

Caring for A Child with Phenylketonuria: Struggles and Psychological Well-being of Parents and Children

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Abstract

Background: Phenylketonuria is a genetic disorder resulting from lack or inadequacy of phenylalanine hydroxylase enzyme activity. Caring for and making nutritional decisions for a child with phenylketonuria puts a great deal of stress on the parents. Moreover, the financial and psychological burden of the disease as well as its chronic nature could affect the psychological well-being of both parents and their children with phenylketonuria. **Aim:** to assess the struggles and psychological well-being of parents and their children with phenylketonuria. **Subjects & Method:** a correlational descriptive study design was conducted on of 145 parents and their children with phenylketonuria. **Tools:** three tools were used to collect the necessary data for the current study, tool (I): Self-administered questionnaire to collect bio-socio-demographic data, tool (II): Depression, Anxiety, and Stress Scale, tool (III) The Strengths and Difficulties Questionnaire. **Results:** it was found that 17.9%, 29%, and 15.9% of the participant parents have extremely severe levels of depression, anxiety, and stress respectively. The results also reveal that 64.8%, 59.3%, 53.1%, and 58.6% of the studied children have emotional, conduct, hyperactivity, and peer problems respectively. Moreover, 62.8% of the studied children have a negative impact on different areas of their everyday life related to the difficulties they have. **Conclusion:** the studied parents struggle to provide care and special foods for their children with phenylketonuria. Behavioral and psychological difficulties facing children with phenylketonuria affect the psychological well-being of their parents. **Recommendations:** further qualitative studies are needed to provide deep insights into challenges perceived by parents of Phenylketonuria children. Designing and implementing training programs for parents to improve their coping strategies and psychological well-being is mandatory.

Keywords: Phenylketonuria, Struggles, Psychological well-being, Difficulties, Parenting.

Introduction

Phenylketonuria (PKU) is an autosomal recessive metabolic disorder characterized by phenylalanine hydroxylase (PAH) deficiency and abnormally high phenylalanine (Phe) concentrations that impair brain function. This brain malfunction leads to significant intellectual impairment, epilepsy, and behavioral issues if left untreated⁽¹⁾. In PKU body's inability to eliminate excessive phenylalanine can cause growth retardation and catastrophic brain abnormalities⁽²⁾.

Phenylketonuria (PKU) is caused by one or more inborn mutations in the phenylalanine hydroxylase gene. Loss of phenylalanine metabolism, as a result, needs strict dietary guidance and/or medication to prevent toxic Phe buildup⁽³⁾. If treatment is initiated and sustained early, children's intelligence is maintained within normal ranges with, on average, some poor neurocognitive function⁽⁴⁾. Newborn screening programs are essential in early detection of phenylketonuria⁽⁵⁾.

Children with PKU may have several behavioral problems that are closely related to the severity of the disease and adherence to a Phe-restricted diet from an early age. Hyperactivity, stereotypy, anxiety, signs of autism, and aggressive behavior are behavioral problems often seen in children who did not follow a strict low Phe diet. Early-diagnosed children who adhere to a Phe-restricted diet from an early age may tend to have high Phe levels which are associated with less severe behavioral problems such as low motivation to achieve, low self-esteem, problems in school, and attention

problems. These children may also have difficulty with reasoning, attention, impulse control, and mental flexibility⁽⁶⁾.

Parenting a child with phenylketonuria is incredibly stressful. These parents are always seeking resolutions for their child's illness-related issues. The measures used by parents differ depending on their culture, religion, level of education, and other background considerations⁽⁷⁾. Early in their child's life, parents of PKU children describe severe emotional problems, including bereavement and trauma reactions along with raising their newborn⁽⁸⁾.

At the time of weaning, the process of natural protein restriction and introduction of protein substitutes may impose a huge psychological burden on parents of PKU children. Maternal anxiety tends to increase over time because of child rejection of protein substitutes⁽⁹⁾. Also, agitation and behavioral problems were associated with a higher parental score of anxiety and depression⁽¹⁰⁾.

The primary goal of providing nursing care for the child with phenylketonuria and his family is making sure that the caregiver will be able to provide the appropriate nutritional needs of the infant. The pediatric nurse should offer the needed emotional support to the family during the stressful time of disease diagnosis. Another crucial goal of caring for PKU children is to help the child achieve a maximum level of growth and development through strict control of phenylalanine levels. Educating the family about disease processes, dietary restrictions, getting dietary advice from the child's dietitian

only, and maintaining safety in case of convulsions is the responsibility of the pediatric nurse⁽¹¹⁾.

