



# The PKU Paradox (Johns Hopkins Biographies of Disease)

*Diane B. Paul, Jeffrey P. Brosco*

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PKU (phenylketonuria) is a genetic disorder that causes severe cognitive impairment if it is not detected and treated with a strict and difficult diet. Programs to detect PKU and start treatment early are deservedly considered a public health success story. Some have traded on this success to urge expanded newborn screening, defend basic research in genetics, and confront proponents of genetic determinism. In this context, treatment for PKU is typically represented as a simple matter of adhering to a low-phenylalanine diet. In reality, the challenges of living with PKU are daunting.

In this first general history of PKU, a historian and a pediatrician explore how a rare genetic disease became the object of an unprecedented system for routine testing. *The PKU Paradox* is informed by interviews with scientists, clinicians, policymakers, and individuals who live with the disease. The questions it raises touch on ongoing controversies about newborn screening and what happens to blood samples collected at birth.]]>

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