Book Review

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Robert Guthrie: The PKU Story. By Jean Holt Koch. Pasadena: Hope Publishing House, 1997. Pp. 190. $20.00.

One of the great stories of modern medicine is the development of routine newborn screening for phenylketonuria, better known as "PKU," and the resulting prevention of mental retardation from this biochemical genetic disorder. For this development, thousands of families throughout the world thank Robert Guthrie. With the addition of coverage for other important disorders (notably, congenital hypothyroidism), newborn screening now has become an integral component of newborn health care. Moreover, the presymptomatic detection of metabolic disorders through newborn screening has been a powerful stimulant of research in biochemical genetics. In this book, Jean Holt Koch tells the story of Bob Guthrie and how newborn screening came to be.

Reading the book called to mind a personal experience. Bob and I were in a taxi in New York, heading for a meeting. It was a long ride, ~45 min, so there was an opportunity to discuss (what else?) newborn screening. At the time, I was involved in research into histidinemia. I mentioned this to Bob and explained that, so far, the data strongly indicated that histidinemia was benign. Bob only allowed himself to hear the word "histidinemia" and then launched into a monologue about his "bug test" for histidine and the importance of adding it to newborn screening. Periodically, I was able to get in a word or two—for instance, about how the case reports of mental retardation and histidinemia that he was mentioning might have been only coincidental occurrences and about other case reports of clinically normal children with histidinemia—but I knew that I was outclassed by a master. In his soft, understated monotone, Bob pressed on relentlessly, convinced that there was no disorder, not one, that should not be added to newborn screening. This insistence and refusal to even allow, much less admit the possible validity of, a contrary opinion about newborn screening could be infuriating. It was not that Bob was unkind. To the contrary, he was one of the kindest and most concerned human beings I have ever known. He could not be rude if he tried. In his single-mindedness about newborn screening, shaped by his extraordinary concern for children and their right to health care and for the prevention of mental retardation, he simply was oblivious to intrusions. It was this sort of uninterrupted focus, shared by all individuals who do great things, that enabled Bob to get newborn screening adopted throughout the world.

The book begins with a description of Bob's upbringing and education. His true love was microbiology, a love that continued throughout his life. He was talked into obtaining a medical degree, which he always said he never used—"the only thing his medical degree did was get his car into the doctors' parking lot at hospitals" (p. 9)—but which was, as Koch points out, essential to Bob's success. Being a physician allowed him to understand the human-disease implications of his "bug tests" and, very importantly, gave him the air of authority that was essential for influencing parent groups to lobby for PKU screening as a public-health measure.

The core of the book is, of course, the newborn-screening test for PKU. It is a fascinating story, and, when reading it, I learned several facts that I had not known. One question frequently asked is whether Bob ever considered obtaining a patent and marketing the test. I knew that he never patented or sold the test and that he never made a penny from it, but I did not know that he had considered allowing the PKU test to be commercialized, as a means for widespread and efficient use of the test. Reading the short chapter "In the Lawyers' Den" is an educational experience that we all can use, especially today.

Koch has performed a wonderful service in bringing to us the story of Bob Guthrie and newborn screening. As the wife of Richard Koch, who has organized and directed national and international studies of PKU, she knew Bob personally. He had stayed in their home on several occasions. She spent many hours obtaining the information for this book from Bob and from others who had known Bob or who had worked closely with him. Most importantly, she has put the story together in an interesting, informative, and memorable manner. She shows us how Bob and his wife's personal tragedy of having a mentally retarded son (who did not have PKU) put Bob on the path toward developing the so-called Guthrie test for PKU; how his intelligence, education, and curiosity about everything scientific allowed him to develop the test; and how his inheritance and upbringing gave him the persistence to insist on the test's adoption for newborn screening and the prevention of mental retardation from PKU, despite vigorous, and at times even vituperative, opposition from the medical establishment. Koch weaves these facets of Bob Guthrie's life into a fascinating and unforgettable story.

This book should be required reading for anyone directly or indirectly involved in newborn screening. The book also will be informative and interesting to all geneticists, since so much of the excitement and interest in genetics began with the ability to prevent, by newborn screening, mental retardation from a genetic disease, namely, PKU. The enjoyment of reading this short book is guaranteed to be a rich return on the very modest price.

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