Operational definition

The word struggles means very difficult tasks that need great effort to be accomplished. In the present study, it describes the hard tasks facing parents in caring for their children with phenylketonuria including physical and psychological burdens, adherence to a low protein diet, as well as, costs of special foods (protein substitutes) these struggles were assessed by the self-administered questionnaire that was developed by the researchers. Also dealing with the child's behavioral problems is also a struggle that was investigated by the strengths and difficulties questionnaire.

Aim: The current study aimed to assess the struggles and psychological well-being of parents and their children with phenylketonuria.

Research questions:

1. What are the struggles facing parents of children with PKU?
2. What are the behavioral and psychological problems of children with PKU?
3. Are parents of children with PKU suffering from psychological problems?
4. Is there a correlation between difficulties facing children with PKU and the psychological well-being of their parents?

Subject and methods:

Research design: A descriptive correlational design was used in carrying out the present study.

Participants and setting: The study was conducted on a convenient sample of 145 parents who have PKU children

from 13 governorates of upper Egypt and lower Egypt. Parents filled out an anonymous online questionnaire shared via social media targeting parents of children aged 4 to 18 years old. In the case of multiple children with PKU, the parent was asked to report on one child only.

Sample calculation

The estimated number of metabolic disorder parents on WhatsApp groups was 418 (Awlad Misr1 (208 parents) from Lower Egypt and Awlad Misr2 (210 parents) from Upper Egypt). The estimated percentage of depression was 17.3% among parents of children with PKU⁽¹²⁾. A confidence level is 95% with a power of study at 80%. The required sample size was 145 parents and their children.

Tools of Data Collection

Three tools were used to collect the necessary data for the present study.

Tool (I): Self-administered questionnaire was developed by the researchers after reviewing recent related literature and used to collect bio-socio-demographic data, it included the following three parts:

Part I: Characteristics of the studied parents such as age, education, occupation, residence, and monthly income.

Part II: Characteristics of children; age, sex, and child's educational grade.

Part III: Medical History and Disease Burden: time of disease diagnosis, positive family history of the disease, type of therapeutic support, child's behavioral problems caused by uncontrolled Phenylalanine levels, the financial and psychological burden of the disease and caring for the child on the studied parent.

Tool II: Depression, Anxiety, and Stress Scale – 21 (DASS-21)

Depression, Anxiety, and Stress Scale – 21 (DASS-21) is a self-administered form that provides on a 4-point scale a measure of individual symptoms indicating depression, anxiety, and stress⁽¹³⁾. DASS-21 was used in the current study to investigate parents' individual perceptions of depression, anxiety, and stress. The rating Likert scale ranges from 0 to 3 as follows: 0 "Did not apply to me at all", 1 "Applied to me to some degree, or some of the time", 2 "Applied to me to a considerable degree or a good part of the time", 3 "Applied to me very much, or most of the time"⁽¹⁴⁾.

Total scores for depression, anxiety, and stress are calculated by summing the scores for the relevant items:

-Depression scale (3, 5, 10, 13, 16, 17, 21) assesses depressive symptoms including dissatisfaction, despair, loss of interest in life, loss of passion, unwillingness to change, and self-criticism.

-Anxiety scale (2, 4, 7, 9, 15, 19, 20) evaluates situational anxiety, the effect of anxiety on autonomic stimulation and muscles as well as personal beliefs about the effect of anxiety.

-Stress scale (1, 6, 8, 11, 12, 14, 18) measures an individual's level of repose, nervousness, restlessness, irritability, impatience, and hyperactivity.⁽¹⁵⁾⁽¹⁶⁾

Tool III: The Strengths and Difficulties Questionnaire (SDQ):

The parent report form of the Strengths and Difficulties Questionnaire (SDQ)⁽¹⁷⁾ was used to investigate behavioral and psychological problems in PKU children. The Arabic double-sided

version with an impact supplement was used to collect the required data in the current study⁽¹⁸⁾. The front side of the SDQ consists of 5 subscales (emotional problems scale, conduct problems scale, hyperactivity scale, peer problems scale, and prosocial scale) each scale has 5 items. The other side of the SDQ is a short impact scale, consisting of 5 questions about the impact of any difficulties on the child and different aspects of his daily life as home life, friendships, classroom learning, and leisure activities⁽¹⁹⁾.

Scoring of SDQ: - Each subscale is measured by 5 items, rated on a 3-point scale. "Somewhat True" is always scored as 1, "Not True" scored as 0, and "Certainly True" scored as 2 except for items 7, 11, 14, 21, and 25. The total score of difficulties ranges from 0 to 40 and is obtained by summing scores from all the scales (emotional problems, conduct problems, hyperactivity, and peer problems) except the prosocial scale. The impact score ranges from 0 to 10 and is generated by summing the items on overall distress and impairment⁽¹⁹⁾.

