humility, for uncertainty, for chance, in medicine and life? Do we know enough?

Dr Victor Frankenstein and the Perfection of the Human Form

Ellen Golub writes of the abiding popularity of Mary Shelley's 1818 novel *Frankenstein*:

Frankenstein, a modern story if ever there was one, touches us deeply because Victor Frankenstein's wish meshes with our own ambition. We fear that Dr. Frankenstein's hubris is ours; moreover, we find ourselves in the disquieting transformation into monster. In this century we have conflated humankind with technology. Out of a desire to "renew life where death had apparently devoted the body to corruption," we have used "extraordinary means" to save our terminally ill.^{5(p725)}

With genetic screening, counseling, and engineering, we will have the ability to alter life before birth. This technical prowess arouses feelings and fantasies that will

From the Department of Family Medicine, Health Sciences Center, University of Oklahoma. Send requests for reprints to Howard F. Stein, PhD, Department of Family Medicine, University of Oklahoma Health Sciences Center, Oklahoma City, OK 73190.

soon become part of our reality. Dr Victor Frankenstein "himself" said,

Life and death appeared to me ideal bounds, which I should first break through, and pour a torrent of light into our dark world. A new species would bless me as its creator and source; many happy and excellent natures would owe their being to me. No father could claim the gratitude of his child so complete as I could deserve theirs.^{6(p55)}

When a newspaper headline reads "Scientists Link Gene to Disease,"⁷ we need to be reminded that we are in the realm of magic, myth, and religion as well as of science. In secular gown, the practitioners and technicians of biotechnology are both god and devil, omnibenevolent and terrorizingly malevolent, priest and witch. In our fantasies of controlling genes and genetic potential, our unconscious anxieties, unspeakable wishes over life and death, our role as creator and created, as father-mother-child, are expressed. Through genetic engineering, we may literally fulfill the wish to be self-made men and women.

Abraham J. Heschel wrote, "We not only describe the 'nature' of man [people], we fashion it. We become what we think of ourselves."⁸ We are what we imagine, then make ourselves to be. Theory and method are inseparable; what we do flows from who and what we think we are. The Human Genome Project will empower us to become what we believe we should be. In this process, many versions of what it means to be human will collide in the physician's office as well as in public policy making.

The Human Genome Project and the Definition of Disease

Joan Ablon writes that "few of us are aware of the potential challenges posed by our genetic inheritance. Yet every individual and every family is at risk for genetic disorders whose expressions are found in every age group."^{9(p10)} One little-explored psychosocial implication of genetic disorders is that, in contrast to the popularly held view that genetic disorders are a special affliction of "them," from which condition the healthy "us" are exempt, we all are in fact "them." Yet, the splitting of conditions of health into "normal" and "abnormal" dominates our thinking. Culturally, one purpose that diagnosis serves is to confirm that the diagnostician is not ill.

All known genetic disorders are not socially equal, and even what constitutes a "disorder" (itself a social construct) will need to be redefined. Some disorders have historically drawn great compassion (Tay-Sachs), others ostracism (sickle-cell anemia), and still others harbor special, unspeakable terrors about an unknown future (Huntington's disease).

In the search for the perfect baby, who is to qualify as "expert" on defining and identifying perfection? What is perfect, and according to what shifting cultural standard? Who will be authorized to choose what "disorders" to keep or eliminate? Who will enforce and execute these standards? To what extent should we permit control over our genetic products, such as parents' choice of their baby's sex? In a still patriarchal society, many parents will wish to have a boy. Does femaleness then become a disorder? In a society that values size (where everything bigger is regarded as better), will short stature be a disease?

Genetics often enters clinical cases in an offhand, even folksy way. For example, I have participated in case conferences in which physicians discussed the treatment of insulin-dependent diabetic children. Although the patient was treated "aggressively," still the question was raised about the advisability of prolonging a "negative gene" such as diabetes in the wider population pool by treating the patient long enough that he or she could grow to adulthood and procreate. How advantageous was it, they wondered, to perpetuate this gene? What genes should we eliminate to create a more "healthy" population? Today we define coronary artery disease by angiogram. What will happen when an infant can be diagnosed as having the CAD gene? If anatomy is destiny, is genetics even more so?

Diversity and Purity: Single- and Multi-Gene Images of the Human Condition

Will American culture domesticate the gene, or will the gene transcend its culture and transform the way that we think as well? This will depend partly on the scientific discoveries themselves. Can the qualities of an individual person be characterized by the simple listing of DNA pairs, or will we learn that personhood is characterized by an exceedingly complex multi-gene interaction that defies understanding in terms of elementary biological particles? If the Human Genome Project goes the former way, our Aristotelian and Cartesian world view will seem confirmed. Like everything else, we will press genetics to become a matter of either/or and black/white, and we will define genes simply as the tiny, invisible machinery of life. If the Project takes the other direction, then the discovery of polygenetic interaction, the tremendous complexity of what we had expected and hoped to pare down to cultural "size," will require that we rethink our biopsychosocial world view. We could come to perceive life with a new innocence and with wonder.

Genes, Metaphor, and Physicians

Genetic knowledge does not occur in a social vacuum. The scientific account is neither the only story, nor the entire story. Decisions to know or not to know, to have children or not to have children, to label as diseased or not, are part of wider life histories, language, and group fantasies. The Human Genome Project and its unprecedented opportunity for intervention in our own genetic structure will confront us with complex questions that defy neatly packaged answers. Physicians will need to serve as mediators, translators, and guides to help patients and families deal in new ways with their lives. Physicians will need to understand the meaning, in human terms, of the nucleotide pairs that we can now so meticulously define. To assume this role, physicians will need to understand the gene as metaphor.

References

1. Whittaker LA. The implications of the human genome project for family practice. *J Fam Pract* 1992; 35:294-301.
2. Balint M. *The doctor, his patient and the illness*. New York: International Universities Press, 1972.
3. Stein HF. *The psychodynamics of medical practice*. Los Angeles: University of California Press, 1985.
4. Stein HF, Mold JW. Stress, anxiety and cascades in clinical decision-making. *Stress Med* 1988; 4:41-8.
5. Golub E. Cancer and death in the Promethean age. *J Popular Culture* 1981; 14:725-31.
6. Shelley MW. *Frankenstein: or the modern Prometheus*. New York: Pocket Books, 1974 (orig 1818):55.
7. Associated Press. Scientists link gene to disease. *The Daily Oklahoman* 1992 June 2, p 8 [Charcot-Marie-Tooth neurological disease].
8. Heschel AJ. *Who is man?* Stanford, Calif: Stanford University Press, 1965:7.
9. Ablon J. Social dimensions of genetic disorders. *Pract Anthropol* 1992; 14:10-13.

See article on page 294.
