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Outcomes of referrals to Child Protective Services for medical neglect in patients with phenylketonuria: Experiences at a single treatment center

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ABSTRACT

Phenylketonuria (PKU) results in an accumulation of phenylalanine (phe) in the blood which can lead to multiple health consequences in affected individuals. Treatment for PKU is available; however adherence to medical management recommendations can be difficult. When recommendations are not followed and the health of a child is at risk, one intervention that may be necessary is a referral for medical neglect to the local child protective services (CPS) agency. This study summarizes the cases that were referred from our metabolic clinic at the Children's Hospital of Michigan to CPS, and the outcomes of that intervention. CPS referrals helped to improve adherence to medical management recommendations in the majority of cases, including a lower blood phe level for the child; however, at times that improvement did not occur until after a second referral and/or the child's temporary removal from the home.

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1. Introduction

Phenylketonuria (PKU; OMIM: #261600) is a rare, autosomal recessive inborn error of metabolism in which affected individuals are unable to convert the amino acid phenylalanine into the amino acid tyrosine. The result is an accumulation of phenylalanine (phe) in the blood and cerebrospinal fluid and a deficiency of tyrosine. Excessive phenylalanine can directly damage nerve cells in both white and gray matter by multiple mechanisms, including increased myelin turnover and disturbed myelin synthesis [1,2]. Lack of tyrosine leads to low neurotransmitter levels such as dopamine and serotonin [3]. Persistently elevated phe and reduced tyrosine levels can cause a multitude of clinical and subclinical effects, including physical and psychological effects (cognitive, emotional and behavioral) [1,4,5]. While some health consequences of persistently elevated phe levels in PKU are reversible, others are not [6].

Newborn screening for PKU was first implemented in the 1960s through detection of elevated blood phe levels in dried blood spots allowing the diagnosis of PKU to be made early in life. When treatment is initiated early and is maintained consistently throughout life most of the signs and symptoms of PKU can be avoided. However, even with proper treatment subtle signs and symptoms of PKU can still manifest in some individuals [7].

The mainstay of treatment for PKU in most individuals includes dietary protein restriction and supplementation with a phe-free metabolic formula. The diet aims to lower blood phe levels, while the formula supplements the required tyrosine, essential amino acids, and other nutrients. The addition of low-protein modified foods and pharmacological therapies may be useful for some patients. Overall, the goal for medical management of PKU is to maintain blood phe levels in the recommended goal range of 2–6 mg/dl [8] while increasing tyrosine levels to near normal.

In order to accomplish this goal, blood phenylalanine and tyrosine levels must be monitored on a regular basis. The medical management recommended to caregivers of the children with PKU includes the submission of blood samples taken from the child at home (dried blood spots) along with a 3-day diet record for each sample. The dried blood spots are then sent to a pre-designated laboratory for determination of blood phe and tyrosine levels. This process, termed home monitoring, allows for dietary and formula adjustments to maintain blood phe and tyrosine levels within the goal range on a consistent and regular basis. Medical management guidelines recommend that home monitoring be completed weekly during the first year of life and every other week after the 1st year until the individual with PKU reaches 12 years of age [8]. Optimum treatment of PKU, including the monitoring of growth and development, requires that individuals with PKU establish consistent and regular care with an experienced metabolic disease management team. The recommended frequency of such clinic visits is every 3 months in the first year of life and every 6 months after that until 12 years of age [8].

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The treatment regimen for PKU can be rigorous and at times seem burdensome, which can lead to problems with adherence to medical management recommendations. Lack of adherence is evident when blood phe levels are consistently elevated above the goal range, which usually indicates that recommendations regarding the amount of dietary protein and/or metabolic formula to consume are not being followed. Absence of home monitoring blood samples and diet records, missed clinic visits, and poor communication between the caregiver and clinic can also be considered as non-adherence to medical management recommendations.

Difficulties with adherence to PKU medical management recommendations, as evidenced by elevated blood phe levels, have been well documented [9]. One study found that phe levels were elevated above goal ranges in up to 30% of children with PKU under 4 years of age [10]. Another smaller study from Germany found that up to 53% of children with PKU under 9 years of age had elevated blood phe levels [11].

Problems with adherence to medical management recommendations are not limited to PKU and are seen in many other chronic childhood diseases, including asthma [12], human immunodeficiency (HIV) [13], sickle cell [14], and leukemia [15]. Non-adherence in chronic disease, including PKU, can be observed in any or all of three areas: participation in health care (e.g. not attending appointments), taking medication (not taking prescribed formula), and health behaviors (not following dietary recommendations or not completing home monitoring) [16]. In particular, maintaining and improving health behaviors can be the most difficult and may require additional interventions, compared with interventions needed to maintain and improve participation in health care and taking medication [17].