Pilot study

A pilot study was conducted on a group of 15 parents and their children prior to data collection to assess the feasibility, internal consistency, and reliability of the questionnaire. The DASS appeared to have good internal consistency and reliability (Cronbach's =0.879) and SDQ appeared to have excellent internal consistency and reliability (Cronbach's =0.903).

Ethical Consideration

Approval of the ethical committee at the Faculty of Nursing, Zagazig University was obtained. Before taking

part, the participants gave their consent. Participants who consented to engage in the research were also given the assurance of confidentiality and anonymity of the collected information and that they had the freedom to leave the study at any time.

Fieldwork

The current study was completed within 4 months from March to June 2022. The data was collected through an anonymous online questionnaire created through google forms (https://docs.google.com/forms/d/e/1FAIpQLScO4k9VQDJ4XrOTJQgUAvu7vOF6g-BRzbXjXrFSiksPiEiM1Q/viewform?usp=sf_link) and targeting parents of children aged 4 to 18 years old. In the case of multiple children with PKU, the parent was asked to report on one child only. The questionnaire was distributed to the participants through social media (2 WhatsApp groups for parents of children with PKU). The researchers communicated with the studied parents individually to explain the aim of the study and to take oral approval for sending the link of the questionnaire and the consent to their private WhatsApp. Some parents preferred to be interviewed over the phone rather than to fill out the online questionnaire.

Statistical Analysis

All data were collected, tabulated, and statistically analyzed using SPSS 20.0 for windows (SPSS Inc., Chicago, IL, USA 2011). Quantitative data were expressed as the mean \pm SD, and qualitative data were expressed as absolute frequencies (number) & relative frequencies (percentage). ANOVA (One way analysis of

variance) test and The student "t" test were used for comparison of means. Percent of categorical variables were compared using the Chi-square test or Fisher's exact test when appropriate. Pearson correlation coefficient was calculated to assess relationship between study variables, (+) sign indicates direct correlation & (-) sign indicate inverse correlation, also values near to 1 indicate strong correlation & values near 0 indicate weak correlation. The developed tools were evaluated for their reliability by using Cronbach's alpha coefficient test. P-value $<$ 0.05 was considered statistically significant, p-value $<$ 0.001 was considered highly statistically significant, and p-value \geq 0.05 was considered statistically non-significant.

Results

Table 1 shows that the mean age of the studied parents was 34.38 ± 6.95 years and 93.8% of parents were mothers. Highly educated parents represented 46.9% of mothers and 40% of fathers. As regards parent occupation, 77.2% of the studied mothers were housewives and 25.5% of the studied fathers were farmers or workers. According to parents' report monthly income was not enough for 87.6% of them. Single parents represented 20% of the participant parents. On the other hand, the mean age of the studied children was 7.22 ± 3.86 years. Males represented 51.7% of the studied children.

As observed from **table 2**, 69% of the studied children were diagnosed with phenylketonuria during the first month of birth. Moreover, 28.3% of the

studied children had a positive family history of phenylketonuria. Concerning the disease burden, expenses of the child's special foods constitute a financial burden for 89% of the studied parents. Also, 91% of the studied parents reported that caring for their child and his special needs constitutes a physical and psychological burden on them.

In the same context, 62.8%, 49.7%, 51% and 65.5% of parents reported that their children suffer from attention deficit, hyperactivity, learning difficulties, and other behavioral problems respectively and these problems have a negative effect on 76.6% of the studied parents.

Table 3 reveals that 17.9%, 29%, and 15.9% of parents have extremely severe levels of depression, anxiety, and stress respectively.

As regards to **Table 4**, the total score of difficulties it was found that 65.5% of the studied children have behavioral and psychological difficulties. Meanwhile, 51.7% of the studied children have normal prosocial behavior. It was also found that 62.8% of the studied children have a negative impact on different areas of their everyday life related to the difficulties they have.

According to **table 5**, there was a highly statistically significant relation between the studied mothers' educational level and the total mean score of DASS ($P= 0.001$). Another statistically significant relation was found between the total mean score of DASS and the monthly income of the studied parents ($P= 0.046$).

As observed from **table 6** there were statistically significant relations

between the total mean score of DASS and the time of the child's diagnosis with PKU ($P=0.040$), the child's attention deficit ($P=0.028$), hyperactivity ($P=0.001$), and the psychological effect of the child's problem on his/her parent ($P= 0.001$).

