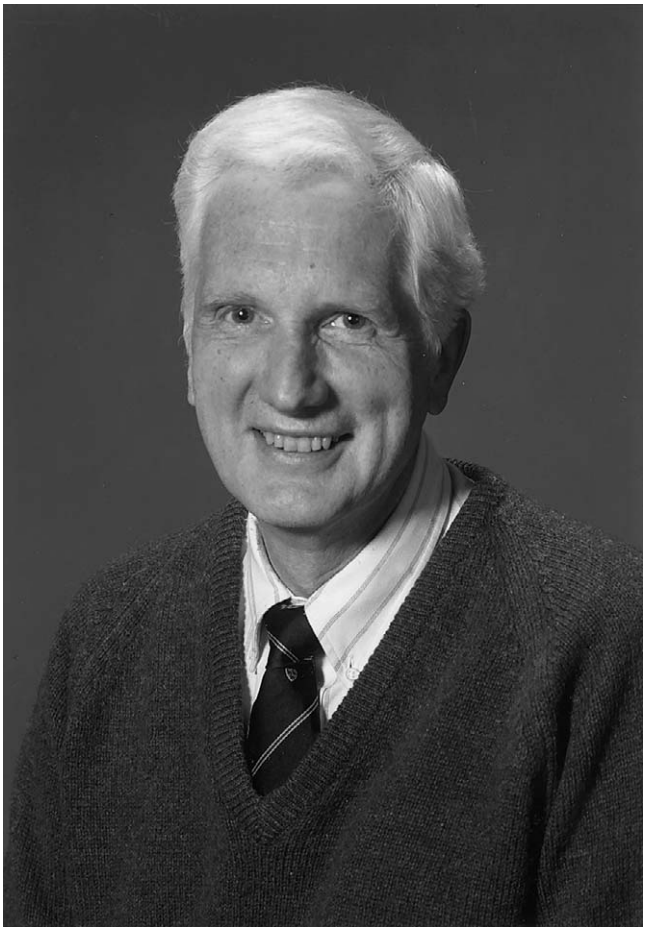
Am. J. Hum. Genet. 70:317–323, 2002

2001 ASHG AWARD FOR EXCELLENCE IN EDUCATION … And Know the Place for the First Time*

Charles R. Scriver



Charles R. Scriver

This is an occasion to celebrate excellence. But first I must recognize the apocalyptic events of September 11, 2001—an occasion for despair and failure. Despair on that bleak day when human evolution took another false step. Failure, if we respond in kind and repeat a cycle of useless violence; greater failure if we learn nothing from the useless violence of the 20th century or from continuing violence in the Middle East, the Balkans, and Northern Ireland, and a hundred other places. Bleaker still that an intelligent species cannot do anything but repeat collective bleak experiments in violent behavior. Would an intelligent person repeat a failed experiment over and over again—and expect the grant to be renewed? Yet we renew the funds, again and again, for the experiments in violence. Why? The answer eludes me, so I will turn to celebrate better things.

Received October 24, 2001; accepted for publication November 28, 2001; electronically published January 4, 2002.

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* Previously presented at the annual meeting of The American Society of Human Genetics, in San Diego, on October 16, 2001.

0002-9297/2002/7002-0006$15.00

Gratitude

Gratitude at receiving this award is a much better feeling. I recognize that you also honor my colleagues, without whom I would not be standing here, and I am grateful for the effort that went into the nomination by those who so generously prepared and supported it. The Selection Committee was kind to me, but undoubtedly there were other deserving candidates, and I acknowledge their own excellence. And then there is the American Society for Human Genetics itself, which honors and celebrates excellence in education, the vehicle that can bring the awareness to all persons that we are both individuals and connected to all life and are called upon to be respectful of all life.

My own human genetics education was enhanced some 35 years earlier by our daughter, who was still a child but very aware that genetics was something of interest; she gave me a poem (illustrated) on the topic for my birthday (fig. 1). Whatever one may think now about the technical quality of the poem and its depiction of complex traits, I have been touched by the message, both then and now. Out of a child's mouth came a truth.

