Phenylketonuria (PKU): Screening and Management

January 1980 through July 2000

3394 Citations
Phenylketonuria (PKU): Screening and Management

January 1980 through July 2000, plus selected earlier citations

3394 Citations

Prepared by

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INTRODUCTION

Phenylketonuria or PKU is a rare, inherited metabolic disorder that, if untreated, causes mental retardation. Approximately one of every 10,000 infants in the United States is born with PKU, which usually results from a deficiency of a liver enzyme known as phenylalanine hydroxylase (PAH). This enzyme deficiency leads to elevated levels of the amino acid phenylalanine in the bloodstream.

All infants in this country undergo blood testing for PKU. The current treatment for this disorder involves dietary modification. When a very strict diet is begun early and maintained, children with PKU can expect normal development and a normal lifespan. The diet generally excludes all high protein foods, such as meat, milk, eggs, and nuts, since all protein contains phenylalanine. Dietary noncompliance can result in a decline in mental and behavioral performance. Women with PKU must also maintain a strictly controlled diet before and during pregnancy to prevent fetal damage. Scientists are actively exploring nondietary treatments for PKU.

This bibliography was prepared in support of the National Institutes of Health (NIH) Consensus Development Conference titled Phenylketonuria (PKU): Screening and Management held in Bethesda, MD on October 16–18, 2000. It includes citations to journal articles, books and book chapters, conference proceedings and conference papers, and dissertations in all languages published primarily from January 1980 through July 2000. Works of historical significance and other selected references from earlier years have also been included. Citations have been arranged by subject and then alphabetically by author within each subject. A citation may appear under more than one subject. For example, a citation discussing the economic aspects of screening for PKU would be found under both “Economics and Socioeconomics” and “Screening and Diagnosis.”

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Journal Article:

Authors                  Article Title

Monograph:

Authors/Editors                               Title

*For details of the formats used for references, see the following publication:

History and Classic Material


Guttler F. Phenylketonuria: 50 years since Folling’s discovery and still expanding our clinical and


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