Table (7) reveals statistically significant relations between the total mean score of difficulties and age of the participant parent ($P= 0.049$), mother's and father's education ($P=0.027$ and $P= 0.001$ respectively) as well as father's occupation ($P= 0.001$). In addition, there were statistically significant relations between total impact score and age of participant parent ($P= 0.048$) and age of the child ($P= 0.001$).

Table (8) represents highly statistically significant relations between the total score of difficulties and the presence of someone helping the parent in caring for the child and the psychological effect of the child's behavioral problems on the parent ($P= 0.001$). Moreover, there were highly statistically significant relations between the total impact score and the child's attention deficit, hyperactivity, and the psychological effect of the child's behavioral problems on the parent ($P= 0.001$). As shown in **table 9** there were highly statistically significant positive correlations between total score of DASS, total score of difficulties and total impact score ($p= 0.001$).

Table (1): Socio-Demographic Characteristics of Participant Parents and Their Children (n=145)

Socio-demographic Characteristics	No.	%
Age of participant parent (year)		
20-	45	31.0
30-	80	55.2
40-	18	12.4
>50	2	1.4
Mean \pm SD	34.38 \pm 6.95	
Participant parent		
Mother	136	93.8
Father	9	6.2
Mothers' education		
Illiterate or read and write	2	1.4
Primary education	12	8.3
Secondary education	63	43.4
Collage or post graduate	68	46.9
Mothers' working status		
Working	33	22.8
Housewife	112	77.2
Fathers' occupation		
Farmer or worker	37	25.5
Employee	35	24.1
Handicraft	20	13.8
Professional	19	13.1
Other	34	23.4
Fathers' education		
Illiterate or read and write	11	7.6
Primary education	17	11.7
Secondary education	59	40.7
Collage or post graduate	58	40.0
Both parents live together		
Yes	116	80.0
No	29	20.0
Monthly income		
Enough	18	12.4
Not enough	127	87.6
Age of child (year)		
4-	84	57.9
6-	43	29.7
12-18	18	12.4
Mean \pm SD	7.22 \pm 3.86	
Sex of the child		
Male	75	51.7
Female	70	48.3
Child's educational grade		
Nursery school	84	57.9
Primary school	45	31.0
Preparatory school	10	6.9
Secondary school	6	4.1

Table (2): Medical history of studied children and disease burden on their parents (n=145).

Medical History and Disease Burden	No.	%
Time of diagnosis with phenylketonuria		
First month of birth	100	69.0
After first month and less than one year	8	5.5
After first year	37	25.5
Positive family history of phenylketonuria		
Yes	41	28.3
No	104	71.7
Expenses of the child's special foods constitute a financial burden		
Yes	129	89.0
No	16	11.0
Type of therapeutic support		
Personal	54	37.2
Health insurance	88	60.7
Ministry of health	3	2.1
Caring for the child and his special needs constitute a physical or psychological burden		
Yes	132	91.0
No	13	9.0
Is there someone to help in caring for the child?		
Yes	43	29.7
No	102	70.3
The child suffer from any of the following*		
Attention deficit	91	62.8
Hyperactivity	72	49.7
Learning difficulties	74	51.0
Behavior (mood changes, lack of confidence, stress, exhaustion, isolation, depression)	95	65.5
If suffers, these problems have effect on the parent		
Yes	111	76.6
No	34	23.4
If yes, What are these effects? (n=111)		
Depression	57	51.4
Anxiety	31	27.9
Stress	23	20.7

*: More than one answer

Table (3): Total Score of Depression, Anxiety and Stress Scale – 21 (DASS-21) (n=145).

	Depression		Anxiety		Stress	
	No.	%	No.	%	No.	%
Normal	24	16.6	39	26.9	44	30.3
Mild	40	27.6	20	13.8	23	15.9
Moderate	36	24.8	18	12.4	24	16.6
Severe	19	13.1	26	17.9	31	21.4
Extremely severe	26	17.9	42	29.0	23	15.9

Table (4): Total Scores of Strengths and Difficulties among The Studied Children (n=145).