Education

On education: The word *education* has a Latin root in the verb *educere,* meaning "to lead out"; how different is its meaning from the word *training,* rooted in the Latin verb *trahere,* meaning to pull or draw out. The New Shorter Oxford English Dictionary has several definitions for the English word, of which two are relevant here: one arising in 17th century Puritan England, implying the "whole course of instruction received by a person," and the other formulated in post-Enlighten-

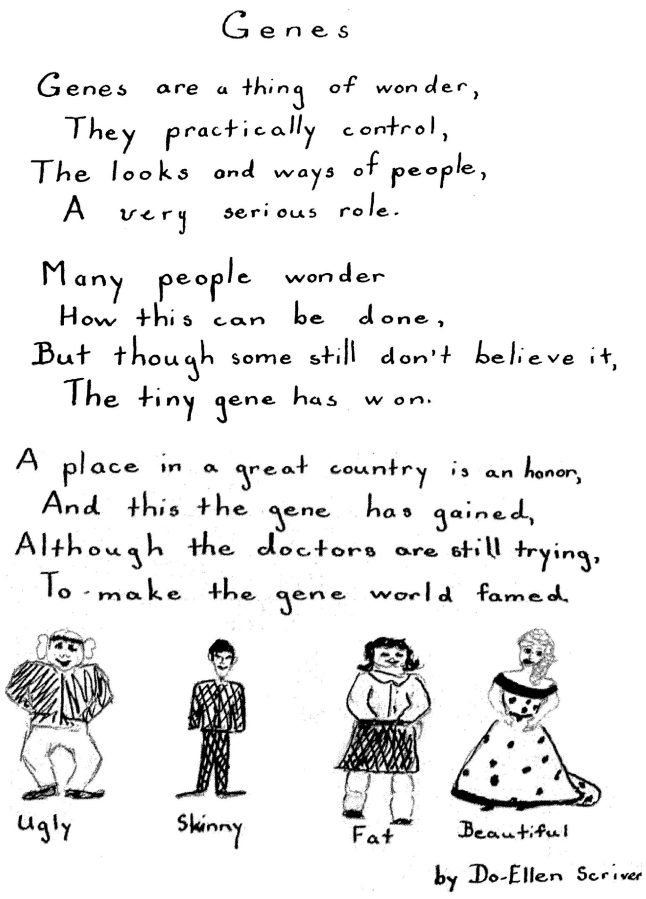

Figure 1 An example of human genetics education in a poem by Dorothy Ellen Scriver given to her father (in the 1960s).

ment 19th century England, describing "the development of mental and physical powers and the moulding of character." Perhaps these definitions inspired James D. Watson to address a school assembly of young women in 1975, in this fashion:

> Because the American public as a whole still believes that a knowledge of science is best left to scientists and most certainly not to young ladies, we remain as a people, dismally ignorant of science and its mode of operation. And so countless decisions, in which the utilization of scientific facts is a key ingredient, are made by persons totally ignorant of its vocabulary, suspicious of its better practitioners, and loathe to take advice for fear that they will be asked to run counter to short term political common sense.

As noted by a reviewer (Postgate 2001) of Watson's book (2000), from which this extract is taken, deletion of the word "American" does not invalidate his message.

So, at its heart, what is science? The best definition I know is clear and simple: "science is an assault on ignorance" (Ridley 1991). Accordingly, it is a natural topic for education. But whose ignorance, and what are the values surrounding science that matter to society? The values differ according to the point of view (table 1): that of the scientist, who claims value-free enquiry, or that of society, which sees value-laden issues in the enquiry and in the process of using the science and its associated technology. Biology and genetics are not exempt from the divergent viewpoints.

I am inclined to believe that the following questions, posed by a poet long ago (Eliot 1963; "Choruses from the Rock"), apply to biology and genetics—all the more so if we are in danger of neglecting our duty to public and personal education in these disciplines.

> Where is the Life we have lost in living?
> Where is the wisdom we have lost in knowledge?
> Where is the knowledge we have lost in information?

Questions of this order take on great significance when *order* in materialistic life, as revealed by biological science, can be united with its spiritual *meaning*, as revealed by other means (and as implied by the poet). Perhaps then *Homo sapiens* will escape extinction in the pursuit of bleak experiments on itself.

