COMMITTEE ON CHILDREN WITH HANDICAPS

PHENYLKETONURIA AND THE PHENYLALANINEMIAS OF INFANCY*

In 1965 this Committee issued a statement outlining the responsibilities of the physician to the child with phenylketonuria, an inherited abnormality of amino acid metabolism.

A lack of knowledge about the disorder and about the results of treatment placed constraints on the 1965 statement. The Committee therefore feels that, with recent advances in knowledge about the disorder, a new statement is needed.

An increased level of phenylalanine in the blood can occur under sporadic and transient conditions in the absence of disease, with or without a concomitant elevation of serum tyrosine; it is always present in the disorder now called phenylketonuria. Because of incomplete information, simple classification of a specific case of phenylalaninemia is often not possible.

Screening programs allow for the detection of infants with elevated blood levels of phenylalanine. Screening programs should be encouraged and supported because they are the best available means for identifying all infants with abnormalities of protein metabolism resulting in serum phenylalanine elevations.

Two important, unresolved issues need clarification: (1) the effect of a persistently elevated blood level of phenylalanine on the intellectual growth of the child when there are no other indications of disease, and (2) the possibility of harmful effects of a diet low in phenylalanine.

The basic treatment of phenylketonuria is to reduce circulating phenylalanine by dietary restriction; and, because a spectrum of disorders causes an elevation of that amino acid, differing approaches in management seem indicated.

The relative rarity of phenylketonuria precludes the opportunity for individual physicians to gain widespread experience and expertise in management outside of a hospital specialty clinic setting. Furthermore, it is desirable to group children with phenylketonuria and related disorders in a clinical setting because these children need the skillful management possible in a therapeutic environment with supportive laboratory and dietary care.

There is a definite need for continuing research and for clustering cases of the disease so investigators in the field will have access to adequate clinical material.

The management of a youngster with an elevated blood level of phenylalanine is complex, and controversy often arises about the appropriate course of action to be taken by a physician attempting treatment. There is no simple solution to the problem. Therefore, after careful consideration of available information, this Committee recommends that, wherever feasible, a child with phenylketonuria should be followed regularly in a clinic or university setting by physicians with expertise in the field, as well as by the child's primary physician, who should be encouraged to participate fully in the treatment program. The needs of the child and the physician, as well as the research needs of the discipline, will best be served by this approach.

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* This statement has been reviewed and approved by the Academy's Council on Child Health.

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