	Normal		Border line		Abnormal	
	No.	%	No.	%	No.	%
Emotional Problems (n=145)	29	20.0	22	15.2	94	64.8
Conduct Problems (n=145)	39	26.9	20	13.8	86	59.3
Hyperactivity (n=145)	58	40.0	10	6.9	77	53.1
Peer Problems (n=145)	39	26.9	21	14.5	85	58.6
Total Difficulties	37	25.5	13	9.0	95	65.5
Prosocial (n=145)	75	51.7	27	18.6	43	29.7
Impact (n=108)	17	11.7	0	0.0	91	62.8

Table (5): Relation between Socio-Demographic Characteristics of Parents and Their Children and Total Mean Score of Depression, Anxiety and Stress (N=145)

Socio-demographic Characteristics	Depression, Anxiety and Stress Scale – 21 (DASS-21)		
	Total DASS score (Mean ± SD)	Test of significance	p-value
Age of participant parent (year)			
20-	28.22±16.31	F=2.111	0.102
30-	24.86±14.12		
40-	33.72±15.09		
>50	18.00±0.00		
Participant parent			
Mother	34.88±18.24	t=1.649	0.101
Father	26.38±14.77		
Mothers' education			
Illiterate or read and write	63.00±0.00	F=4.558	0.001**
Primary education	22.08±10.96		
Secondary education	22.08±10.96		
Collage or postgraduate	27.06±15.08		
Mothers' working status			
Working	28.57±16.51	t=0.721	0.472
Housewife	26.41±14.67		
Fathers' occupation			
Farmer or worker	26.43±15.29	F=1.117	0.351
Employee	25.71±13.06		
Handicraft	29.75±20.31		
Professional	20.31±15.62		
Other	29.85±12.63		
Fathers' education			
Illiterate or read and write	28.81±14.60	F=0.680	0.566
Primary education	25.94±16.92		
Secondary education	28.72±17.73		
Collage or post graduate	24.98±11.32		
Both parents live together			
Yes	25.68±15.50	t=-1.969	0.051
No	31.79±12.29		
Monthly income			
Enough	20.27±9.55	t=-2.015	0.046*
Not enough	27.85±15.50		
Age of child (year)			
4-	25.71±14.55	F=0.709	0.494
6-	29.06±15.74		
12-18	27.33±16.11		
Sex of the child			
Male	24.71±12.65	t=-1.837	0.068
Female	29.27±17.08		

F : One way Anova t: student t-test, non-significant($p>0.05$),*: statistically significant ($p<0.05$), **: statistically highly significant ($p<0.001$)

Table (6): Relation between Medical History of Studied Children and Disease Burden on Their Parents and Total Mean Score of Depression, Anxiety and Stress (N=145)

Medical History and Disease Burden	Depression, Anxiety and Stress Scale – 21 (DASS-21)		
	Total DASS score (Mean ± SD)	Test of significance	p-value
Time of diagnosis with phenylketonuria (PKU)			
First month of birth	27.15±13.92	F=3.289	0.040*
After first month and less than one year	14.25±9.89		
After first year	29.00±17.81		
Positive family history of phenylketonuria			
Yes	29.31±16.90	t=1.209	0.229
No	25.96±14.27		
Do the expenses constitute a financial burden?			
Yes	27.68±15.71	t=1.763	0.080
No	20.68±5.400		
Is caring for the child constitutes burden?			
Yes	27.65±15.28	t=1.923	0.056
No	19.31±10.45		
Is there someone to help?			
Yes	24.79±15.86	t=-1.100	0.273
No	27.80±14.72		
Does the child suffer from any of the following?			
Attention deficit	29.02±14.52	t=-2.219	0.028*
Hyperactivity	31.88±15.12	t=4.167	0.001**
Learning difficulties	29.13±15.85	t=-1.829	0.070
Behavior (mood changes, lack of confidence, stress, exhaustion, isolation, depression)	27.94±14.88	t=-1.143	0.255
If suffers, Do these problems have effect on the parent?			
Yes	28.65±15.95	t=2.570	0.011*
No	21.21±10.01		
If yes, What are these effects?			
Depression	33.51±16.47	F=7.187	0.001**
Anxiety	20.83±14.07		
Stress	27.17±12.71		

©: More than one answer F: One way Anova t: student t-test, non-significant (p>0.05),*: statistically significant (p<0.05),)

** : statistically highly significant (p<0.001)

Table (7): Relation between Socio-Demographic Characteristics of Parents, Children and Total Score of Difficulties and Total Impact Score (N=145)

