Quality-of-life Evaluation of Healthy Siblings of Children with Phenylketonuria

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Introduction: The diagnosis of a chronic disease can be stressful for the entire family, and it can also have a psychosocial influence on the children's siblings. Phenylketonuria (PKU) is an inherited autosomal recessive metabolic disorder caused by a deficiency of the enzyme phenylalanine hydroxylase (PAH). Due to high phenylalanine (phe) concentrations, untreated PKU results in severe irreversible neurological and especially cognitive impairments already within the first year after birth. We compared the health-related quality of life (HrQoL) scores of siblings of children with PKU to those of siblings of healthy children.

Patients and Method: The healthy siblings of 34 children with PKU and 100 healthy children were included in this study. The physical and psychosocial health scores were calculated using the replies of the sibling and parent on the Pediatric Quality of Life Inventory questionnaire.

Results: Physical health scores of healthy siblings of children with PKU were significantly lower than those of healthy siblings of healthy children, although psychosocial health and total health scores were comparable between the groups. There was no difference in the psychosocial health, physical health, or total scores of siblings of children in the PKU group and the control group, according to parents' reports. The healthy siblings of the children in the control group had no impairment in QoL, whereas 14.7% of the healthy siblings of the children in the PKU group had an impaired QoL.

Discussion: Daily dietary management, frequent blood tests for monitoring the phe levels and regular visits to a pediatric center for metabolic disorder make the therapy complex and require a lot of time and the effort for the patients and their families. Previous research showed that parents and siblings of chronically ill children are at risk of developing psychological disorders and psychosocial problems and to have poorer health than parents and siblings of healthy children. The majority of healthy siblings of children with PKU had their QoL impacted in varied degrees. The relationship between the siblings is influenced by their cognitive, social, and emotional development, which are the determinants of QoL. Because brotherhood/sisterhood is a lifelong relationship that defines one's identity and personality during adulthood, including healthy siblings in support programs for parents of children with PKU may be useful.