

12-1-2014

# Review Of "The PKU Paradox: A Short History Of A Genetic Disease" By D. B. Paul And J. P. Brosco

John B. Jenkins

*Swarthmore College*, [jjenkins1@swarthmore.edu](mailto:jjenkins1@swarthmore.edu)

Let us know how access to these works benefits you

Follow this and additional works at: <http://works.swarthmore.edu/fac-biology>



Part of the [Biology Commons](#), and the [Genetics Commons](#)

---

## Recommended Citation

John B. Jenkins. (2014). "Review Of "The PKU Paradox: A Short History Of A Genetic Disease" By D. B. Paul And J. P. Brosco". *Quarterly Review Of Biology*. Volume 89, Issue 4. 409-409.  
<http://works.swarthmore.edu/fac-biology/460>

This Book Review is brought to you for free and open access by the Biology at Works. It has been accepted for inclusion in Biology Faculty Works by an authorized administrator of Works. For more information, please contact [myworks@swarthmore.edu](mailto:myworks@swarthmore.edu).

ulations could be broadly beneficial. If, however, they are suppressing immune responses, therapeutic inoculations could make a person vulnerable to infectious diseases. On balance, the evidence supports immune suppression rather than tuning, thus weakening the Hygiene Hypothesis and emphasizing the dangers associated with therapeutic inoculations of parasites.

I strongly recommend this book to anyone who wants a wide-ranging, accessible account of the evidence that is being used to support the Hygiene Hypothesis. The caveat is that readers will need to do their own critical thinking and outside reading to adequately evaluate which alternative explanations are most compelling. Still, Velasquez-Manoff opens the door to considering the ways that the myriad residents of our bodies may influence our health, and the implications for reducing the damage that our immune systems sometimes inflict on us.

PAUL W. EWALD, *Biology, University of Louisville, Louisville, Kentucky*

THE PKU PARADOX: A SHORT HISTORY OF A GENETIC DISEASE. *Johns Hopkins Biographies of Disease.*

By Diane B. Paul and Jeffrey P. Brosco. Baltimore (Maryland): Johns Hopkins University Press. \$24.95 (paper). xxv + 289 p.; ill.; index. ISBN: 978-1-4214-1131-6 (pb); 978-1-4214-1132-3 (eb). 2013.

In 1995, Diane Paul wrote an important book, *Controlling Human Heredity: 1865 to the Present* (Atlantic Highlands (NJ): Humanities Press), about the eugenics movement. She explored the ethical, legal, and social issues posed by the rapidly emerging powers of medical and genetic science and how those powers have enriched our understanding of the roles they played in the modern history of Western civilization, both positive and negative. One of my human genetics seminars was centered on this volume and it was a remarkable experience for all.

Paul, who is currently Professor Emerita at the University of Massachusetts and a research associate at Harvard University, has now teamed with Jeffrey Brosco, a clinical pediatrician at the University of Miami School of Medicine, to explore the history of phenylketonuria (PKU), a rare but well-understood inherited metabolic disorder. PKU is the paradigm for successful medical intervention in genetic disease. If left untreated, a person born with PKU will develop debilitating intellectual disabilities. But if detected early and treated with a strict although difficult dietary regimen that restricts the intake of the amino acid phenylalanine, an essential amino acid, the prognosis for normal neurological functions is excellent.

Why do Paul and Brosco use the word “paradox” in the title? It is because even though PKU is rare and most pediatricians will never encounter a case dur-

ing their entire medical career, every newborn child is tested for PKU. This is an inexpensive test (the Guthrie test) and it has an enormous upside if a child is found to have PKU. If undetected and untreated, the consequences—physically, emotionally, and socially—are enormous.

The authors begin by introducing us to the family of Pearl S. Buck, the winner of the Nobel Prize in Literature (1938) for her writings about life in China and her biographical masterpieces. Buck had a child, Carol, who was born with PKU, although she kept this issue very private. She wrote about her daughter in her short, moving book, *The Child Who Never Grew* (1950. New York: J. Day Company). Carol was born before there was a test for PKU so the neurological damage was done before caregivers could ascertain the problem. The narrative moves on to Ivar Asbjørn Følling’s landmark studies of the cause of PKU (Peter Harper includes Følling’s study in his collection, *Landmarks in Medical Genetics: Classic Papers with Commentaries*. 2004. Oxford (United Kingdom): Oxford University Press). Uncovering the metabolic basis of PKU led to strategies for trying to control the disorder by dietary intervention.

Mandatory testing for PKU was championed by President John F. Kennedy and his family, not only because they had an intellectually impaired family member, but because they thought that PKU would serve as the model to search for other metabolic disruptions that could result in mental disorders. Paul and Brosco discuss the fascinating development of mandatory testing for PKU as well as the skepticism that surrounded it.

The final chapters examine new paradigms for PKU and look closely at the highly charged debates about the infamous genetics and IQ controversies of the late 1960s, 1970s, and 1980s (i.e., Arthur Jensen). The authors look at issues of patients living with PKU, the problem of maternal PKU, where a mother who has been successfully managed for PKU, then stops following the phenylalanine-restricted diet becomes pregnant with a non-PKU child who then becomes intellectually impaired because of the high levels of phenylalanine in the maternal circulatory system. They conclude with a thought-provoking discussion of political and eugenic issues.

Paul and Brosco are to be commended for reminding us all how central the PKU story has been to the development of genetics and medicine, the connection of genotype to phenotype to society, and to the way this society views people with intellectual disabilities. This is a book that should be read by all geneticists and physicians and people with an interest and concern about individuals born with intellectual impairments.

JOHN B. JENKINS, *Biology, Swarthmore College, Swarthmore, Pennsylvania*