Socio-demographic Characteristics	Total Score of Difficulties						Total Impact Score							
	Normal (n=37)		Borderline (n=13)		Abnormal (n=95)		χ^2	p-value	Normal (n=17)		Abnormal (n=91)		χ^2	p-value
	No.	%	No.	%	No.	%			No.	%	No.	%		
Age of participant parent (year)														
20-	10	27.0	6	46.2	29	30.5	12.616	0.049*	3	17.6	25	27.5	6.083	0.048*
30-	27	73.0	4	30.8	49	51.6			14	82.4	48	52.7		
40-	0	0.0	3	23.1	15	15.8			0	0.0	18	19.8		
>50	0	0.0	0	0.0	2	2.1			0	0.0	0	0.0		
Participant parent														
Mother	34	91.9	13	100.0	89	93.7	1.092	0.579	17	100.0	85	93.4	FET	0.587
Father	3	8.1	0	0.0	6	6.3			0	0.0	6	6.6		
Mothers' education														
Illiterate or read and write	0	0.0	0	0.0	2	2.2	14.225	0.027*	0	0.0	2	2.2	2.736	0.434
Primary education	3	8.1	2	15.4	7	7.4			0	0.0	9	9.9		
Secondary education	15	40.5	11	84.6	37	38.9			6	35.3	35	38.5		
Collage or post graduate	19	51.4	0	0.0	45	51.6			11	64.7	45	49.5		
Mothers' working status														
Working	9	24.3	0	0.0	24	25.3	4.221	0.121	6	35.3	23	25.3	FET	0.387
Housewife	28	75.7	13	100.0	71	74.7			11	64.7	68	74.7		
Fathers' occupation														
Farmer or worker	9	24.3	9	69.2	19	20.0	26.907	0.001**	4	23.5	20	22.0	2.953	0.566
Employee	8	21.6	0	0.0	27	28.4			6	35.3	23	25.3		
Handicraft	4	10.8	0	0.0	16	16.8			2	11.8	13	14.3		
Professional	10	27.0	0	0.0	9	9.5			3	17.6	9	9.9		
Other	6	16.2	4	30.8	24	25.3			2	11.8	26	28.6		
Fathers' education														
Illiterate or read and write	0	0.0	4	30.8	7	7.4	22.775	0.001**	2	11.8	5	5.5	6.711	0.082
Primary education	2	5.4	3	23.1	12	12.6			0	0.0	15	16.5		
Secondary education	14	37.8	6	46.2	39	41.1			5	29.4	40	44.0		
Collage or post graduate	21	56.8	0	0.0	37	38.9			10	58.8	31	34.1		
Monthly income														
Enough	7	18.9	0	0.0	11	11.6	3.343	0.188	4	23.5	7	7.7	FET	0.070
Not enough	30	81.1	13	100.0	84	88.4			13	76.5	84	92.3		
Age of child (year)														
4-	26	70.3	6	46.2	52	54.7	8.998	0.061	15	88.2	42	46.2	10.360	0.001**
6-	9	24.3	7	53.8	27	28.4			2	11.8	37	40.7		
12-18	2	5.4	0	0.0	16	16.8			0	0.0	12	13.2		
Sex of the child														
Male	22	59.5	6	46.2	47	49.5	1.241	0.538	8	47.1	47	51.6	FET	0.795
Female	15	40.5	7	53.8	48	50.5			9	52.9	44	48.4		

χ^2 : Chi square test FET: Fisher exact test, non-significant($p > 0.05$),*: statistically significant ($p < 0.05$), **: statistically highly significant ($p < 0.001$)

Table (8): Relation between Medical History of Studied Children and Disease Burden on Their Parents and Total Score of Difficulties and Total Impact Score (N=145)

Medical History and Disease Burden	Total Score of Difficulties							Total Impact Score						
	Normal (n=37)		Borderline (n=13)		Abnormal (n=95)		χ^2	p-value	Normal (n=17)		Abnormal (n=91)		χ^2	p-value
	No.	%	No.	%	No.	%			No.	%	No.	%		
Time of diagnosis with phenylketonuria														
First month of birth	28	75.7	11	84.6	61	64.2	4.905	0.297	12	70.6	61	67.0	4.296	0.117
After first month and less than one year	3	8.1	0	0.0	5	5.3			3	17.6	5	5.5		
After first year	6	16.2	2	15.4	29	30.5			2	11.8	25	27.5		
Positive family history of phenylketonuria														
Yes	9	24.3	4	30.8	28	29.5	0.392	0.822	2	11.8	24	26.4	FET	0.353
No	28	75.7	9	69.2	67	70.5			15	88.2	67	73.6		
Do the expenses constitute a financial burden?														
Yes	30	81.1	13	100.0	86	90.5	4.191	0.123	15	88.2	82	90.1	FET	0.683
No	7	18.9	0	0.0	9	9.5			2	11.8	9	9.9		
Is caring for the child constitute burden?														
Yes	32	86.5	11	84.6	89	93.7	2.411	0.300	14	82.4	87	95.6	FET	0.076
No	5	13.5	2	15.4	6	6.3			3	17.6	4	4.4		
Is there someone to help?														
Yes	23	62.2	0	0.0	20	21.1	27.593	0.001**	9	52.9	25	27.5	FET	0.049*
No	14	37.8	13	100.0	75	78.9			8	47.1	66	72.5		
Does the child suffer from any of the following?[®]														
Attention deficit	22	59.5	7	53.8	62	65.3	0.869	0.648	6	35.3	66	72.5	FET	0.001**
Hyperactivity	18	48.6	3	23.1	51	53.7	4.305	0.116	3	17.6	54	59.3	FET	0.001**
Learning difficulties	16	43.2	4	30.8	54	56.8	4.317	0.115	6	35.3	53	58.2	FET	0.112
Behavior (mood changes, lack of confidence, stress, exhaustion, isolation, depression)	20	54.1	11	84.6	64	67.4	4.395	0.111	9	52.9	59	64.8	FET	0.416
If suffers, Do these problems have effect on the parent?														
Yes	23	62.2	7	53.8	81	85.3	12.018	0.001**	17	100.0	70	76.9	FET	0.039*
No	14	37.8	6	46.2	14	14.7			0	0.0	21	23.1		
If yes, What are these effects? (n=111)														
Depression	7	30.4	4	57.1	46	56.8	18.109	0.001**	5	29.4	40	57.1	16.988	0.001**
Anxiety	14	60.9	0	0.0	17	21.0			10	58.8	9	12.9		
Stress	2	8.7	3	42.9	18	22.2			2	11.8	21	30.0		