Since a child is dependent on caregivers, it is the caregiver's responsibility to ensure adherence to medical management recommendations. Unfortunately, caregivers can encounter many barriers to adherence, most of which are not unique to PKU itself. These barriers include cognitive (not able to understand PKU and treatment recommendations), educational (lack of information about PKU and rationale for treatment recommendations), social (lack of supportive relationships), financial (insurance, transportation, cost of food and metabolic formula), and family dynamics (unstable marital relationship, multiple caregivers, etc.). In addition, a lack of time to manage the PKU regimen successfully, the sometimes unpalatable metabolic formula and low-protein food substitutes, as well as a lack of immediate visible symptoms of elevated phe levels can contribute to non-adherence [18]. Recognition of such barriers and individualizing the interventions of the metabolic team is critical to optimizing the care of the PKU patient.

In rare instances when such interventions are unsuccessful, it may be necessary to utilize the authority that exists within the local Child Protective Services (CPS) agency by filing a complaint alleging medical neglect. The primary goal of such an approach is to require that the caregiver comply with medical recommendations, with the anticipated outcome of a decrease in blood phe level for the child. A secondary goal is to provide necessary resources for the caregiver that will facilitate continued adherence long-term.

Since 2005, Children's Hospital of Michigan Metabolic Clinic (CHMMC) has been responsible for follow-up of abnormal metabolic newborn screens for the entire state of Michigan through a grant provided by the Michigan Department of Community Health. In the years of 2007 through 2012, approximately 250 patients with PKU requiring dietary treatment were followed at CHMMC. Our metabolic clinic staff includes 2 physicians dually board-certified in Clinical Genetics and Clinical Biochemical Genetics, 2 pediatric nurse practitioners, 1 genetic counselor, 2 nurses, 3 dietitians, 1 psychologist, and 1 social worker. Between 2007 and 2012, we filed 12 complaints on 8 patients for medical neglect with CPS. The purpose of this study was to assess whether this particular intervention was successful in improving adherence to medical management recommendations in patients we referred.

2. Methods

Medical records of children with PKU followed at the Children's Hospital of Michigan Metabolic Clinic and reported to CPS for medical neglect between January 2007 and July 2012 were retrospectively reviewed. Data collected included date of birth of the child, date of and reason for CPS referral, clinic visit dates, dried blood spot submission dates, blood phenylalanine levels, clinic interventions prior to filing, caregiver-identified barriers, socioeconomic status, and CPS interventions. The protocol was approved as an exempt study by the Wayne State University Institutional Review Board. In addition, a waiver of consent was obtained as all information collected was de-identified, with no elements of private health information (PHI) recorded.

Data analysis was completed using primarily descriptive statistics due to the limited sample size and large number of variables that affect adherence. Pre-CPS and post-CPS referral dried blood spot submission rates and blood phe levels were compared, and Excel spreadsheets were exported to SPSS for graphical representation of submission frequency and phe levels.

3. Results

Medical neglect complaints were filed on 8 children affected with PKU. A total of 12 complaints were filed, which includes second complaints filed on the same child in 4 instances.

The average age of the child when the first CPS report was filed was 4 years, with an age range of less than 1 year to 7 years. In cases when a second CPS referral was made the average age of the child was 5 years, with a range of 3 to 9 years.

In reviewing the socioeconomic data and care-giver identified barriers, there were no identified common themes for socioeconomic status or barriers. This may be in part due to the small sample size. However, many of the barriers listed previously in this article (Section 1) were cited by the non-adherent caregivers of our children with PKU, and we did not identify any new or unique barriers.

Socioeconomic information, including parental marital status, employment status, educational status, and income level, were obtained when the information was available in the patient's medical record. Four of the patients were from single parent homes with no involvement of the other parent. Two of the patients had parents that were married and living together in the same home, while the remaining two patients had 2 parents that were involved in their care, but were not living together. One or both parents were employed consistently in 3 of the 8 families. Three more families had parents with sporadic employment, while one family was unemployed, and the employment status of the eighth family is unknown. Financial concerns were voiced by four of the families. Individual income information is not typically obtained for our patients. Highest educational levels of parents was not assessed in the majority of these cases, though 2 out of 8 of the families had one parent that was attending post-secondary educational classes.

Barriers to adherence with the medical management recommendations for their child's PKU were a combination of PKU-specific barriers along with specific situations within each family that were not related to the child's PKU. PKU-specific barriers identified by parents of our patients included: putting off or forgetting to obtain blood samples, resistance to causing the child discomfort by collecting the blood sample, not liking to poke the child (inflict discomfort), lack of time to complete diet records, increased appetite of the child, food-sneaking behavior by the child, and the child's difficulty to adhere to the restrictive diet. General life stressors that our parents identified that interfered with adherence were: the addition of another child (birth of a sibling), lack of employment, difficulties with finances, parental marital status (single, other parent in jail), lack of telephone and transportation, parent's work and school schedule, and multiple children for one caregiver. Only one family was unable to state any barriers to

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