Anxiety

Anxiety about the place of science and technology in human affairs is nothing new. Whereas the technologies gave us a productive century of steam (the 19th century) and an even more remarkable century of electricity and electronics (the 20th century), and we have subsequently celebrated their corresponding benefits, citizens were nonetheless deeply worried about the impact of science and technology in those earlier days, and if the 21st century is to be the one of DNA and biotechnology, citizens will once again be very anxious. COSMOS was a recent international exhibition examining the relationships between science and society, and it offered an important cultural perspective in the accompanying book (Clair 1999), where this phrase appears: "After the ex-

Table 1

Two Views of Science: The Viewer, Not the View

Scientist's Science	Citizen's View
"Value-free"	"Value-laden"
Biology (and genetics)	AIDS; eugenics
Physics	Food irradiation
Chemistry	Acid rain; pesticides
Geology	Earthquakes
Forestry	Reye syndrome
Electronics	Air traffic safety
Sociology	Crime rate; immigration
Ethnology	Native peoples' claims

NOTE.—Adapted from Scriver (1993).

altation of natural beauty in the Enlightenment came the sense of apprehension that permeated Romanticism." Replace the words "natural beauty" by "genes," "Enlightenment" by "The Human Genome Project," and "Romanticism" by "the early 21st century," and ask yourself: what is different? Goethe—scientist, artist, and philosopher of the Enlightenment—reflected in *The Travels of Young Werther* on "the immense that ceases to be sublime, surpassing our faculty of comprehension and threatening to annihilate us" (cited by Clair [1999], p. 20). Someone might use those words today about contemporary legacies of science, notably in physics, and one hopes they won't apply to genetics and biology. Collective and personal education that fosters an integration of wisdom, knowledge, and information is a way to reduce the likelihood that they will.

On Frankenstein and Prometheus

A young woman of the early 19th century wrote a story to fulfill a wager taken up with her illustrious partner, Percy Bysshe Shelley, and his friend George Gordon, Lord Byron. Mary Wollstonecraft Shelley was the only one of the three who met the challenge, and, in so doing, the story of *Dr. Frankenstein: or the Modern Prometheus* emerged. (In my talk, I used three of the powerful illustrations by Barry Moser in the Pennyroyal edition of the story [Shelley 1984]). In Mary's version of the Promethean legend, the good doctor used available recombinant methods, and from diverse human parts, rather than from clay, he made his creature. Dr. Frankenstein's creation was supposed to be beautiful, and the whole venture was meant to be truly Promethean in its achievement—but it was NOT! The project went wrong, and the result was unpleasant. Yes, the project went wrong, and the result was—shall we say—unsatisfactory.

Thus, when genetic science states it can recombine or reuse genomes and parts of genomes and make something good and useful, such as a genetically modified organism for food, are we surprised that citizens are worried? They remember Dr. Frankenstein, and they know instinctively that life as they perceive it is being corrupted by humankind when it tinkers with the genomes and life forms created by evolution (or by God). Accordingly, geneticists and biotechnologists are being called upon, with all the wisdom at their disposal, to show that they understand the concerns of others and, indeed, acknowledge them. Education is the tool I would use to deepen the understanding of life, its forms, its connections, and what we are doing when we recombine it.

It is no surprise to me that "genetics," the thing I love and do, is now a word likely to bring a shadow or a look of concern to my neighbors' faces, when, as recently as a decade ago, those same persons thought I was a good person doing wonderful things. As I see it, there is both a call for and a hunger for education in modern society about genetics, and I congratulate the American Society of Human Genetics for taking initiatives to enhance our role in that endeavor. I also congratulate, for example, the Public Broadcasting System for the recent television series on *Evolution,* in which contending scientific and religious views about the origins of species—and of humankind in particular—were honorably exposed.

Culture, Spaces, and Science

Do we recognize and have we identified the cultural spaces where science is actually being defined? They are *not* in a convention center like this one, nor are they in the pages of our *Journal.* They are in the halls of government and in the news media. The people with the power and those in much need of continuing education about genetics are out there—not in here. Controversies will be worked out or not worked out, and policies will be made or not made, ultimately, by government assisted by the media. We can help them in their tasks, and our assistance with their education is one means to do so. I can illustrate this particular theme from personal experience.

Experience

In the 1960s, the province of Quebec was called the infantile-rickets capital of the Western world, because the incidence of this disease was so high in our population. The principal cause was environmental vitamin D deficiency, and its origin was both in the atmospheric conditions (cold winters and low-angle ultraviolet radiation) and in an anomaly of government regulations that, contrary to the actual intent, kept vitamin D out of bottled dairy milk—the major source of nutrition in those days for a large sector of the Quebec infant population. The solution seemed obvious, the relevant government regulation had to be changed, a process that proved to be unexpectedly difficult.

