Phenylketonuria - an inherited metabolic disorder associated with mental retardation

U.S. Dept. of Health, Education, and Welfare, Social Security Administration, Childrens Bureau - Perioperative care of children with inherited metabolic disorders

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Perioperative care of children with inherited metabolic disorders

In the event of an acute crisis, these children should be rehydrated, have infections aggressively identified and treated, and have measures to prevent catabolism instituted. The number and complexity of metabolic processes required to maintain homeostasis is vast and so it follows that the spectrum of human disorders ascribed to inherited defects in metabolism is considerable. People with the same disease may not have all the symptoms listed.

Long

For the rest of their lives, people with PKU — babies, children and adults — need to follow a diet that limits phenylalanine, which is found mostly in foods that contain protein.

Phenylketonuria is still a major cause of mental retardation in Tunisia despite the possibility of treatment

Since 1991, CDC has conducted MADDSP, an ongoing, population-based surveillance system for selected developmental disabilities i. Some registries collect contact information while others collect more detailed medical information. An original paper copy of this issue can be obtained from the Superintendent of Documents, U.

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This is a medical emergency and may be the first presentation of an IMD. An overview of some of the commoner IMDs is presented in Table. The measurement of the anion gap is essential in evaluating these patients.

Phenylketonuria: An Inborn Error of Phenylalanine Metabolism

NORD is a patient advocacy organization for individuals with rare diseases and the organizations that serve them. Andersen OA, Flatmark T, Hough E. This site provides information and resources about screening at the local, state, and national levels and serves as the Clearinghouse for newborn screening information.

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