Research Article

The Effect of Phenylketonuria on the Quality of Life of the Affected children, Adolescents and their Parents at Minia Governorate.

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Abstract
Background: Phenylketonuria (PKU) is an inborn error of metabolism that results in decrease metabolism of the amino acid phenylalanine (phe). This error causes increased PHE blood concentrations and leads to a toxic accumulation in the brain, resulting in cognitive deficiencies, emotional disturbance and psychosocial disabilities. Therefore, PKU patients must follow the lifelong long protein restricted diet. It is possible that PKU affects PKU patients' quality of life negatively. **Objectives: Aim of the study:** We aimed in this study to assess the impact of phenylketonuria on quality of life of PKU children. **Methods:** This study was conducted on children and adolescents less than 18 years who were diagnosed as PKU Patient over the period from April to October 2019. PKU patients were classified into three group : **Group I:** children between 2 and 9 years, **Group II:** children between 9 and11 years and **Group III:** adolescents between 12 and 17 years. The three groups were subjected to complete history taking, through clinical examination and laboratory investigations determining serum phe level . **Results:** Most of the individuals were male (55.2%) with mean age 5.72±3.80 years. Most of them have normal BMI (85.1%), but 14.9% of children were overweight .(65.7%) were under height, and (34.3%) had normal height. Our results showed that the mean phenylalanine level in all studied group was 507.54±279.24, there is no significant differences between groups in the mean phenylalanine level ($p > 0,05$). **Conclusion:** Phenylketonuria (PKU) have more negative effects on quality of life of children and younger individuals.
Keywords: Phenylketonuria, Quality of life, Metabolic disorders, Nutrition.

Introduction

Phenylketonuria (PKU) is an inherited metabolic disease which affects cognitive functions due to an inability to metabolize phenylalanine which leads to the accumulation of toxic by-products (Phe) in the brain. PKU can be effectively treated with a low phenylalanine diet, but some cognitive deficits remain [(1)].

PKU is the most common inborn error of amino acid metabolism in Egypt with a relatively higher incidence of 1/3000 (0.03%), which means that about 333 neonates are affected with PKU every year as one million babies are born yearly [(2)].

The need to adhere to a strict diet, frequent blood sampling to monitor the levels of (Phe) and regular visits to health services, can affect daily life and therefore have a negative impact on the health-related quality of life (HRQ oL) of individuals affected by PKU [(3)].

It seems that the National PKU screening will provide a big chance to diagnose the PKU patients very early in life, so as to start the special diet and to prepare the family and the patient for a lifelong disease [(4).]

We aimed in this study to assess the impact of phenylketonuria on quality of life of PKU children .

Patients and Methods

This descriptive cross-sectional hospital based study was conducted on children and adolescents less than 18 years who were diagnosed as PKU Patient attending pediatric outpatient clinic of Maternity and children's Hospital of Minia university, Minia Governorate.

The study was conducted over the period from April to October 2019.

Our patients were classified into 3 groups;

Group I: children between2 and 9 years.
Group II: children between 9 and11 years.

Group III: adolescents between 12 and 17 years
Then included children were subjected to the following:

a- Careful history taking including:

Name, Gender, Age, Socioeconomic state, Educational level, Developmental history (Motor, mental), Past history of other co-morbid medical illness., Family history about PKU .

b- General Examination:

* Weight (kg): using digital scale, recording the weight to the nearest decimal fraction.
* Height (cm): is measured in the standing position using astadiometer.
* BMI (kg/m²).

c- Systemic examination:

* Chest Examination.
* Cardiac Examination.
* Abdominal and Pelvic Examination.
* Neurological Examination.

d- Laboratory investigation:

Assessment of serum phenylalanine level.

Results

The study included (67) children between age of (2-17) years. They were classified into 3 groups: First group included 46 patients, their age ranged (2-8) years.
Second group included 15 patients their age ranged (9-11) years.
Third group included 6 patients their age ranged (12-17) years.`
The majority of participants were male (55.2%) with mean age 5.72±3.80 years.
Most of them have normal BMI (85.1%), but 14.9% of children were overweight.
(65.7%) were under height, and (34.3%) had normal height. **(Table 1).**
Our results showed that the mean phenyl-alanine level in all studied group was 507.54± 279.24 **(Table 2),** the mean phenylalanine level in group I was 477.39± 250.92, in Group II was 557.07±310.55 and in group III was 614.83±403.7 **(Table 2).**
Our results showed that there is no significant differences between groups in the mean phenylalanine level (p > 0,05) **(Table 2).**

