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Nursing Intervention Program for Family Caregivers Having Children with Phenylketonuria

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Abstract: *Phenylketonuria is a genetic disorder in which individuals are unable to metabolize Phenylalanine to tyrosine, which may lead to mental retardation. The study aimed to evaluate the effect of a nursing intervention program on knowledge and practices of family caregivers having children with phenylketonuria. A quasi-experimental design was used. The study was carried out at two settings, the Pediatric Genetics Clinic in Ain Shams University Children's Hospital and the Child Clinic of Phenylketonuria Metabolic Disorder in the Pediatric Hospital Campus of Cairo University. A purposive sample included 153 children with phenylketonuria and their family caregivers. For data collection, two tools were used:* ***1)*** *A structured interview questionnaire included six parts to assess demographic data for children and family caregivers' knowledge and practices regarding the Phenylketonuria.* ***2)*** *A child's medical record. The results revealed that 55.6 % of the studied children were males, while 52.9% of them their age was ranged from 2- < 6 years, and 13.7% of them are enrolled in special needs schools. Most of family caregivers were females with a mean age of 35.3±3.2. There was improvement in children's weight and phenylalanine level post program with statistically significant differences. Also, there were highly statistically significant differences in the total satisfactory knowledge for family caregivers and their practices post program implementation (X^2 = 28.564 at P <0.001). This study proved that the nursing intervention program significantly improved the family caregivers' knowledge and practices related to the care of their children with phenylketonuria. The study recommended the publication and dissemination of the nursing intervention program in all maternal and child health care centers for families having children with phenylketonuria to raise their awareness about the disease management to prevent its complications.*

Keywords: *Phenylketonuria Children - Family Caregivers - Community Health Nurse Role - Nursing Intervention Program.*

I. Introduction

Phenylketonuria (PKU) is an autosomal recessive genetic disorder in which individuals are unable to metabolize the amino acid called phenylalanine (Phe) to tyrosine that causes phenylalanine to build up in the body. The PKU is caused by a defect in the gene that helps create the enzyme needed to break down phenylalanine. Without the enzyme necessary to process phenylalanine, a dangerous buildup can develop when a person with PKU eat foods that are high in protein, because of this metabolic defect, phenylalanine rises to toxic levels in the body and can cause mental retardation, seizures, learning disabilities, and emotional problems when the infant or adult is untreated or inadequately treated. The gene defect for PKU is an autosomal recessive genetic defect and is unknowingly passed down from generation to generation.[1].[2]

People with PKU can have different levels of phenylalanine hydroxylase (PAH) deficiency, resulting in severity differences in Phe levels and symptoms. **Type I,** or classical, PKU: Classic PKU is present when plasma phenylalanine levels exceed 20 mg/D1L (1200 µmol/L) without treatment. **Type II:** There are cases of hyperphenylalaninaemia (HPA) where up to 5% of enzyme activity is retained, due to less critical mutations in the enzyme's sequence.[3]

Phenylketonuria is diagnosed through performing serum Phe test between one and seven days after birth. Blood is obtained by pricking the heel of the newborn and analyzing it for phenylalanine concentration. Children with PKU need to keep Phe levels low. So, keeping Phe levels between 120-360 µmol/L (2-6 mg/DL) for life is required. High blood Phe levels are toxic to the brain and can lead to lower intelligence, slowed reaction time, decreased ability to focus, decreased ability to remember, delayed speech and decreased thinking.[4]

Globally, PKU has an incidence of around 1 in 20,000 newborns. The highest incidence of PKU is seen among white infants of European descent (1/5,000 to 1/15,000 births). An exception is Finland, where the disorder is extremely rare (1/200,000 births). Black, Asian, and Hispanic infants are less commonly affected. The incidence in Japan has been estimated at 1 in 125,000. Data are not available from African populations. This is an autosomal-recessive disorder with males and females being equally affected.[5]

Phenylketonuria symptoms can be mild or severe and may include mental retardation. Babies with PKU seem perfectly normal at birth. The first effects are usually seen around 6 months of age. Untreated infants may be late in learning to sit, crawl and stand. They may pay less attention to things around them. Without treatment, a child with PKU will have

intellectual disabilities. Some of the effects of untreated PKU include: Intellectual disabilities, behavior problems, hyperactivity, restlessness or irritability, seizures, a skin condition called eczema , a "musty" or "mousy" body odor, and fair hair and skin, stunted growth, and small head size. EEG abnormalities, recurrent vomiting and severe learning disabilities have been major clinical problems later in life. All of these symptoms can be avoided though when proper treatment is put into place and continues throughout life**.** [6].[7]