☺: more than one answer, χ^2 : Chi square test FET: Fisher exact test, non-significant ($p > 0.05$), *: statistically significant ($p < 0.05$), **: statistically highly significant ($p < 0.001$)

Table (9): Correlation between Total Scores of DASS, Difficulties and Impact Scale (n=145).

	Total Score of Depression, Anxiety and Stress Scale – 21 (DASS-21)		Total Score of Difficulties		Total Impact Score	
	r	p	r	p	r	p
Total Score of Depression, Anxiety and Stress Scale – 21 (DASS-21)						
Total Score of Difficulties	0.486	0.001**				
Total Impact Score	0.396	0.001**	0.669	0.001**		

** : statistically highly significant ($p < 0.001$), r: correlation coefficient

Discussion

Families of children with phenylketonuria experience parental struggles, parenting stress, and child's emotional and behavioral issues, all of which are linked to poor psychological well-being⁽²⁰⁾. The present study was conducted on 145 parents and their PKU children aged 4 to 18 years, to assess the struggles and psychological well-being of parents and their children with phenylketonuria.

Regarding characteristics of the studied parents, the mean age was 34.38 ± 6.95 years, most of them were mothers, nearly half of them were highly educated and more than three-quarters of them are housewives. Most of the studied parents were living together and did not have enough monthly income. These results are supported by another study where findings were as follows; the mean age of the studied parents was 37.7 ± 5.49 years, and more than half of them had a university degree or postgraduate degree⁽⁷⁾. Also, these results were matched with other reported research findings in which most of the studied parents were married and living in the same home⁽²¹⁾.

The mean age of the studied children was 7.22 ± 3.86 years and males represented nearly half of them. These findings are inconsistent with the results of **Morawska et al. (2020)**, who reported that the mean age of the studied children was 6.89 ± 3.6 years and that about two-thirds of them were females⁽²¹⁾.

The results of the present study revealed that more than two-thirds of the studied children were diagnosed with phenylketonuria during the first month of birth. Those children were early diagnosed through the neonatal screening program for inborn errors of

metabolism (IEM). These findings were consistent with **Öztürk et al. (2022)** who reported that most of the studied children were diagnosed through the scanning program⁽²³⁾.

Positive family history of phenylketonuria was found among slightly less than one-third of the studied children. As phenylketonuria is an autosomal recessive metabolic disorder⁽²⁴⁾ and like other IEMs, it runs in many Egyptian families causing variable levels of morbidity and mortality⁽²⁵⁾. Phenylketonuria and other metabolic disorders are common in Arab countries because of the tradition of consanguineous marriage⁽²⁶⁾.

