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Effect of Instructional Guidelines on Mothers' knowledge, Practice and Lifestyle of their Children with Phenylketonuria

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Introduction

Background: Phenylketonuria is an autosomal recessive condition in which phenylalanine accumulates in the blood and bodily fluids due to a lack of Phehydroxylase activity. **The study aimed** to evaluate the effect of instructional guidelines on mothers' knowledge, practice and lifestyle of their children with phenylketonuria. **Design:** A quasi-experimental research design was utilized to achieve the study's aim. **Setting:** The study was conducted at genetic counseling clinic in the basic health care unit at Sohag City affiliated to the Ministry of Health (MOH). **Sample:** A purposive sample included 100 mothers accompanied by their children with Phenylketonuria. **Tools**: Two tools were used: **Tool I:** Mothers' knowledge and reported practice regarding phenylketonuria: a structured interview questionnaire was utilized and **Tool II:** Children's lifestyle assessment questionnaire. **Results:** The study revealed that there was a statistically significant improvement in post-intervention mothers' knowledge and reported practices and their children's lifestyle with phenylketonuria than pre-intervention. A statistically significant correlation detected between compliance with the prescribed diet and growth problems. No statistically significant correlation found between compliance with the prescribed diet and sleep problems. **Conclusion:** The study concluded that the instructional guidelines were effective in improving mothers' knowledge, reported practice, and their children's lifestyle with phenylketonuria. **Recommendations:** Health education program for mothers should be provided to increase their knowledge and reported practices about phenylketonuria, as well as identify the importance of a prescribed diet for children with this condition.

Keywords: Children, Knowledge, Practice, lifestyle, Mothers & Phenylketonuria.

Introduction:

Phenylketonuria (PKU) is an inherited condition characterized by phenylalanine (Phe) buildup in the blood and bodily fluids due to a lack of Phe hydroxylase activity. Classical PKU (Phe N 1200 mol/L), mild PKU (Phe 600–1200 mol/L), and mild hyperphenylalaninaemia (Phe b 600 mol/L) are the three types of Phenylalanine hydroxylase (PAH) deficiency. The prevalence of PKU varies with ethnicity. PKU affects roughly one in every 10,000 white European births in the UK, with around 70 babies born with PKU each year, implying that over 6000 people in the UK have PKU (**Williams et al., 2018**). Excess phenylalanine in the blood can be caused by mutations in the PAH gene, which result in diminished activity or full inactivation of the PAH enzyme. This can result in severe and irreversible intellectual disability if untreated (**Said & Draz 2019**) A provisional diagnosis of Metachromatic Leukodystrophy (MLD) / Aminoaciduria / Leigh syndrome, with urine and serum amino acid tests, thyroid hormone stimulating tests, creatinine phosphokinase (CPK), nerve conduction study (NCS), and serumlactatepyruvate tests recommended. Physiotherapy and anti-epileptic medicines are being used. During the second month after birth, the child was admitted to the outpatient department with global developmental delay, right focal seizures, impaired visual perception, and a lack of social grin, head control, and rolling eyes **(MacDonald, et al., 2017**)

If the child is not treated, elevated Phe concentrations in the blood for an extended period can cause significant cognitive impairment, seizures, behavioral issues, and autism-like symptoms. Such irreparable consequences could be averted by starting a low-Phe diet as soon as possible following diagnosis and continuing it throughout life. The synthetic formulation of tetrahydrobiopterin (BH4), sapropterin dihydrochloride, and medical food or amino acid supplements, which comprise formulations of major neutral amino acids such as tryptophan and tyrosine, are currently accessible as pharmacological therapy (**Hanley, 2017)**

Blood tests should be done at home by parents every week for children under the age of five and between twice monthly and monthly for children aged six. As a result, several parts of PKU management appear to be time-consuming for caregivers who are also responsible for normal childcare. PKU has previously been found to have an impact on caregiver's lives, including their ability to continue regular work duties, with 11% of parental caregivers saying that they had to quit working and 20% reporting that they had to change employment to fulfill the child's demands in one survey (**Loeber, 2017**).

This treatment plan's adherence is taken into account. Children supervising the intake of Phe-free L-amino acid supplements are demanding since they need to purchase specialized diets. Daily Phe consumption planning, low-Phe meal preparation (sometimes needing additional cooking), Phe intake monitoring, and clinic visits (**Hanley, 2017).**

Until November 2015, the Egyptian Ministry of Health and Population did not conduct a national screening program for PKU. As a result, the moms had some information and experience with phenylketonuria. Children with untreated PKU appear normal at birth, but lose interest in their surroundings by the age of 3 to 6 months. Seizures and tremors, difficulties in executive functioning, psychiatric and behavioral concerns, social difficulties, poor growth, irritability, and dermatitis are all symptoms of increased blood Phe in PKU children. Children are developmentally delayed by the age of one year. If left untreated, it is usually severe, and most children will require institutional care. Dietary and/or pharmaceutical therapy are used to reduce blood Phe concentrations in PKU patients **(Khaton, 2016).**

PKU is treated with a low-Phe diet that significantly restricts natural protein intake to maintain blood Phe concentrations under control. Natural foods are included in the low-Phe diet. Fruit, various vegetables, fats, and oils, as well as specially manufactured low-protein items like low-protein flour, pasta, and bread, are all low in protein. Phe-free L-amino acids are required for all patients on dietary treatment, which are frequently supplemented with additional carbohydrates, with or without fat, vitamins, and minerals. Dietary therapy compliance is critical, which necessitates careful planning, dietary supervision, and monitoring by the caregiver **(Blau, et al., 2018).**

