PATIENTS & PROVIDERS FOR MEDICAL NUTRITION EQUITY

HOW COVERAGE OF MEDICAL FOODS CAN SAVE LIVES & COSTS

INBORN ERRORS OF METABOLISM

Inborn Errors of Metabolism (IEM), also referred to as Inherited Metabolic Disorders are lifelong genetic conditions in which specific enzyme defects interfere with the normal metabolism of protein, carbohydrate, or fat. Federal newborn screening policy and state screening programs identify the majority of IEM. Medical nutrition intervention is a mainstay of patient management and must begin shortly after birth to prevent death, intellectual disability, and other adverse health outcomes.

Individually, IEM are rare. For example:

- Glutaric Acidemia Type 1 (GA-1) occurs in 1 in 92,300 live births
- Very Long-Chain Acyl-CoA Dehydrogenase (VLCAD) Deficiency 1 in 63,500 live births
- Phenylketonuria (PKU) 1 in 16,500 live births

There are no cures for IEM, but treatment with medical food, foods modified to be low in protein, and/or supplementation with individual amino acids, and large doses of vitamins allow patients to achieve normal or near-normal health. **Twenty-two of the 34 core conditions on the HHS Recommended Uniform Screening Panel (RUSP) must be treated with medical nutrition for life. There is no alternative to this treatment.**

The consequences of not treating these conditions are devastating. For example:

- GA-1: Metabolic crisis that can be fatal. For survivors irreversible brain damage that can affect the ability to walk, talk or even swallow safely, often with uncontrolled, painful movements called dystonia
- VLCAD: Metabolic crisis that can be fatal. For survivors poor growth, liver failure, heart failure, and episodes of painful muscle breakdown called rhabdomyolysis that can cause kidney failure
- PKU: Irreversible cognitive impairment, hyperactivity, autistic behavior, seizures
- Maternal PKU Syndrome: Affects children of women with poorly treated PKU and may include microcephaly (small brains), irreversible intellectual disabilities, congenital heart defects and other birth defects, and low birth weight
- Homocystinuria (HCY): Strokes that can be fatal. For survivors, paralysis, cognitive impairment, abnormal vision because of dislocated lenses, osteoporosis, and skeletal deformities

The annual total cost to treat IEM with medical nutrition ranges from $2,254 for an infant to almost $25,000 for an adult male or pregnant woman. Without coverage, treatment is unaffordable for the majority of patients. However, the cost of NOT providing accessible and appropriate treatment for these patients is much greater.

The inequity of medical nutrition coverage across the United States has been, and continues to be, detrimental to individuals with IEM, their families, and society. In 2008, the Newborn Screening Saves Lives Act passed with overwhelming support. The Medical Nutrition Equity Act is the best way to achieve the aims of that act: healthy lives for those with IEM. The Patients & Providers for Medical Nutrition Equity Coalition calls on Congress to address this issue by passing the bi-partisan Medical Nutrition Equity Act. Please contact the offices of the bill leads (find them at nutritionequity.org/leads) to co-sponsor.

[1] B. L. Therrell, et al., Inborn errors of metabolism identified via newborn screening: Ten-year incidence data and costs of nutritional interventions for research agenda planning, Mol. Genet. Metab. (2014), http://dx.doi.org/10/1016/j.ymgme.2014.07.009
[2] K.M. Camp, M.A. Lloyd-Puryear, K.L. Huntington, Nutritional Treatment for Inborn Errors of Metabolism: Indications, Regulations, and Availability of Medical Foods and Dietary Supplements Using Phenylketonuria as an Example, Mol Genet Metab. 2012 September; 107(1-2); 3-9. Doi:10.1016/j.ymgme.2012.07.005