Serum phenylalanine levels

Without early diagnosis and appropriate treatment, phenylketonuria (PKU) leads to mental retardation. Loss of intelligence is preventable with good management. Serum phenylalanine levels of 1-10 mg% are safe and acceptable, but levels of 1-6 mg% are ideal and especially important for infants and young children. The current standard of care in the United States is to maintain these safe levels throughout life. No one with PKU should ever be "taken off diet."

Once newborn children are under satisfactory management, a monthly serum phenylalanine (or "phe") level is recommended, which is drawn at the monthly clinic. If that level is out of the recommended range, a follow-up level is requested.

Food patterns

Children with PKU must eliminate high protein foods from their diet (meat, fish, dairy products, eggs, poultry) and restrict their intake of moderate protein foods (potatoes, grains). As children grow older, they need to increase their intake of low protein foods (certain fruits and vegetables, low protein breads and low protein pasta products) to maintain appropriate energy (calorie) intake and prevent hunger. Adequate formula intake plus low protein foods provide a generous amount of protein, phenylalanine, and tyrosine at the requirement level, energy to support growth, and all essential nutrients.

Formula

All formula is prescribed by a physician and monitored by a nutritionist. Those infants and children who receive a consistent daily intake of a phenylalanine -free formula/medical food (roughly 85% of protein needs and 90% of their energy needs) have the most stable and appropriate serum phenylalanine levels. The clinic policy is "formula first." Parents are taught that formula is given before other foods, both to meet nutritional needs and to satisfy appetite. Formula is necessary to maintain low blood phe levels that support long-term compliance and lead to the completion of school, employment, and independence.

Nutrition education

Teaching the children to manage their diets by themselves is an important aspect of the program, starting with "yes/no foods" and moving on to "how much." At the monthly clinics, nutrition education activities are provided for each age group to aid the children in learning about their diets. Two concepts are reinforced by this clinic, the first being that the parent is the teacher. Materials and ideas with which the parent can continue to teach their child about the diet at home are provided. The importance of consistency and appropriate development of food habits are emphasized. The second concept is that of health. Children with well-managed PKU are normal, healthy children. Every possible attempt is made to treat them as such and not as if they have a disease. All aspects of the child—diet, physical fitness, self-esteem, and self-worth—are considered. The consistent food patterns and routines established by parents in early childhood are essential for dietary compliance when the children are older and more independent.

Illness

It is important to aggressively treat the usual childhood illnesses to prevent tissue catabolism and consequent elevated blood phenylalanine levels. During an illness or infection, it is important to maintain the formula intake as much as possible. The continuation of formula is to help prevent the breakdown of muscle protein during illness that will increase the level of phenylalanine in the blood. However, if a child refuses to drink formula, then whatever clear liquid the primary care physician recommends is appropriate. Parents need to contact the primary care physician for management of the illness before calling the PKU staff.

Lifetime diet

The current standard of care is a low phenylalanine diet, including formula/medical food, for life. The PKU Collaborative Study documented long-term dietary intervention as the only method to achieve optimal intellectual ability. When children are taken off the low phenylalanine diet and their blood phenylalanine levels increase, their IQ or achievement test scores drop. Cranial MRI studies have documented abnormalities consistent with demyelination with the severity of changes correlating with high blood phenylalanine levels. Central to long-term compliance is regular monitoring of progress: monthly (or more frequent) serum phe levels and food records are important "benchmarks of progress" for persons with PKU.

Clinic Format

A monthly clinic is held in Seattle at the University of Washington. It is always held on the third Wednesday of the month and the following Thursday. Instead of giving children individual appointments, children and their parents are seen in groups based on age. The children meet with the nutritionists to work on a nutrition education project while the parents meet in a separate group to discuss issues related to raising a child with PKU. After the group time, the families meet individually with the pediatrician, nutritionist, and social worker.

This group format was chosen for several reasons. First, it is a reliable and efficient way to educate children and parents and keep them up to date on recent developments in PKU management. Additionally, parents can support each other in important ways, and children benefit immeasureably from knowing other children with PKU who make the same food choices they do.

A clinic in Spokane is held about five times per year for families in Eastern Washington. Seattle-area families are strongly urged to attend the monthly clinic. For families who live too far away to attend monthly clinics, a monthly serum phenylalanine level and three day food record is required. Experience has shown that this monthly feedback is essential to maintain good serum phenylalanine levels.

Maternal PKU

There is a special dilemma for females with phenylketonuria. It is difficult for a woman with PKU to have a normal baby. Even when serum phenylalanine levels are kept low, there may be some damage to the fetus. The higher the serum phenylalanine level, the poorer the fetal outcome. Infants of mothers with PKU exhibit microcephaly, mental retardation, and growth retardation. Due to this adverse outcome, we encourage women with PKU to carefully consider all the risks of pregnancy; we recommend other alternatives such as adoption. This matter is discussed early with the family so that the consequences are well understood and appropriate decisions can be made.

Clinic Staff

The PKU Program at the University of Washington is primarily funded by the Washington State Newborn Screening Program. The State Genetics Program provides families with subsidized formula (paid for by their medical insurance). The programs are closely coordinated to endure follow-up and appropriate management of individuals with PKU.

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