Nurses play a vital role in the assessment of PKU children and give health education to the general public about the condition, which includes early case detection to avoid complications, nutrition, and diet restrictions, and referral for treatment of Phenylketonuria disease. During the preconception stage, they provide premarital counseling about consanguineous marriage to identify if parents carry the PKU gene (**Khaton, 2016**). Also, following each clinic appointment, follow-up calls, or e-mails can help examine children with PKU (**Abd-Elkodoos et al., 2018).**

Significance of the study:

PKU is found in different geographic places around the world. With a rate of 1: 4,500, Ireland has the highest incidence rate in Europe. It's also popular in a few sections of China, but it's uncommon in African countries. An incidence of 1: 2,600 has been observed in Turkey **(Ford et al., 2018).** In the governorate of Menoufiya, the prevalence was 1/3000 (**Morad et al., 2019**). In 2017, the incidence rate in Fayoum Governorate was 2.8:10000. The incidence rate in Egypt as a whole was estimated to be 1 in 7500 (or 1.3 in 10,000) (**Gad et al., 2019**).

On the other hand, a preliminary study in Sohag was published in this study, which included 18,000 cases seen in the pediatric clinics of Sohag University hospital over three years, one hundred of which were suspected of having PKU and two of which were confirmed **(Sadek et al., 2018 & Morad et al., 2019).**

Because the gene abnormality is unknown and passed down from generation to generation, Phenylketonuria is a major health concern throughout a child's life and that of their family when harmful protein buildup occurs in people with Phenylketonuria who eat high-protein diets (**Anderson & Leuzzi, 2018**). PKU continues to have negative impacts on children in Upper Egypt, including developmental issues such as mental retardation, convulsions, delayed social and intellectual abilities, delayed weight and length growth, body odor, skin rash, and eczema, as well as behavioral anomalies (**Abdel Rahim et al., 2017**).

Aim of the study:

To evaluate the effect of instructional guidelines on mothers' knowledge, practice and lifestyle of their children with phenylketonuria through:

- Assess mothers' knowledge level regarding phenylketonuria.
- Assess mothers' reported practices level regarding phenylketonuria.
- Assessing lifestyle of children with phenylketonuria.
- Design and implement instructional guidelines based on the mother's and children's needs.
- Investigate the association between demographic characteristics of mothers and their knowledge and reported practice regarding phenylketonuria.
- Determine the effect of mothers' knowledge and reported practice on the lifestyle of their children with phenylketonuria

Research hypothesis:
H1: Mother's knowledge and practices regarding phenylketonuria expected to improve after implementing the instructional guidelines.
H2: The lifestyle of children with phenylketonuria expected to improve after implementing the instructional guidelines.

Subjects and Method:

Research design:
A quasi-experimental research design was utilized to achieve the study's aim.

Setting:
The study was conducted at genetic counseling clinic in the basic health care unit at Sohag City affiliated to the Ministry of Health (MOH), Egypt. These settings were chosen because of the high prevalence of children with phenylketonuria in the selected setting, as well as the fact that it serves the most populous region of the country.

Subjects:
A purposive sample included 100 mothers with ages ranged from 18-to 45 years who agrees to participate in the current study accompanied by their children with Phenylketonuria according to the following criteria; Children aged from one year to 6 years old and free from other chronic diseases.

Sample size:
The sample size was calculated based on a power analysis of 0.95(β=1-0.95=0.5) at alpha .05 (one-sided) with a large effect size (0.5) was used as the significance, 0.001 was used as the high significance.

Tools of data collection:
Two data collection tools were used to carry out the current study.

Tool (I): Mothers' knowledge and reported practice regarding phenylketonuria: a structured interview questionnaire was utilized. It was designed by the researcher after reviewing related literature and translated into the Arabic language it consisted of four parts as the following:

Part (1): Assessment of demographic characteristics of the mothers and their children:
It included 3 open-ended questions:

- Demographic characteristics of the child, such as age, gender, and child ranking.
- Demographic characteristics of mothers include age, relative degree, educational level, and occupation.

Part (2): Assessment of medical history:
It is related to the medical history of children with PKU, including duration, the discovery of the disease, follow up frequency per month, the degree of follow up, history of family members, the relationship of a family member suffering from the disease, complications of the disease and measurement of the child (Height, weight, body mass index and the serum level).

Part (3) mothers' knowledge about phenylketonuria:
It consisted of 16 questions divided into three sections: the first section asked six questions concerning the mothers' knowledge of PKU condition, including definitions, risk factors, causes, early symptoms, latent signs, and sequelae. The second section included four questions about artificial feeding, including the temperature at which free phenylalanine formula should be kept and for how long, the frequency with which the nipple of the feeding bottle should be changed, and the type of pot used to boil water. The final component included five questions concerning the supported services, including a source of dietary plan, restricted foods, contraindicated medications, and a question about social support **(Abd-Elkodoos et al., 2018; Cazzorla et al., 2018; & Elsayed et al., 2020).**

Scoring system of knowledge:
The overall score for knowledge was 37 points; answers to questions were either correct or incorrect, with one point awarded for correct answers and zero for erroneous answers and I don't know. There were a few questions that had several answers. Each component was added together and converted to a percentage. Mothers' knowledge was divided into three categories: poor knowledge (less than 50% of total score), fair knowledge (50 % to 70% of total score), and high knowledge (greater than 70% of total information) (**Abd-Elkodoos et al., 2018**).