Complications of untreated children with PKU include behavioral and psychological problems such as; hyperactivity, irritability, inability to sit, sleep disturbances, psychomotor agitation, temper tantrums, uncontrollable attacks of rage, short attention span, aggressive behavior, poor ability to follow directions, poor ability to learn new things, psychotic behavior, severe behavioral disturbance, destructiveness and self-injury. Complications include also neurological problems such as; mental retardation, seizures, tremors, muscular hypertonicity, whole body repetitive movements, paraplegia, as well and quadriplegia, developmental delay or mental impairment, congenital anomaly and mental impairment in untreated PKU and low self-esteem. Others complications include, eczema (can involve large areas of the body); sensitivity to sunlight and very light skin (due to excess Phe inhibiting melanin formation) body odor identified as "musty". [8]

The following treatments are often advised for children with PKU: Medical formulas even though they need less Phe, children with PKU still need a certain amount of protein. The medical formula gives babies and children with PKU the nutrients and protein they need while helping keep their Phe levels within a safe range. Low-Phe food plan is made up of foods that are very low in Phe. This means the child must avoid or strictly limit the following foods: milk and all dairy products including cheese, yogurt, ice cream, regular formula, meat and poultry, fish, eggs, nuts and peanut butter, dried beans, regular flour, and sugar substitute aspartame. Tracking Phe levels of babies and young children with PKU need to have regular blood tests to measure their Phe levels. If there is too much or too little Phe in the blood, the diet and formula may need to be adjusted. The BH4 (tetrahydrobiopterin) supplements is a substance made by the body. It works to help the PAH enzyme change Phe into Tyrosine. [7].[9]

Family caregiver is someone who is responsible for attending to the daily needs of another person. Family caregivers are responsible for the physical, emotional and often financial support of another person who is unable to care for him/herself due to illness, injury or disability. The care recipient may be a family member, life partner or friend. Family caregivers are sometimes described as "informal," a term used to describe those who care for family members or friends in the home, typically without pay. "Formal" caregivers, including home health care providers and other professionals, are trained and paid for their services. Some formal caregivers are trained volunteers associated with an agency.[10]

Community health nurses can be family advocates to coordinate information and resources. Nurses can help parents gain confidence in their abilities to care for their child by giving appropriate guidance and positive reinforcement. All nurses adapted to children, suffering from PKU should be aware of the dietary requirements and restrictions. Some medicines contain phenylalanine as a sweetener and should be avoided. Living with dietary restrictions can be very difficult for children. They may wish to share concerns or feelings about being "different" from peers. If there is insufficient intake of phenylalanine, which is an essential amino acid, levels may to be too low for growth and body functions, so routine screening is especially important. Successful management of PKU requires a team effort from health care providers, the child, family, and friends. [11]

1. 1. Significance of the Study:

The incidence of PKU varies widely in different human populations. A low incidence is reported in African Americans (1/50,000). Turkey has the highest documented rate in the world, with approximately 1 case in 2600 births, while countries such as Finland and Japan have extremely low rates with less than one case of PKU in 100,000 births. These disorders are equally frequent in males and females. [12]

In Egypt the incidence of PKU is unknown, but all cases about 8000 cases follow up in genetic clinics in Egypt. The prevalence in the population is approximately 4 cases per 100,000 individuals. It is diagnosed through performing screening test of serum Phe level between one and seven days after birth, through pricking the heel of the newborn and test the blood obtained for phenylalanine concentration. PKU unfortunately in Egypt this screening test is not routine consequently it is discovered later and the parents of child have to cope with the burden of daily care related to the illness**.**[13].[14]

According to [15], who concluded that in their research about 24 children diagnosed with phenylketonuria and their mean age was 3.37 years in Sohag University Hospital, Upper Egypt that, PKU still has adverse effects on children in Upper Egypt leading to developmental problems, mental retardation, and behavioral abnormalities.