The news media took up the cause accurately and responsibly and they were indeed a helpful "cultural space" from which awareness of the problem was widely disseminated. But nothing changed in the halls of government, and I soon learned that a problem like this must ultimately be worked out there—that is, in a cultural space where the decision would be taken, in the context of either votes or money. Rickets was not an issue rich in votes. However, I learned that a truthful economic argument based on accurate information would help to bring about change. Accordingly, I showed that the potential cost savings in the prevention of vitamin D deficiency rickets would far outweigh the

costs of changing legislation and modifying milk marketing. My argument was a form of education; it prevailed, the legislation was changed, and the marketing of milk was modified. Education had had its effect. During the course of this event, two particular people became better educated: myself, when I learned how government actually works and what it needs to know when making decisions, and the government's minister, who needed a perspective so that he could solve a social problem affecting health and well-being in the society he served.

I discovered something else of a later and particular educational interest. Following a rapid decline in the incidence of environmental rickets (thanks to the changes brought about as I described) heritability of the condition in the population of Quebec rose dramatically. Thereafter, virtually all new patients with infantile rickets, albeit far fewer in number than in earlier days, had a Mendelian disorder of calcium or phosphate ion homeostasis (Scriver and Tenenhouse 1981). I saw this transition in the nature of a particular disease in the population as a paradigm for the change in incidence and heritability of disease, in general, in any society where public health has been improved. I have used the particular Quebec experience ever since as an educational perspective in defending the relevance of putting genetics into health care (Scriver 1988).

My second personal experience with education as a transforming social force was in the selective area of genetic screening, both in the newborn population and among ethnic heterozygous carriers of disease-causing mutations and also in the broader area of genetics in medicine (Scriver et al. 1978). We used top-down and bottom-up educational approaches to create an integrated newborn genetic screening program in Quebec that now reaches into a population of 7 million people through public education, population screening, and resources for diagnosis, counseling, and treatment of a wide variety of genetic diseases (National Academy of Sciences 1975). The public is informed prior to and at the time of screening and is given the option for voluntary participation or withdrawal. The government provides infrastructure and funding for the integrated program. The combined approaches make the project greater than the sum of its parts, reflecting a societal phenotype and cooperative points of view. Mutual education is the effective glue, and the disease consequences of a wide spectrum of early-onset genetic disorders have been altered in Quebec.

My second example in genetic screening again involves the Quebec population, which comprises a substantial portion of immigrants from overseas during the 20th century, among whom are healthy, innocent carriers of mutations causing Tay-Sachs disease and β-thalassemia. The work of Kaback and Cao inspired us to emulate and develop programs for carrier screening and reproductive counseling in the Quebec populations at particular risk.

In pilot studies (Beck et al. 1974), we noted that participation rates were highest among senior high school students. There are many explanations for this finding, and we subsequently received a range of criticisms for even considering that high school students could be the population window for genetic screening of this type; some of the criticism can be found in *Assessing Genetic Risks,* a report from the Institute of Medicine (Andrews et al. 1994). Nonetheless, we pursued our course, because attitudes in our communities were receptive. A top-down approach allowed government to provide resources; rabbis, bishops, and priests, along with school boards, principals, and teachers, spread the word and supported participation. However, nothing would have happened without a bottom-up approach as well. It came from the students themselves (who were introduced to information and participated voluntarily in the screening process), their parents, and the communities who wanted the programs. In the final analysis, the communities perceived they owned the programs, and it was their wish that the latter should exist. The key was communication, shared information, and community education. Twenty years later, the programs for voluntary carrier screening for Tay-Sachs disease and β-thalassemia in Quebec are imbedded in the fabric of community awareness and health care. As reported (Mitchell et al. 1996), normal fertility rates were maintained in the families at risk, while incidence of these two diseases declined by >90% in the communities. For some strange reason, this long standing project in community genetics suddenly became news in Canada's national print and television media in April, 2000. Fortunately, no harm came from our 15 minutes of fame, and the broadcast news appeared to help people to learn that genetics can be good news.

Internet

The citation for this Education Award refers to a pair of books with which I have been involved. They are *Amino Acid Metabolism and its Disorders* (Scriver and Rosenberg 1973) and *The Metabolic and Molecular Bases of Inherited Disease* (MMBID) (Scriver et al. 2001). It would not be right if I did not acknowledge that both books reflect the combined efforts of colleagues and myself. The citation also mentions "the application of electronic communications to genetic education at all levels." Let's begin at the human level to look at these accomplishments.