Part (4) mothers' reported practice about phenylketonuria:
It included 30 statements in three sections: Formula preparation (4 statements), feeding process (2 statements), dietary record (4 statements), making baked goods for the PKU children (11 statements), follow up (1 statement), hair and nail care (2 statements), hand wash (2 statements), clothing and bathing (2 statements), and skincare (2 statements) **(MacDonald et al., 2017; Abd-Elkodoos, et al., 2018; Elsayed et al.,2020).**

Scoring system of reported practice:
The total score was 30 points, with one point for each statement. The checklist's answers were either done or not done, with one mark for done and zero for not done. Each component was added together and converted to a percentage. The practices of mothers were categorized as either satisfactory (60 % or more) or unsatisfactory (less than 60 %) **(Fouad and Elmoneem 2016; Abd-Elkodoos et al., 2018; Khdair et al., 2021).**

Tool (II) Children's lifestyle assessment questionnaire for children with PKU:

It was adapted from **National Society for Phenylketonuria, (2019)** and was used to assess existing strengths in a child's health, any risk factors that may be present, and highlight recommendations that may be worth considering. It was divided into nine sections: general information, maternal health during pregnancy, family history and information, the first few years of a child's life, past and present health concerns, general diet information, an atypical day for a child, child patterns of behavior, and a review of the child's physical system.

Scoring system:

Scoring system for each of the knowledge items, almost always (2 points) - some of the time (1point) - hardly ever (0 point)". For each area of knowledge, it was considered satisfactory if the percent score was 60% or more and unsatisfactory if less than 60%.

Validity of the tools:

The content of the tools and instructional guidelines was evaluated for clarity, thoroughness, appropriateness, and relevance. To ensure that the questionnaire was clear and relevant to PKU, five expert professors with more than ten years of experience, two professors in Pediatric Nursing, one professor in Community Health Nursing, and two professors in Medical Biochemistry from Sohag University examined and revised the content validity of the tools. Based on the panel's recommendation, no adjustments were done.

Reliability of the tools:

The reliability of the tools was tested using Cronbach's Alpha for the Interviewing questionnaire was 0.878 and the lifestyle assessment Questionnaire for children with PKU was 0.815.

Data collection procedure:

Data collection was carried out in three phases:

Preparatory phase:

Design the study questionnaire for data collection, including analyzing the existing literature in various sections of the review utilizing textbooks, publications, different studies, the internet, and journals. The health directors received an official consent letter from the Dean of the Faculty of Nursing, Sohag University, to conduct the study.

Pilot study:

Before beginning data collection, a pilot study was conducted on 10% of the total sample (10 mothers and their children) in the study. The pilot research data was collected around the middle of January 2021. The goal of this study was to see how clear the tools were and estimate how long it would take to fill out the questionnaire based on the findings of the pilot study; no changes to the tools were done.

The study followed standard clinical research ethics guidelines. The mothers were informed that there would be no risk to them during the research, that they could decline or participate at any time, and that they had the right to refuse or participate. During data collection, the privacy of study participants was taken into account. The researcher also met with the mothers to explain the study's goal and gain their consent to participate. They were assured that the acquired data would be kept anonymous and secret and that it would only be used for the scientific study. Subjects were guaranteed the ability to withdraw from the study at any time.

Data collection phase (Fieldwork):

Data were collected from February to August 2021, three days per week for the mothers who were being investigated, and typically 5-6 questionnaires were collected every day. The questionnaire took between 30-and 35 minutes to complete. Before enrolling the mothers in the study, verbal consent was obtained from each of them, and the study objectives were thoroughly done.

The purpose and significance of the research project were discussed after selecting the sample that matched the inclusion criteria.

The data collection tools were given to the mothers twice: (1) as a pre-test to examine their knowledge, practices, and lifestyle before adopting instructional guidelines, and (2) as a post-test to assess their knowledge, practices, and lifestyle of children after the instructional guidelines have been implemented.

After evaluating the associated research and assessing the real needs of the investigated mothers, the simplified booklet was used as a supportive resource and delivered to the mothers in the Arabic language to cover all points related to knowledge and practice of PKU. Lectures, discussions, photographs, and posters were all employed as instructional methods. The researchers created and implemented PKU educational guidelines that included both a theoretical and practical component. The mother's knowledge of PKU was incorporated into the theoretical portion. Lectures, posters, educational videos, scenarios, and role-plays were used to implement it. The patients were given an instructional pamphlet about PKU written in plain Arabic with informative photos provided by the researchers.

The subject contents have been sequenced through 5 sessions (3 sessions for the theoretical part and 2 sessions for the practical part), and each session took about 25-30 minutes. The total time was 2 hours for each one. At the beginning of the first session, an introduction to the educational guidelines regarding PKU was given and each session started with summary feedback about the previous session.

The instructional guidelines included knowledge regarding PKU as follow:

- Definition of PKU
- Risk factors of PKU

- Causes of PKU
- An early symptom of PKU
- Latent symptoms of PKU
- Complications of phenylketonuria.
- Artificial feeding such as temperature that keeps and duration for use of free phenylalanine formula
- Period of changing nipple of feeding bottle and lastly kind of pot to boil water.
- Supported services as the source of nutritional plan, prohibited foods, contraindicated types of medications

The practical part contained information regarding PKU. The interview took approximately 25-35 minutes for each mother to answer and fill out the questionnaire to assess the practices of the studied mothers. It was implemented through lectures, posters, and educational films.

