WHAT ARE PKU COMORBIDITIES

Comorbidity is the presence of one or more adverse medical conditions that occur along with a primary medical condition. For example, obesity is a common comorbidity of Type-2 diabetes. Management of individual comorbidities often requires different strategies than if the comorbidity was occurring as a single primary medical condition. PKU patients often experience multiple comorbidities. Comorbidities contribute to the complexity of treatment, less favorable clinical outcomes, increased cost of healthcare for PKU patients compared to non-PKU individuals, and negatively impact quality of life. [1, 2]

A recent survey of health insurance records showed that PKU patients suffer from a wide range of comorbidities compared to non-PKU controls. [3] Executive function deficits, neuropsychological deficits, behavior challenges, and depression are well-described primary morbidities of PKU around which most of the therapeutic effort is focused through management of blood Phe levels. [4]

In the early 1960’s, the low Phe diet emerged as the primary treatment for PKU which dramatically reduced the devastating outcomes of untreated PKU. However, as the first generation of patients to adopt and follow the PKU diet aged, it became clear that these patients also suffered from a wide array of comorbidities affecting multiple organ systems beyond the established neurological deficits. The two most common comorbidities are kidney failure with high blood pressure and obesity. These are followed in decreasing order of prevalence by kidney failure without high blood pressure, inflammation of the stomach and throat, kidney stones, hair loss and baldness, esophageal disorders, osteoporosis, acid reflux, hives, anemia, asthma, gallbladder diseases (gallstones, cholecystitis), dermatitis and eczema, and allergic and chronic rhinitis.

The presence of comorbidities increases the cost of healthcare for PKU patients. In one study, comorbidities in the PKU patient population were estimated to increase medical care costs fourfold over the 16 year period of retrospective insurance claim review. [3] It has also been suggested that some of the comorbidities are caused or exacerbated by the low Phe diet. This makes treatment of the comorbidity more difficult, increases the burden of care, and negatively impacts the quality of life of PKU patients. [1]

While more studies are needed to strengthen the correlation of comorbidities with PKU; these initial studies point to the need for careful monitoring of PKU patients throughout their lifetimes and the need for better treatments. It’s important to note that no individual patient experiences every listed comorbidity and no single comorbidity is experienced by every patient increasing the complexity of treatment and monitoring. While every treatment plan shares the common goal of lowering blood Phe levels, each patient will also have unique aspects of their treatment to address their set of comorbidities. The cumulative deleterious effect of high blood Phe in PKU patients and emerging recognition of PKU comorbidities justifies the clinical practice guideline recommendations for lifelong management to control blood Phe. [5, 6]


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