1. 2. Aim of the study:

The aim of this study was to evaluate the effect of a nursing intervention program on the knowledge and practices of family caregivers having children with phenylketonuria through:

1- Assessing the family caregivers' knowledge and practices regarding the care of their children with phenylketonuria.
2- Developing and implementing a nursing intervention programs for family caregivers, according to the health needs and problems of children with phenylketonuria.
3- Evaluating the effect of the nursing intervention program on improving the family caregivers' knowledge and practices related to the care of their children with phenylketonuria.

1.3. Research hypothesis:

The nursing intervention program will improve family caregivers' knowledge and practices related to the care of their children with phenylketonuria.

II. Subjects and Methods

2.1. Research design: A quasi-experimental design was used to achieve the aim of the study.

2.2.Setting: This study was conducted at two settings, the first one was the Pediatric Genetics Clinic, Children's Hospital in Ain Shams University and the second was the Child Clinic of PKU Metabolic Disorder, affiliated to the Center for Social and Preventive Medicine (CSPM) of the Pediatric Hospital Campus of Cairo University.

2.3.Subjects: A purposive sample was used to conduct this study. The total number of children, who attended with their family caregivers at the Pediatric Genetic Clinic at Ain Shams University Children's hospital annually were 500 children, and 280 children at the Child Clinic of PKU Metabolic Disorder in the Pediatric Hospital Campus of Cairo University.

2.4.Sampling size: was calculated according to the sample size equation as

$$SS = \frac{Z^2 * (p) * (1-p)}{c^2}$$

Z = Z value (e.g. 1.96 to 95% confidence level), P = percentage picking a choice, expressed as decimal , (.5 used for sample size needed), C = confidence interval, expressed as decimal, (e.g., .04 = ±4)

The total sample was 153 PKU children and their family caregivers, 81 children selected from Ain Shams University Children's Hospital and 72 from the Pediatric Hospital Campus of Cairo University with inclusion criteria, all children diagnosed with PKU, attending at the two clinics, their aged ranged from 2- 12 years, and excluded all children suffering from any physical and mental disabilities.

2.4. Study Tools:

Two tools were used in this study for data collection:

First tool: An interview questionnaire, which included the following parts:

Part I: Demographic characteristics of child, such as age, gender, school level, and child ranking between their siblings and going to the nursery.

Part II - Socio-demographic characteristics of family caregiver regarding age, sex, relative degree, marital status, education, occupation, and family monthly income.

Part III - Medical history of children with PKU, including duration, discovery of the disease, follow up frequency per month, the degree to follow up, history of family members, and the relationship of family member suffering from the disease and complications of the disease.

Part IV- Family caregiver's knowledge, pre/post intervention program included questions regarding, meaning, causes, signs and symptoms, different methods of treatment, prevention of potential complications of the disease, and the warning signs to go to the doctor.

Part V- Family caregiver's role towards the health needs and problems for the child with PKU, including adequate nutrition, management of treatment, home safety, exercise of the children, sleep problems, the practice

at high temperature, protect the child from complications, practices to meet psychological needs, and practices to meet social needs.

Knowledge scoring system:

For each of the knowledge items, a correct response was scored "one", and an incorrect "zero". For each area of knowledge, it was considered satisfactory if the percent score was 50% or more and unsatisfactory if less than 50%.

Part VI- Family caregiver's practices related to the care of their children with PKU disease pre/post intervention program (self reported questions) through asking questions about diet management, such as "the formula preparations, dietary record, diet restriction", medication management, vaccination, suitable type of sport for children, monitoring Phe serum level twice per week, follow up for growth and development and the practices of high temperature.

Practice scoring system:-

Each of the practice items done correctly was scored "one", and not done "zero". for each area of practice was considered adequate if the percent score was 60% or more and inadequate if less than 60%.

Second tool: Children medical record to assess health status of children, which included weight, height, body mass index (BMI), and Phe serum level.

Scoring system:

Phenylalanine levels: the normal range of Phe levels between 120-360 µmol/L (2-6 mg/DL). The normal range of weight = age of the child x 2 + 8 and the normal range of length = age of the child x 5 + 80, it was calculated as follows according to [16], The formulas to calculate BMI was based on two of the most commonly used unit systems:

BMI = weight (kg)/height^2(m^2) recently according to the recommendation of [17], BMI categorization for children and teens between age 2 and 20was as follows.