The instructional guidelines included practices regarding PKU as follow:

- Formula preparation
- Feeding process
- Dietary record
- Making baked goods for the PKU children
- Follow up
- Hair and nail care
- Hand wash
- Clothing and bathing
- Skincare

Evaluation: occurred after three months, each mother was re-interviewed to assess their knowledge, practices, and children's lifestyle. Re-assessment of the mothers was done using the same pre-test tools.

Administrative Design:

An official letter requesting permission to conduct the study was directed from the dean of the faculty of nursing at Sohag University to the directors of the genetic counseling clinic in the basic health care unit in Sohag affiliated to MOH to obtain their approval to carry out this study.

Statistical Design:

Upon completion of data collection, the data was reviewed, prepared for computer entry, coded, scored, tabulated, and analyzed by computer using the "statistical package. Descriptive statistics (i.e., frequencies, percentage ….etc.) were done using the computer program SPSS version 20. Frequency and percentage were used for numerical data also, Fisher exact test was used. The Chi-square test was used to compare differences in the distribution of frequencies among different groups. It is considered * significant when P-values were less than 0.05 or (P< 0.05).

Results:

Table (1): Distribution of the studied children regarding their demographic characteristics (n=100)

Demographic characteristics	No	%
Age: (years)		
<2	26	26.0%
2 - < 5	44	(44.0%)
≥5 years	30	(30.0%)
Mean ± SD	3.35 ± 3.64	
Gender:		
Boys	58	(58.0%)
Girls	42	(42.0%)
Child ranking:		
First	29	29.0
Second	39	39.0
Third or more	32	32.0
Child weight		
9:<13 kg	64	64.0
13:<17 kg	36	30.0
≥17 kg	6	6.0
Child length		
<75 cm	14	14.0
75 <95 cm	36	36.0
95 < 105 cm	27	27.0
≥105 cm	23	23.0
Body mass index		
Normal	44	44.0
Less than normal	24	24.0
More than normal	14	14.0
Obese	18	18.0

Table (2): Distribution of the medical history of children with PKU (n=100)

Medical history	No.	%
Effect of the disease on general health		
Yes	53	53.0
No	47	47.0
Effect of the disease on general health (no=53)		
Delayed growth of weight and length	23	44.0
Behavioral problems	6	11.0
Delayed social and intellectual skills which obtained from child's medical record	9	17.0
Skin Turgor	1	2.0
Convulsions	2	4.0
Bad odor of the body	1	2.0
All of the above	11	20.0
Presence of complications of the disease		
Yes	38	38.0
No	62	62.0
Complications (no=38)		
Mental retardation	11	29.0
Behavioral problems	6	17.0
Delayed social and intellectual skills	6	15.0
Convulsions	10	25.0
Skin rash and eczema	5	14.0
Types of regular investigations		
Phenylalanine level in the blood	100	100.0
Urine analysis	0	0.0
CBC	0	0.0
All of the above	0	0.0
Number of investigating Phenylalanine levels in the blood		
Once monthly	44	44.0
Once every two months	48	48.0
Once every three months	8	8.0

Table (3): Distribution of the studied mothers regarding their demographic characteristics (n=100)

Demographic characteristics	No.	%
Age in years		
- 18 < 30	64	64.0
- 30 – 40	36	36.0
Mean ±Stander deviation	22.43 ± 5.36	
Educational level		
- Read and write	26	26.0
- Secondary education	39	39.0
- University education	35	35.0
Occupation		
- Working	25	25.0
- Housewives	75	75.0

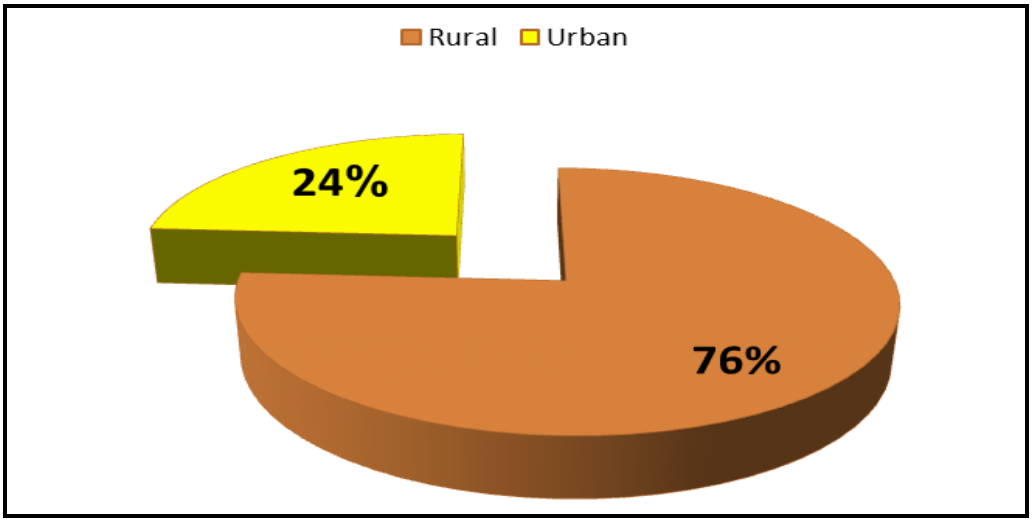

Figure (1): Distribution of the studied mothers according to their residence (n=100)