You are looking at a wedding scene (fig. 2). The photograph (which I took) links books, electronic communication, and people to make a coherent story. Irene,

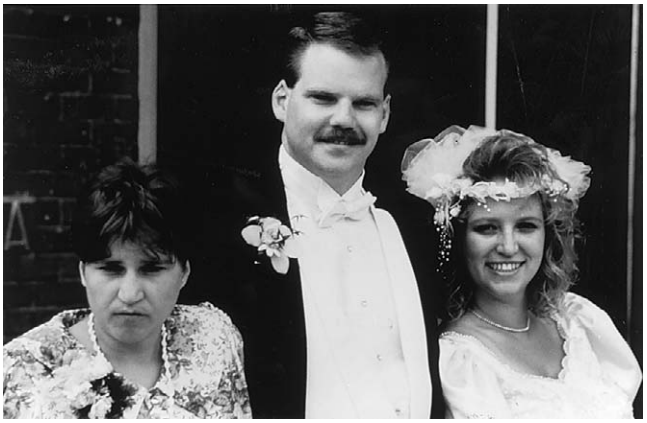

Figure 2 Irene at her wedding. The bridesmaid is her sister, Gaby. Irene and Gaby both have PKU-causing alleles at the *PAH* locus. This image appears on the PAHdb Web site.

the bride, is a woman I have known for almost 40 years, through diagnosis and treatment of her phenylketonuria (PKU). The other woman in the photograph is her older sister, who was born too early to benefit from screening, escaped early diagnosis, and did not receive effective treatment. My own interest in the treatment of genetic disease in general and of PKU in particular began with these two sisters. This particular photo has now been reproduced (with permission) in the 5th edition of Hartl and Jones's *Genetics: Analysis of Genes and Genomes* (2001). Its readers are being educated (virtually) by Irene.

PKU-causing mutations harbored by Irene and by thousands of other PKU probands are increasingly known and recorded in a locus-specific mutation database that my colleagues and I curate; PAHdb has become a heavily annotated knowledgebase (Scriver et al. 2000). It was focused initially to provide information for scientists, until Irene and a graduate student (Piotr Nowacki) suggested the addition of a clinical field in the database that would provide information for parents. We took the advice, and few days now pass without communication to and from patients, families, or students somewhere in the world. PAHdb has become an educational device.

PAHdb is also connected to chapter 77, on the topic of hyperphenylalaninemia, in the 8th edition of MMBID. The book itself is scheduled to move from print to an online electronic version, at which time it will become a continuous book on an open schedule in the manner familiar to all who use OMIM. Hereafter, Irene, you, and I will be connected to MMBID, OMIM, PAHdb, and other electronic resources in ways imagined a generation ago perhaps only by authors of science fiction.

Enlightenment

On September 11, 2001, different human values collided violently. In the aftermath, *Homo sapiens* has been challenged to ponder the merits of justice, truth, and respect for our individuality and to weigh those convictions in the balance against revenge and further violence. I found myself reflecting on the convictions that fostered the Enlightenment, particularly in England and Scotland, two centuries earlier. Its scientists and philosophers recognized that reason is superior to irrationality, moderation is better than fanaticism, and liberalism offers freedoms preferable to authoritarianism. Together, those convictions offered relative freedom from political, racial, and religious fanaticism. The Enlightenment was also a great friend of science (indeed, one of its legatees was Charles Darwin). It recognized that the natural tools of science are hypotheses and measurement, while the particular tools are observations and experiments. The thinkers of the Enlightenment knew that intuitive certainty about the materialistic world, or any other world, is no substitute for well-tested empirical knowledge (Berlin 2001).

Whereas science seeks to find some form of *order* in our existence—for example, Copernican, Linnean, Darwinian, or genetic (fig. 3)—we are usually left with a deep yearning to know the *meaning* of it. Life on Earth is a biochemistry that transcends chemistry in its emergent properties. Moreover, humankind is aware, or should be aware, that all life is connected through interacting hierarchies—from DNA through cells, organ-