Category	Percentile Range
Underweight	<5%
Healthy weight	5% - 85%
Overweight	>85%

Validity and Reliability: Content and face validity were performed by 3 professors of the community specialty of nursing faculty and two professors from the Pediatric Department ,Faculty of Medicine, all experts were affiliated to Ain Shams University, Egypt who reviewed the tools for content accuracy. The developed tool was tested for reliability on a sample of 10 subjects. The reliability test of translated version was established by using the Cronbach alpha and Pearson correlation which showed good internal consistency construct validity Cronbach alpha = (0.887).

2.5. Pilot study:

A pilot study was conducted on 10 caregivers of the total study sample 153 to test and evaluate the clarity, and applicability of the study tools and time required for completion of each study tool, also pilot study sample was excluded from the main study sample.

2.6. Administrative Design and Ethical Considerations:

Permission for conduction of the study was obtained by submission of an official letter issued from the Faculty of Nursing, Ain Shams University to the directors of Pediatric Hospital at Ain Shams University, and Cairo University. It was necessary for the researchers to get the consent of the family caregivers of children with PKU. So, strict confidentiality was ensured throughout the study process. The study subjects were assured that all data will be used only for research purpose and will be only used to improve their children's health. They were also informed about their right to withdraw from the study at any time without giving any reason.

2.7. Operational Design:

Field work:

After official permissions to carry out the study, the aim of the study was explained to the caregivers. The study was carried out along 6 months started from beginning of November 2015 to the end of Abril 2016. The average time consumed to filling the tool was 45 minutes. The previously mentioned settings were visited by the researchers three days/week (Saturdays, Mondays & Wednesdays) from 10.00 a.m. to 2.00 p.m.

III. Nursing intervention program development phases

This program was conducted on four consecutive phases, assessing, developing, implementing and evaluating.

Phase 1: A pre-program assessment test, using the interview questionnaire for data collection from the previous mentioned settings. This phase aimed at assessing family caregivers' knowledge and practices related to the care of their children with PKU according to their health needs and problems.

Phase 2: Developing an intervention program for family caregivers meeting their PKU children with PKU health needs and problems.

The general objective of the program: To improve family caregivers' knowledge, and enhance their practices related to health needs and problems of their children who have phenylketonuria

The content of the intervention program is based on needs assessment of children with PKU which include:

The Theoretical components of the nursing intervention program are:

- Identifying the meaning of phenylketonuria.
- Determining the causes of phenylketonuria.
- Describing early signs and symptoms of phenylketonuria**.**
- Explaining the importance of early detection of phenylketonuria.
- Identifying risk factors that help to develop phenylketonuria.
- Detecting the disease complications.
- Enumerating proper investigations for early detection of phenylketonuria.
- Explaining the importance of treatment.
- Describing prevention of delayed development of the child.
- Detemining the proper management for control, and prevention of complications.
- **The contents of the practical part included in the program:** It involves the diet management (the formula preparations, dietary record, and diet restriction), medication management, vaccinations, and suitable types of sport for children and monitoring Phe serum level and schedule for follow up.

Phase 3: Implementation of the program:

Implementation of the program was carried out at the previous mentioned settings. At the beginning of the first session, an orientation to the program and its purpose was presented. Each session started with a summary about what had been given through the previous sessions and the objectives of the new topics, taking into consideration the use of simple language to suite the level of family caregivers.

The theoretical part of the program was presented in two sessions in the form of lectures/ discussions followed by the practical part which consisted of two subsequent reinforcement sessions in the form of demonstration and redemonstration by using a doll, **3**days/week (Satudays, Mondays & Wednesdays), from 10.00 am to 2.00 pm). Time of each session ranged between 30 to 45 minutes. Using effective media of conveying information as, laptop, posters, and power point presentation. A booklet was developed for family caregivers as a reference to be used after program implementation. The study was carried out within six months from beginning of November 2015 to the end of Abrile 2016 based on the program implementation.

Family caregivers were divided into 7 groups, and each group consisted of 20-25 approximately. The nursing intervention program was implemented for the family caregivers at the suitable time for them according to their availability, sometimes the session was held for one or more caregivers . To insure that they were exposed to the same learning experience, they received the same intervention program content and used the same teaching strategies with direct reinforcement in the form of a copy of the intervention program booklet.

Phase 4: Evaluation phase:

The evaluation phase was done immediately post implementation of the nursing intervention program by comparing changes in family caregivers' knowledge, practices, and Phe serum level among children with PKU, in order to identify differences, similarities and areas of improvement as well as clinical defects.