Table (4): Distribution of the studied mother's correct knowledge regarding phenylketonuria pre and post instructional guidelines' implementation (n=100)

Mother's knowledge	No =(100)		P-value
	Pre instructional guidelines' implementation(%)	Post instructional guidelines' implementation(%)	
Definition of phenylketonuria	19%	88%	<0.001*
Risk factor of phenylketonuria	29%	87%	<0.001*
Causes of phenylketonuria	18%	84%	<0.001*
Early symptoms of phenylketonuria	33%	82%	<0.001*
Latent symptoms of phenylketonuria	17%	92%	<0.001*
Complications of phenylketonuria	20%	85%	<0.001*
Thesupported services asa source ofnutritional plan	25%	100%	<0.001*
Prohibited foods	37%	89%	<0.001*
Types of medications	38%	87%	<0.001*
Artificial feeding such as temperature that keep and duration for use of free phenylalanine formula	53%	92%	<0.001*

**highly significant at <0.001 levels* *Chi square test was used*

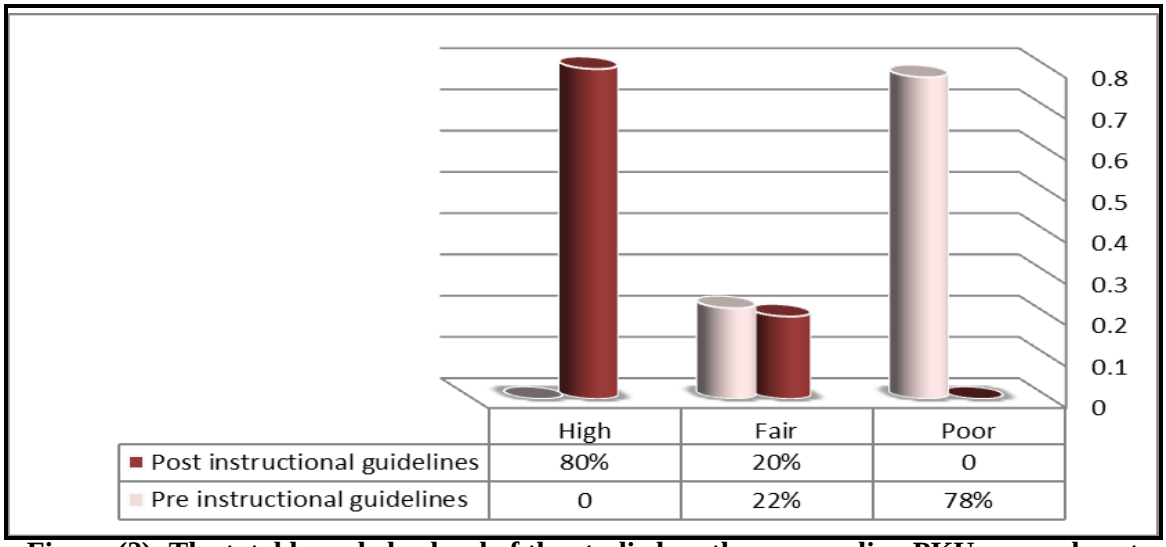

Figure (2): The total knowledge level of the studied mothers regarding PKU pre and post instructional guidelines' implementation

Table (5): Distribution of the studied mother's reported practices regarding phenylketonuria in the pre and post instructional guidelines' implementation (n=100)

Mothers reported practices	No = (100)		P-value
	Pre instructional guidelines' implementation (%)	Post instructional guidelines' implementation (%)	
Formula preparation	17%	87%	<0.001*
Feeding process	19%	83%	<0.001*
Dietary record	18%	89%	<0.001*
Making baked goods for the PKU children	13%	82%	<0.001*
Follow up	14%	90%	<0.001*
Hair and nail care	22%	86%	<0.001*
Hand wash	45%	100%	<0.001*
Clothing and bathing	67%	100%	<0.001*
Skincare	78%	99%	<0.001*

***highly significant at <0.001 levels* *Chi square test was used*

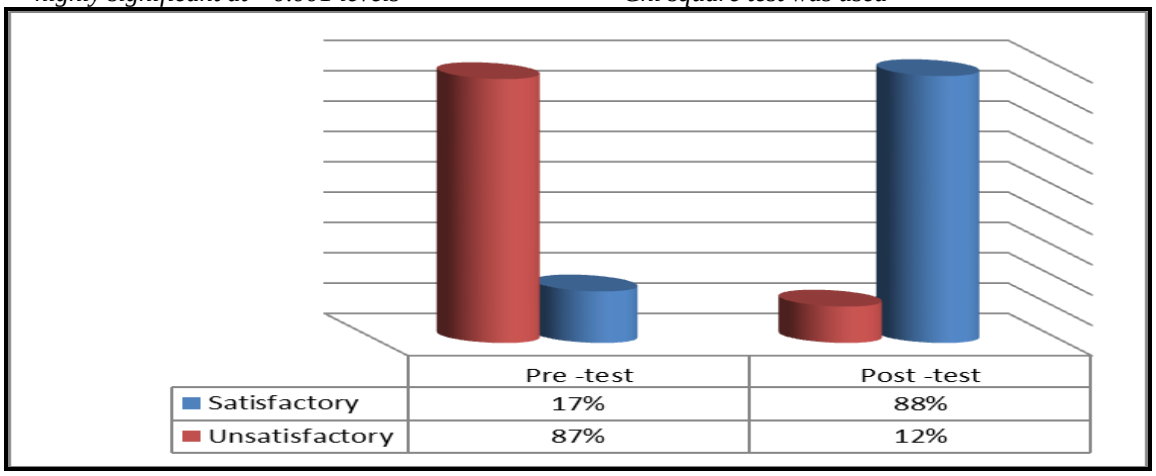

Figure (3): The total practices level of the studied mothers regarding PKU pre and post instructional guidelines' implementation

