2001 ASHG AWARD FOR EXCELLENCE IN EDUCATION
Introductory Speech for Charles Scriver*

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Most here today would summarize their professional activities under some combination of three broad categories: research, clinical practice, and education. Unfortunately, in these times when researchers are faced with an overwhelming flood of new information and when clinicians are under increasing pressure to see more and more patients, education all too often ends up at the bottom of the priority list. This, despite the fact that there is no activity more important than education of our colleagues, the public, and especially our students, the geneticists of the next generation. Thus, it is my distinct pleasure and honor to introduce someone who has put education at the top of his list, the recipient of this year's Award for Excellence in Education, my colleague, friend, and advisor, Charles Scriver.

Charles is well known to you all, so I will mention only a few key points of his history. He was born in Montreal into a scholarly family; both his parents were academic physicians. He received all his formal training at McGill, graduating cum laude with a B.A. in 1951 and with an M.D. in 1955. Over the next 3 years, he completed a pediatric residency, also at McGill, and then, in 1958, began his career in biochemical genetics with a 2-year fellowship with Professor C. E. Dent at University College in London, where he was exposed to exciting early work on disorders of amino acid transport. In 1960, he returned to Montreal to complete a chief residency in pediatrics at McGill, and the rest, as they say, is history. Charles has now published more than 300 papers and numerous reviews and books and is a fellow of the Royal Society and the Royal Society of Canada.

Throughout his career, Charles has been involved in education. Images over the years from the Bar Harbor course, taken by Victor McKusick, document this point (fig. 1). Thus, when Vasken der Kaloustian called to ask if I would second his nomination of Charles for the Award for Excellence in Education, I was thrilled to do so. Of Charles's many educational contributions, I will emphasize several of particular relevance to his selection and to me.

First, from the early 1960s, Charles has played a major role in establishing the field of modern biochemical genetics. His initial publications on disorders of proline and glutamate metabolism and inborn errors of amino acid transport contributed enormously to the early excitement that drove the growth of the specialty. Particularly important, from an educational perspective, was the slender book that he and Leon Rosenberg coauthored, *Amino Acid Metabolism and Its Disorders* (Scriver and Rosenberg 1973). This monograph contains extensive information on normal amino acid biochemistry and physiology, together with chapters on the clinical and metabolic aspects of the abnormal phenotypes. Although

Figure 1 Charles Scriver teaching at the "Bar Harbor Course" in 1972 (*a*) and 1987 (*b*). Photos courtesy of Victor McKusick.

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Figure 2 A full set of *The Metabolic and Molecular Bases of Inherited Disease*. Charles Scriver has been the senior editor for the last three editions: the 6th (*red*), the 7th (*blue*), and the 8th (*green*).

currently out of print, it remains the best available summary of amino acid physiology and, over the years, has contributed to the training of two or more generations of biochemical geneticists.

A second major contribution has been Charles's work in the areas of patient and community education. An excellent example is his work on Tay-Sachs and thalassemia screening and education in Quebec. In a beautiful and comprehensive study described in a series of papers, Charles developed and tested Tay-Sachs and thalassemia screening and educational programs targeted at high school students, reasoning that this is an age when people are highly concerned with their own biology and would be most receptive to this information. Follow-up evaluation of the first 20 years of this effort indicated that the vast majority of those tested correctly retained knowledge of their carrier status and used this information in their family planning (Mitchell et al. 1996). This program contributed to a >90% reduction in the incidence of these two diseases in Quebec, an outstanding example of successful education of an entire population about the value of genetic medicine.

A third educational focus of Charles's work has been his ongoing interest in treatment of genetic diseases. Together with Barton Childs, a previous recipient of this award, Charles has been responsible for a series of studies evaluating treatment of genetic disease over the last 25 years (Costa et al. 1985; Hayes et al. 1985; Treacy et al. 1995). This work has emphasized the enormous challenges involved in our attempts to treat genetic disease and has provided a metric by which we can measure our progress to this end.

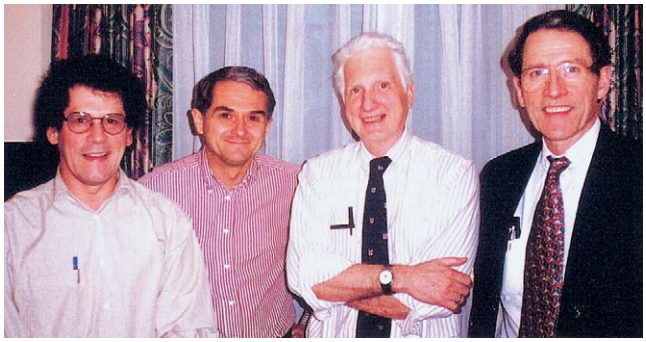

Figure 3 The editors of *The Metabolic and Molecular Bases of Inherited Disease* in 1989, when the 6th edition had just appeared and the 7th was in planning. Charles Scriver is second from the right.

More recently, Charles has been deeply involved in educating all of us about the challenge of understanding complex traits—the next frontier of genetic medicine. His strategy, not surprisingly, has been to use a "simple" monogenic disorder, phenylketonuria (PKU), as an example of a complex trait. Delving deep into PKU and phenylalanine hydroxylase, he has shown how multiple genetic and environmental factors interact to influence the phenotype of mutations at the *PAH* locus (Scriver and Waters 1999). This work, based on a disorder we think we understand, provides a conceptual framework for those we clearly don't. As part of this effort, Charles quickly recognized the potential of cyberspace for genetic education. His phenylalanine hydroxylase locus knowledgebase (PAHdb) is a superb online relational resource, useful not only to workers in the field but also as a model to all who use mutations and mutant phenotypes as a way to understand genetics. It also provides extensive educational material for PKU patients and families.

Last, I would emphasize the enormous contributions Charles has made as the senior editor of the last three editions (6th, 7th, and 8th) of *The Metabolic and Molecular Bases of Inherited Disease* (fig. 2). Without his guidance, expertise, and energy, the book might not have survived transition of its editorial leadership (fig. 3). Nor would it have expanded in scope from straightforward coverage of classical inborn errors of metabolism to a compendium on all genetic medicine. I am clearly biased here, but I would argue that Charles's efforts on this book alone are an invaluable educational accomplishment.

In summary, Charles has served as a model for all of us with his unflagging, diverse, and highly successful educational activities. He is a wonderful choice as this year's recipient of the ASHG Award for Excellence in Education.

Electronic-Database Information

The URL for data in this article is as follows:

PAHdb, http://data.mch.mcgill.ca/pahdb_new/

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