Statistical Design:

Data were coded, scored, tabulated, and analyzed by computer using the "statistical package for the social science" (SPSS windows), version 19. Numerical data were expressed as mean±SD, and range. Qualitative data were expressed as frequency and percentage. Relations between different numerical variables

were tested using Pearson correlation. Chi-square (X^2) and P value less than 0.05 was considered significant and less than 0.0001 was considered as highly significant

IV. Results

Table (1): shows that, 55.6 % of the children were males and 44.4 % of them were females. Which 52.9% their age was ranged from 2- < 6 years, and 36.0% of children were not enrolled, 19.0% of them were in kindergartens, 13.7% of them in special need schools, while 25.4% in primary schools , and 5.9% of them in preparatory schools. Concerning child ranking, 34.6% of the studied samples were the third child in the family.

Table (2): shows that, 94.1% of family caregivers were female with a mean age of 35.3±3.2 years. As regards the marital status, 75.2% were married, 11.1% of them were divorced, and 13.7% were widowed. Regarding caregivers' educational level, the same table reveals that 11.8% of them were illiterate, 13.1% can read and write, 39.8% of them had intermediate education, while only 17.0% were having a university degree. Concerning the caregivers' relationship to the child, 88.2% of the caregivers were the mothers of the children, only 5.9% of them were the fathers of children, and just 2% were other relatives of the children. The table revealed also that 85% of caregivers weren't working with 81.7% of them had insufficient family income.

Table (3): indicates that the age at disease detection was less than one year for 35.3% of the studied children and for 45.7% of them it was at age from 1- < 3 years. Regarding discovery of the disease, 88.2% discovered the disease from investigation, for 79.7% of them the follow up frequency was once per month, and 18.3% of them had another family member suffering from PKU, 89.3% out of them were first degree family relationship, and 67.3% of studied children had complications of the disease.

Table (4): reflects that related to body mas index (BMI) 49.0% of the children with PKU had an over weight pre program and 45.8% of them post program. Regarding to children height, 47.7% of them were tall and 40.5% of the children with PKU had normal length, while11.8% of them had short length with no changes after implementation of the program. Regarding the serum Phe level, 70.6% of the children with PKU had Phe high level pre program and improved to be 56.9% after implementation of the program with statistically significant difference ($X^2 = 6.2345$ and P-value < 0.05).

Table (5): shows highly statistical significant differences in family caregivers' level of knowledge post-test compared to pre-test regarding the meaning of PKU, causes of this disease, its signs and symptoms, the different methods of treatment, prevention of potential complications of the disease and warning signs to visit the doctor, (P- value < 0.0001).

Figure (1): reveals that, after nursing intervention program, a highly statistically significant improvement was obvious in the family caregivers' total satisfactory knowledge, whereas post program there was a highly statistically significant improvement in the caregivers' correct knowledge scores post-test from 26.8% unsatisfactory, to 95.4% satisfactory ($X^2 = 28.564$ at P <0.001).

Figure (2): illustrates that highly statistically significant improvement was detected in total family care givers' practices related to care of their children with PKU post-program than pre-program implementation ($X^2 = 11.04$ at p = 0.0009).

Table (6): represents distribution of family caregivers' adequate practices related to care of their children suffering from PKU pre/post-tests. The table shows that there were highly statistically significant improvements post-program compared to pre-program implementation (p < 0.001). These practices wete related to management of treatment, sleep problems, practice at high temperature, and protecting the child from complications (88.9%, 90.2%, 95.4%, & 97.4 % respectively post-test compared to 69.3%, 67.9%, 77.1%, &73.2% respectively pre-test) . As well statistically significant improvements were detected in items adequate nutrition, home safety, and exercise of the children 96.1%, 83.0%, & 84.3% respectively post-test compared to 88.9%, 70.6%, &71.3% respectively pre-program (p <0.05).

Figure (3): Represents that there were highly statistically significant relations between the caregivers' total knowledge and their total practices toward the care of their children suffering from PKU pre/post program ($X^2 = 82.884$ at $p < 0.001$).

Table (7): shows that there weren't statistically significant relations between family caregivers' total knowledge and their children's BMI and height pre/ post program, (P > 0.05), while there was a highly statistically significant relation between family caregivers' total knowledge toward the care of their children with PKU and their children's serum Phe level which was significantly improved post program (P < 0.001).