Table (6): Distribution of the studied children according to their nutritional lifestyle (n=100)

Items	%
Age of child when taking solid feeding	
< One year	41.0
≥ One year	59.0
Number of the main meals per day	
Two meals	33.0
Three meals and more	67.0
Time of the largest meal	
Breakfast	21
Lunch	65.0
Dinner	23.0
Un-preferred food for children	
Yes	41.0
No	59.0
Last meal of the day	
From 6 pm to 10 pm	71.0
After 10pm	29.0
Receiving adequate fluids	
Yes	69.0
No	31.0
Types of fluids	
Permitted fluids (water – low sugar fruits)	65.0
Not permitted fluids (canned juices)	35.0

Table (7): Distribution of the studied children according to their prescribed dietsystem (n=100)

Items	%
Children follow a special diet system	
Yes	83%
No	17%
Foods prepared by:	
Physician or nurse	0
Dietitian	0
The mother	100.0
Type of diet system	
Free from protein	70.0
Free from Fenyl-Alanin	23.0
Low fat	7.0
Compliance with the diet system	
Yes	71.0
No	29.0
Number of dispensing the prescribed diet during the month	
Once	87.0
Twice	9.0
More than two times	4.0
Adequacy of the dispensed amount of the prescribed diet	
Yes	89.0
No	11.0
Dealing to compensate for the adequacy	
Give the child the permitted vegetables and fruits	14.0
The calculation method to measure the amount of foods	
Yes	89.0
No	11.0
Type of calculation method of measuring the amount of foods (n=89)	
From the metabolic clinic	49.0
15 cups for every 8 meals	40.0
Place of keeping the prescribed diet	
In the freezer	4.0
Out of refrigerator	89.0
In a special place in the refrigerator	7.0

Table (8): Correlation between compliance with the prescribed diet and the effect of the disease on general children's health

Variables	Compliance with the prescribed diet	
Effect on general health	**R** **P**	**0.773** **0.000****

(**) Highly statistically significant correlation at P-value <0.05

Table (9): The total lifestyle level of the studied children with PKU pre and post instructional guidelines' implementation (100)

Lifestyle of children	Pre instructional guidelines implementation		Post instructional guidelines implementation		T	P-value
	No	%	No	%		
Satisfactory	37	37.0	86	86.0	125.34	<0.001*
Unsatisfactory	63	63.0	14	14.0		

***highly significant at <0.001 levels* *Chi square test was used*

Table (10): Correlation between total studied mothers' knowledge, practices, and children's lifestyle with PKU Using Pearson Correlation Coefficient

Variable	Correlation coefficient	P-value
Knowledge- reported practice	0.446	<0.001*
Knowledge- children' lifestyle	0.227	<0.001*
Reported practice - children' lifestyle	0.228	<0.001*

**Correlation significant at <0.001 levels*

Table (1): Depicted the children's demographic features. In terms of age, 44% of them were between the ages of 2 to < 5years, with a Mean ±SD of 3.35 ±3.64, and (58%) of them were males. In terms of kid ranking, 39 % of the studied children were ranked the second. In addition, the table revealed that less than two-thirds of children's weight (64%) was 9<13kg, and more than one-third of children's height (36%) was 75<95 cm. and less than half of children (44 %) had a normal body mass index (BMI).

Table (2): Showed that disease affected 53% of children's general health such as; delayed growth of weight and length, behavioral problems, delayed social and intellectual skills, Skin related disease, Convulsions, and bad odor of the body. Less than two thirds of children (62%) had no complications of the disease. All children (100%) were made Phenylalanine levels in the blood as regular investigation. Less than half of children (44%) are making the Phenylalanine levels in the blood once every two months.

Table (3): Represented that 64% of the studied mother's their age ranged between 18 < 30 years with a mean ± SD of 22.43 ± 5.36, (39%) of them had secondary education, meanwhile, it is pointed out that 75% of the studied mothers were housewives.

Figure (1): Demonstrated that (76%) of the mothers are living in rural areas and 24% of them were from urban areas.

Table (4): Demonstrated frequency and percentage distribution of the studied mothers' correct knowledge regarding phenylketonuria. It was observed that there was an improvement with highly statistically significant differences between mothers' correct knowledge regarding phenylketonuria in pre/post instructional guidelines implementation related all items (P<0.001).

Figure (2): Showed that 87% of the studied mothers had poor knowledge regarding PKU while 22% of them had fair knowledge in the pre instructional guidelines' implementation, but post instructional guidelines' implementation, there was a significant improvement and 80% of mothers had high knowledge regarding PKU

Table (5): Portrayed the frequency and percentage distribution of the studied mothers' reported practices regarding phenylketonuria. It was observed that there was an improvement with highly statistically significant differences between mothers' reported practices regarding phenylketonuria in the pre/post instructional guidelines implementation related all items(P<0.001).