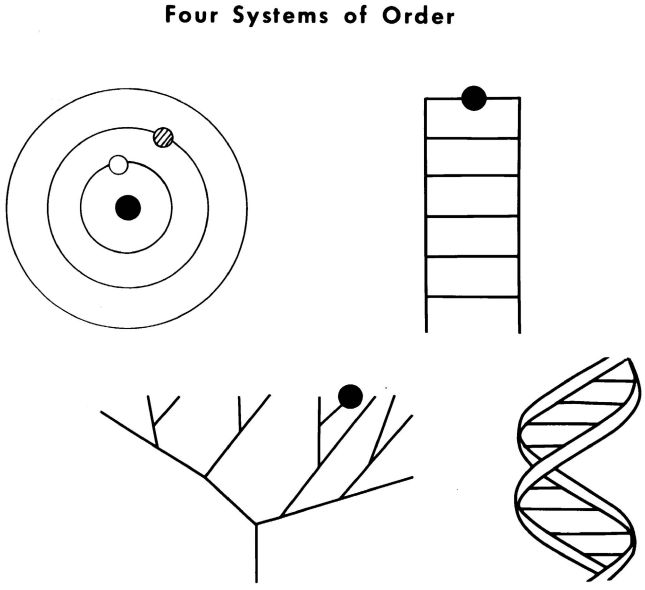

Figure 3 Four systems of order for the place of humankind in the materialistic world. (The systems are Copernican, Linnean, Darwinian, and genetic.)

Figure 4 Earth-rise from the moon; the photo was taken by a member of the species *Homo sapiens,* with a little technical assistance from *Homo modificans*. The image exposes the fragility of Earth and of life on it.

isms, communities, and populations to ecosystems and even the universe (Davies 2000). As individual waves are part of the ocean, so individual organisms are part of life on Earth. The immortal thread through all living organisms is the DNA molecule, a reality of chemical matter whose order we begin to understand. But the meaning of our being is another reality, a spiritual reality, perhaps forever beyond understanding, always a matter of faith. A complete education of *Homo sapiens,* the so-called sapient species, will introduce both realities, and it will see them as complementary and not competitive understandings. If this view of human education is ever fulfilled, I hope the materialistic gods pursued by some sectors of human society and the fanatical beliefs of others will become bad memories of the past.

Education about the biology and the genetics of life itself would fit nicely within the convictions of the Enlightenment. Knowledge of biology will make sense of my self, my genes, and the memes (Blackmore 1999) that I am conveying to you at this moment. Evolution makes sense of my biology. To know that a separation of the germline genome from the somatic genome has introduced a form of immortality is an enlightening perspective. It does not deny other interpretations of immortality.

I recognize five types of knowledge, each relevant to education in human genetics and biology, each apparent in every day life, and each connected in some way with the information we gather and the wisdom with which we use it. First, there is the unknowable—regarding the ultimate mystery of life. Second, there is the unknown, which science ferrets out. Third, there is the known—knowledge used by *Homo modificans,* the subspecies of *Homo sapiens* that has forever been modifying experience and trying to modulate the effects of chance. Then there are two other types of knowledge: one is called "I-don't-want-to-know"; the other is knowledge that is forbidden and censored. Unfortunately, the fourth and fifth types of knowledge are all around us. Yet all five types of knowledge are encountered in the moments of our lives: in mystery and God at one end of the five-part spectrum and in Lysenko and Creationism at the other. However, the chemical, knowable, materialistic view of life *can* coexist with vitalistic unknowable views. Both are realities, and, as complementary memes, they can live and replicate together in our minds.

Envoie

This was probably not the speech you expected to hear (or read), but you will recognize a shared angst about the near- and long-term futures of humankind. My faith is that education will yield the future. The only other alternative I can suggest is a fully penetrant, dominant universal germline mutation sweeping through the human genome, encoding altruism, kindness, and love of ourselves and our neighbors. But, being who we are, someone will probably patent its use and claim royalties every time we are generous and love each other.

To end, I borrow four lines of 20th century poetry (Eliot 1963; "Little Gidding"):

> We shall not cease from exploration
> The end of all our exploring
> Will be to arrive where we started
> And know the place for the first time.

That place is, among other things, our earthly home (fig. 4); I will meet you there.

Acknowledgments

I am grateful for many things, among them the institutions where I have worked for over 40 years; the innumerable ways Huguette Rizziero and Lynne Prevost have helped me over 30 years; the granting agencies, governments, and donors who funded the work; the discussions with Joan Matthews; and the gift of my family.

Electronic-Database Information

Accession numbers and URLs for data in this article are as follows:

Online Mendelian Inheritance in Man (OMIM), http://www.ncbi.nlm.nih.gov/Omim/ (for β-thalassemia [MIM 141900], PKU [MIM 261600], and Tay-Sachs disease [MIM 272750])
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