The findings of the present study revealed that the majority of the studied parents perceived the cost of special foods (low protein diet and protein substitutes) as a financial burden. Also, most of the studied parents reported that caring for their child and his special needs constitutes a physical and psychological burden on them. These reported burdens either financial, physical, or psychological could be explained by other findings of the current study as the majority of the studied parents did not have enough monthly income. Moreover, more than one-third of parents have no therapeutic support and bear the costs of their children's treatment and investigations personally. Regarding caring for the child nearly three-quarters of the studied parents have no one to help in caring for the PKU child. In addition, the child's behavioral problems negatively affect more than three-quarters of the studied parents. Other studies in China, the United States, and Turki reported that parents of PKU children have many burdens related to the scare of their children including high financial burden.^(27, 28, 23)

Severe depression, anxiety, and stress affected less than one-quarter of the studied PKU children's parents respectively. Meanwhile, extremely severe anxiety affected more than one-quarter of the studied parents. Other studies also reported high levels of depression, anxiety, and stress in parents of PKU children⁽²⁹⁾⁽³⁰⁾⁽¹⁰⁾. Many factors may contribute to these psychological problems in the studied parents including the chronic nature of PKU⁽³¹⁾, high risk for neurocognitive impairments and disabilities, learning difficulties of PKU children, frequent medical appointments for follow-up and monitoring of Phe levels as well as limitation of parents' daily activities⁽³²⁾⁽³³⁾. The highly restrictive diet (low protein diet) could be a major factor affecting parents' psychological well-being as the diet restrictions of one family member affect the whole family's feeding style⁽³⁴⁾. Being responsible for the child's dietary control makes parents overprotective of the child and puts too much stress on them⁽³⁵⁾. Provision, and preparing a low-protein diet (restrictive diet) needs extra effort, time, and money^(36,37). In addition, parents' awareness of the danger of non-adherence to a low-protein diet and the possibility of mental retardation is another important factor in parental depression, anxiety, and stress⁽²⁹⁾. In the current study, some factors significantly affected the studied parents' levels of depression, anxiety, and stress including the mother's educational level, monthly income of the studied parents, and the time of the child's diagnosis with PKU. Previous studies showed that the low educational level of parents was associated with higher depression and anxiety scores

^(31,30,10). Education is a very important factor for women's emotional and psychological well-being, especially with low income, and lack of power and authority⁽³⁸⁾. In such circumstances, education becomes the only source of social and economic empowerment. Moreover, education makes mothers feel more confident in providing care for their sick children as parents' educational and cultural factors were found to be the most important indicators for dietary adherence and good clinical outcomes in children with PKU^(39,40,41). Parents of children diagnosed after one year of age had a higher total mean score of depression, anxiety, and stress with a statistically significant difference. The late-diagnosed and untreated infant with PKU gradually develops a profound mental disability that may necessitate institutional care⁽⁴²⁾. Parenting a child with any chronic condition that requires special care needs since birth could lead to many physical and psychological problems for parents^(31,43). Emotional problems affected nearly two-thirds of the studied PKU children, while conduct, hyperactivity, and peer problems were found among more than half of the studied PKU children according to parent reports. These difficulties had a negative impact on different areas of everyday life of nearly two-thirds of the studied children. Other studies agreed with these findings and reported negative effects of PKU on various aspects of patient's life either physical, social, emotional, or psychological⁽⁴⁴⁾⁽⁴⁵⁾. Although early treatment of PKU prevents severe cognitive disabilities some PKU children may display decreased intelligence, behavioral problems, and executive functioning deficits "planning, organization,

working memory, initiation, inhibition of usual response, and cognitive flexibility” (46). Total mean score of difficulties was significantly related to the age of the participant parent also total impact score was significantly related to both age of the participant parent and the child’s age. The results revealed that children of younger parents have higher total scores of difficulties and higher impact scores and younger children were more affected by the difficulties they face. another Egyptian study reported different findings as follows PKU children aged five years old and more had low scores in physical, cognitive, social, and psychological domains of quality of life, and in contrast younger children had better scores in all domains (47).

Findings also revealed highly statistically significant positive correlations between total score of DASS, total score of difficulties and total impact score. Where difficulties the child had negatively impact different aspects of his daily life and in turn negatively affect parents’ psychological well-being and vice versa. Caring for PKU child requires more time and effort to deal with dietary restrictions, address health problems and cope with the child’s behavioral problems (31,32,48,43).

Conclusion

The findings of the present study revealed that the studied parents struggle to provide special care needs for their PKU children. As for disease-related issues, restriction of natural protein, and costs of providing protein substitutes for their children constitute both psychological and financial burdens on them. Parents of PKU children also tussle with the behavioral and psychological difficulties facing their

children. These difficulties negatively affect various aspects of the child’s daily living as well as the psychological well-being of their parents.

Recommendations:

- Further qualitative studies are needed to provide deep insights into challenges perceived by parents of PKU children.
- Designing and implementing training programs for parents to improve their coping strategies and psychological well-being.
- Tailored intervention on managing difficult child behavior may help improve parents’ feelings of competence and psychological well-being.

Conflict of interest: None

Acknowledgment: We appreciate the time and effort of parents who participate in the study.

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