Figure (3): Clarified the total reported practices level of the studied mother's in pre and post instructional guidelines' implementation. It was noticed that (87%) of the studied mother had unsatisfactory practices level about PKU pre- instructional guidelines implementation and decreased to become 12% in post- instructional guidelines implementation. Reversely, 17% of the studied mothers had satisfactory practices level regarding PKU pre-implementation compared to 88 % of them in the post-implementation.

Table (6): Showed that after one year, (59%) of children were eating solid food, (67%) of them were eating three meals per day, and (65%) of the largest meal was lunch. Carbohydrates (rice – beads) were favored by less than half of the children (43%). More than half of children's favored foods (59%) were part of a specified diet. Concerning Children's last meal of the day (71%) was between 6 and 10 p.m., (69%) received appropriate fluids, and (65%) received authorized fluids (water – low sugar fruits).

Table (7): Revealed that the majority of children (83%) followed a particular diet. All of the mothers were cooked for their children, and the majority of them (70%) followed a low-protein diet. The diet approach was followed by less than three-quarters of the children (71 %). According to the table, (87%) were dispensing the specified diet once a month, and (89%) were dispensing the prescribed diet in a certain amount. The majority of children's mothers (89%) used a mathematical method to measure the amount of food their children ate, and 49% of children's mothers used the metabolic clinic to calculate the amount of food their children ate. The majority of mothers (89%) did not maintain the prescribed diet in the refrigerator.

Table (8): Showed that there is a highly statistically significant correlation between compliance with the prescribed diet and the effect of the disease on general children's health (P value 0.000).

Table (9): Portrayed that (86%) of the children with PKU their lifestyles have been improved and become satisfactory lifestyle post instructional guidelines implementation compared to 37% pre- guidelines implementation.

Table (10): Correlation revealed significant positive linear correlations between knowledge- reported practice ($r = 0.446$, $p < 0.001$), knowledge- children's lifestyle ($r = 0.227$, $p < 0.001$), and reported practice - children's lifestyle ($r = 0.228$, $p < 0.001$). This result confirms the positive relationship between the mother's knowledge, reported practice, and children's lifestyle with PKU.

Discussion:

Phenylketonuria (PKU) is a common chronic genetic disorder that necessitates a caregiver's daily effort to maintain a particular food program, rehabilitation, and deal with the child's difficulties (**Elsayed et al., 2020**).

The current study discovered that less than half of the children tested were between the ages of 2 to less than 5 years and that less than half of them had a normal body mass index. This finding is consistent with the findings of **MacDonald et al. (2017)**, who investigated "The personal burden for caregivers of children with phenylketonuria" in a cross-sectional study in the United Kingdom. Molecular genetics and metabolism reports revealed that the majority of participants indicated that their children's weight ranged from 9 to 13 kg, with half of the youngsters having a normal BMI. This finding matches that of **Al-Zyoud et al. (2019),** who investigated "Cultural gut microorganisms lack Escherichia coli in children with phenylketonuria" and found that half of the children's height was 70<90cm.

In terms of medical history, less than one-fifth of PKU children exhibited impaired intellectual skills, according to the findings. More than two-fifths of children with phenylketonuria exhibited higher brain electricity, according to **Abd-Elkodoos, et al., (2018**). These findings were also consistent with those of **Gad et al. (2019**), who investigated "Pediatric Phenylketonuria in Fayoum Governorate" and found that the children had a history of motor developmental history, motor delay, convulsions, and autism. In addition, **Roby et al., (2021**), who conducted "Assessment of Lifestyle for Children with Phenylketonuria," discovered that the disease affected fewer than half of children's overall health; including delayed weight and length, behavioral problems, delayed social and intellectual skills, skin related disease, convulsions, and bad odor of the body.

Regarding distribution of the effect of the disease on studied children's general health, the current study revealed that the disease affected more than half of children's general health such as; Delayed growth of weight and length, Behavioral problems, delayed social and intellectual skills, Skin related disease, Convulsions, and Bad odor of the body. The majority of children had no complications of the disease. All children are making Phenylalanine levels in the blood as regular investigation. Less than half of children are making Phenylalanine levels in the blood once every two months, this result may be due to this disease's effect on the normal development of children in their minds and their bodies.

This result also contradicts **Tiele et al. (2019**), who studied "Investigation of pediatric PKU breath malodor, comparing glycol macro peptide with phenylalanine free L-amino acid supplements and found the majority of children's general health problems, including convulsions, delayed weight and length growth, delayed social and intellectual skills, and bad body odor." This result is also consistent with **Walkowiak et al., (2019**) who conducted a study, which looked at "General health in classical phenylketonuria children: A retrospective cohort study." Advances in medical technology have revealed that the majority of children have their blood Phenylalanine levels checked regularly. Every two months, two-thirds of the youngsters check their Phenylalanine levels in the blood.

The current study found an improvement in mothers' knowledge related phenylketonuria, with a highly statistically significant difference between pre and after the adoption of instructional guidelines. This, in the opinion of the researchers, proved the positive impact of instructional guidelines execution, as evidenced by an increase in knowledge of the research issue.