Table (1): Basic characteristics of study participants

	Total (n=67)	GroupI (2-8 years) (n=46)	Group II (9-11 years) (n=15)	Group III (12-17 years) (n=6)
Age				
Median (Q1-Q3)	4 (2.8-9.4)	3 (2.5-4)	9 (9-10)	13.5 (12.6-14)
Mean±SD	5.72±3.80	3.37±1.07	9.71±0.65	13.77±1.77
Sex				
Male	37 (55.2%)	21 (45.7%)	11 (73.3%)	5 (83.3%)
Female	30 (44.8%)	25 (54.3%)	4 (26.7%)	1 (16.7%)
BMI (kg/m2)				
Median	16.64	15.6	18.06	28.29
(Q1-Q3)	(15.18-18.56)	(14.2-17.42)	(16.95-19.07)	(18.88-30.73)
BAZ				
Median	0.44	0.1	0.95	2.19
(Q1-Q3)	(-0.47 to 1.41)	(-1.01 to 1.41)	(0.44 to 1.34)	(0.60 to 2.86)
<-2SD	0 (0%)	0 (0%)	0 (0%)	0 (0%)
-2SD to +2SD	57 (85.1%)	39 (84.8%)	15 (100.0%)	3 (50.0%)
>2SD	10 (14.9%)	7 (15.2%)	0 (0.0%)	3 (50.0%)
HAZ				
Median	-2.53	-2.93	-1.95	-2.42
(Q1-Q3)	(-3.26 to -1.61)	(-3.66 to -2.01)	(-2.37 to -1.36)	(-2.83 to -1.28)
< -2SD	44 (65.7%)	35 (76.1%)	5 (33.3%)	4 (66.7%)
-2SD to +2SD	23 (34.3%)	11 (23.9%)	10 (66.7%)	2 (33.3%)
> 2SD	0 (0%)	0 (0%)	0 (0%)	0 (0%)

- **Mean±SD: mean ± standard deviation**

Table (2): Comparison between serum phenylalanine in the studied children.

	Total (n=67)	GroupI (2-8 years)	Group II (9-11 years)	Group III (12-17 years)	p value
Serum phenylalanine					0.561
Median (Q1-Q3)	450(280-700)	468 (265-640)	450 (318-816)	580 (340-989)	
Mean±SD	507.54±279.24	477.39±250.92	557.07±310.55	614.83±403.7	

-*: Significant level at P value < 0.05
-Mean± SD: mean ± standard deviation

Discussion

PKU was common cause of severe intellectual disability which is promptly diagnosed and effectively treated now thanks to new born screening programs [5].

New born screening programs are extremely beneficial for preventing late diagnosis of PKU. Despite the fact that there are beautiful developments, there is limited study for PKU patients in terms of quality of life.

The aim of present study was to evaluate patients with PKU in terms of quality of life, Questionnaires specially developed for PKU patients by Regnault et al.,[6] were used. A total of 67 patients with PKU between 2 and 17 years participated in study.

As regards description of characteristics of study participants, showed that Majority of participants was male (55.2%) **(table 1)** This results was similar to the study done in Sohag University Hospital-Upper Egypt(boys more common than girls (62.5% versus 37.5%) [7].

The mean age of participtants was 5.72±3.80 years which was lower than that in Karimzadeh et al.,. study (8.5 ± 6.2) years [8].

Most of studied children have normal BMI (85.1%), but 14.9% of children were overweight. Concerning the BMI, our results were similar to Ilgaz, et al.,. study [9].

In the study of Dokoupil, et al., Patients with PKU have attendency to be overweight relative to subjects without PKU,[10]. But in study done in turkey 55% was under weight (Alptekin, et al.,[11]

65.7% of our patients have aheight for age < -2SD, but (34.3%) were normal height, These results were comparable to other studies (Enns, et al.,.[12] and (Ilgaz, et al.,.[9].

Patients with PKU showed reduced height and growth rates, compared with the reference group, Our explanation for this affection of height for age in our pku patients may be due to the kind of protein they received for supplement as compared to reference group receiving intact protein.

No previous studies showed either severe malnourishment or a higher percentage of overweight or obesity in patients with PKU [13].

The present study showed that the mean phenylalanine level was (507.54±279.24 mmol/L) (8.4±4.6mg/dl) **(table 2),** but in study done in sohag was (15.80±8.84mg/dl 1).[7]

Conclusions

Phenylketonuria (PKU) have more negative effects on quality of life of children and younger individuals.

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