The majority of mothers had poor knowledge before the instructional guidelines were implemented, but after that, there was a considerable change, and the majority of them had high knowledge about PKU. This amplification is in line with **Elsayed et al., (2020**), who did a study titled "Assessment of Mothers Care toward their Children with Phenylketonuria" and found that more than half of mothers were unaware of PKU. Also, according to **Abd-Elkodoos et al., (2018**), slightly less than two-thirds had poor knowledge of PKU, more than one-fifth had acceptable knowledge, and less than one-fifth had good knowledge of PKU. These results reflect the need of those mothers for these instructional guidelines about the proper management of PKU children.

The majority of mothers evaluated had an unsatisfactory practice level before the implementation of the instructional guidelines, according to the current study. The nature of the condition PKU, which is one of the most difficult chronic disorders, could explain these findings. If mothers have a high level of understanding of their children's conditions, their practices toward PKU children will improve (**Abd-Elkodoos et al. 2018 & Elsayed et al., 2020**).

In terms of the nutritional lifestyle of the children analyzed, the current study found that less than two-fifths of the children were eating solid food after one year. More than two-thirds of children ate three meals per day, with lunch being the most important. Carbohydrates (rice–bread) were favored by less than half of the children. While the majority of the children ate their final meal between 6 and 10 p.m. More than two-thirds of the youngsters were getting enough fluids, and the majority were getting only what was allowed (water and low-sugar fruits). This may be because, during the first year of a child's life, the family becomes afraid of food containing PHE, so the mother gives artificial food only after one year

to keep the child healthy; also, the child eats three meals per day, the largest of which was lunch, because the child only takes the formula in the morning, so they become hungry and eat the largest diet at lunchtime, and the lunch meal also contains the preferred food for the child. These findings are consistent with **Demirdas et al. (2017**) study "Micronutrients, essential fatty acids, and bone health in phenylketonuria.

According to a study published in the Annals of Nutrition and Metabolism, one-third of youngsters eat three meals per day, with lunch being the largest. Carbohydrates (rice – beads) were the most popular food among children, and half of them were eating solid food within one year. This finding is consistent with **Tonon et al., (2019)** who did a study about "Food Neophobia in Patients With Phenylketonuria," which indicated that two-thirds of the children were getting enough fluids, and the majority of the children were getting the done fluids (water – low sugar fruits).

Regarding distribution of the studied children towards their prescribed diet system, the current study revealed that the majority of children had followed a special diet system. All of the mothers prepared food for their children. Less than three-quarters of children were taken to a diet system Free from protein. Less than three-quarters of the children Complied about the diet system. The majority of children were dispensing the prescribed diet once every month. The majority of children were dispensed amount of the prescribed diet. The majority of children's mothers were using the calculation method to measure the amount of food, and nearly half of the children's mothers calculate the amount of food from the metabolic clinic. The majority of mothers were keeping the prescribed diet out of the refrigerator. These results may be due to the mothers were interested in the diet system for their children. These results are in agreement with those obtained in a study done by **Medford et al., (2017)** who studied "Diet system influences on treatment adherence for children with PKU": a systematic review and found that majority of children Complied with the diet system, and the majority of children were dispensed the amount of the prescribed diet. This result is in agreement with the study done by **Singh et al. (2016)** who studied "Updated, web-based nutrition management guideline for PKU": evidence and consensus-based approach have they found that three-quarters of children were Using calculation method to measure the amount of foods, and all of the mothers were keeping the prescribed diet out of the refrigerator.

Regarding the correlation between compliance with the prescribed diet and affect general health, the current study revealed a highly statistically significant correlation found between compliance with the prescribed diet and its effect on general health. This result could be because the child's PHE ratio is normal, and hence the child's health is unaffected. This finding is consistent with the findings of **Medford, et al., (2017**), discovered a highly statistically significant link between adherence to the prescribed diet and the disease's impact on the children's overall health. **Jani et al., (2017)** found that "Protein consumption and physical exercise are done to body composition in patients with phenylalanine hydroxylase deficiency." They looked at "molecular genetics and metabolism" and discovered a statistically significant correlation between compliance with the prescribed diet and the effect of the disease on the children's general health.

The findings of the current study highlighted that in most of the children with PKU their lifestyles have been improved and become had satisfactory lifestyles post instructional guidelines implementation with a statistically significant difference. The results of the current convey the success of the implementation of instructional guidelines for mothers of children with PKU which met their needs regarding improving their lifestyle, resulting in acceptance of the research hypothesis and objectives.

Results of the current study revealed significant positive linear correlations between knowledge-practice, knowledge- children's lifestyle, and practice - children's lifestyle. This result confirms the positive relationship between the mother's knowledge, practice, and children's lifestyle with PKU. These results mean that the mothers' knowledge affected their practices when the mothers have satisfactory knowledge about their children's condition; this will improve their practices regarding the care of their children with PKU and enhance their caring roles. This explanation goes in the same line as **Ozel et al. (2018**) who mentioned that, when caregivers are provided with the basic knowledge about their children's condition, developmental prognosis, and various treatment approaches, this will assist them in practicing new and healthy behaviors or also can change unhealthy behavior.

Conclusion:

From the result of the present study, it can be concluded that the instructional guidelines were effective in improving mothers' knowledge, practice, and their children's lifestyle with phenylketonuria.

Recommendations:

In the light of the results of this study, the following recommendations were suggested:

- According to the current study, a health education program should be provided for mothers their children's with phenylketonuria regarding a phenylketonuria-specific diet for their children
- Encourage children's regular assessments of a recommended diet to fit their actual demands to improve their children's lifestyles.

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