Table (8): reveals that there weren't statistically significant relations between family caregivers' total adequate practices toward the care of their children with PKU and their children's BMI and height pre/ post program (P > 0.05), while there was a highly statistically significant relation between family caregivers' total practices and their children's serum phe level which was significantly improved post program (P< 0.01) .

Table (1):- Distribution of the studied children with PKU according to their demographic charactristics (n=153).

Items	N	%
Gender		
Male	85	55.6
Female	68	44.4
Age		
2 -	81	52.9
6 -	33	21.6
8 -	21	13.7
10 -12	18	11.8
Educational level		
Did not enroll	55	36.0
Kindergartens	29	19.0
Special needs schools	21	13.7
Primary school	39	25.4
Preparatory school	9	5.9
Child ranking		
First	31	20.3
Second	45	29.4
Third	53	34.6
Fourth	18	11.8
Fifth	6	3.9

Table (2): Distribution of the family caregivers according to their socio-demographic charactristics (n=153).

Items	N	%
Gender		
Male	9	5.9
Female	146	94.1
Age		
20 -	32	20.9
30 -	107	69.9
> 40 years	14	9.2
Mean ±SD	35.3±3.2	
Marital status:		
Married	115	75.2
Divorced	17	11.1
Widow	21	13.7
Educational level		
Illiterate	18	11.8
Read and write	20	13.1
Basic education	28	18.3
Intermediate education	61	39.8
University degree	26	17.0
Relationship to the child:		
Mother	135	88.2
Father	9	5.9
Grandma	6	3.9
Other relatives	3	2.0
Care giver's job		
Working	23	15.0
Not working	130	85.0
Family Income		
Sufficient	28	18.3
Insuficient	125	81.7

Table (3):- Distribution of the studied children with PKU according to their medical history (n=153)

Items	N	%
Duration of the disease(in years)		
< 1 year	54	35.3
1 - < 3	70	45.7
3 - < 6	22	14.4
6 - < 9	6	3.9
9 - < 12	1	0.7
***Discovery of the disease by**		
Chance	11	7.1
Symptoms of the disease	66	43.1
Complications of the disease	37	24.1
Laboratory investigation	135	88.2
Through follow up of MCH services	12	7.8
Follow up frequency per month		
Once	122	79.7

Twice	31	20.3
History of family member	28	18.3
Relationship of family member suffering from the disease (n=28)		
First degree	25	89.3
Second degree	3	10.7
Third degree	0	0.0
Complications of the disease	103	67.3

** Responses are not mutually exclusive*

Table (4):- Distribution of the studied children with PKU according to their health status (n=153)

Items	Pre program		Post program		Chi-square
	No	%	No	%	
Body mass index(BMI)					$X^2 = 1.257$
Overweight	75	49.0	70	45.8	P=
Normal weight	44	28.8	53	34.6	0.53339129
Under weight	34	22.2	30	19.6	(NS)
Height					
Tall	73	47.7	73	47.7	$X^2 = 0$
Normal	62	40.5	62	40.5	P=1
Short	18	11.8	18	11.8	(NS)
Serum Phe level					
High	108	70.6%	87	56.9	$X^2 = 6.23$
Normal	45	29.4%	66	43.1	P= 0.0125
Low	0	0.0%	0	0.0	(S)

Table (5): Family caregivers' satisfactory knowledge about PKU disease (n=153).

Satisfactory Knowledge about PKU	Pre- prog. Satisfactory		Post- prog. Satisfactory		Chi-square	
	No	%	No	%	X^2	P-value
Meaning of PKU	118	77.1	149	97.4	28.24	< 0.0001***
Causes	116	75.8	150	98.0	33.24	< 0.0001***
Signs and symptoms	121	79.1	148	96.7	22.41	< 0.0001***
Different methods of treatment	111	72.5	138	90.2	15.71	< 0.0001***
Prevention of potential complications	115	75.2	147	96.1	27.1811	< 0.0001***
Warning signs to visit the doctor	118	77.2	145	94.8	19.7254	< 0.0001***
Total satisfactory knowledge	112	73.2	146	95.4	28.564	< 0.0001***

**Not Significant (NS) **Significant (S) ***Highly significant (HS)*

Figure (1): Family caregivers' total knowledge related to the care for their children with PKU throughout program phases (n=153).

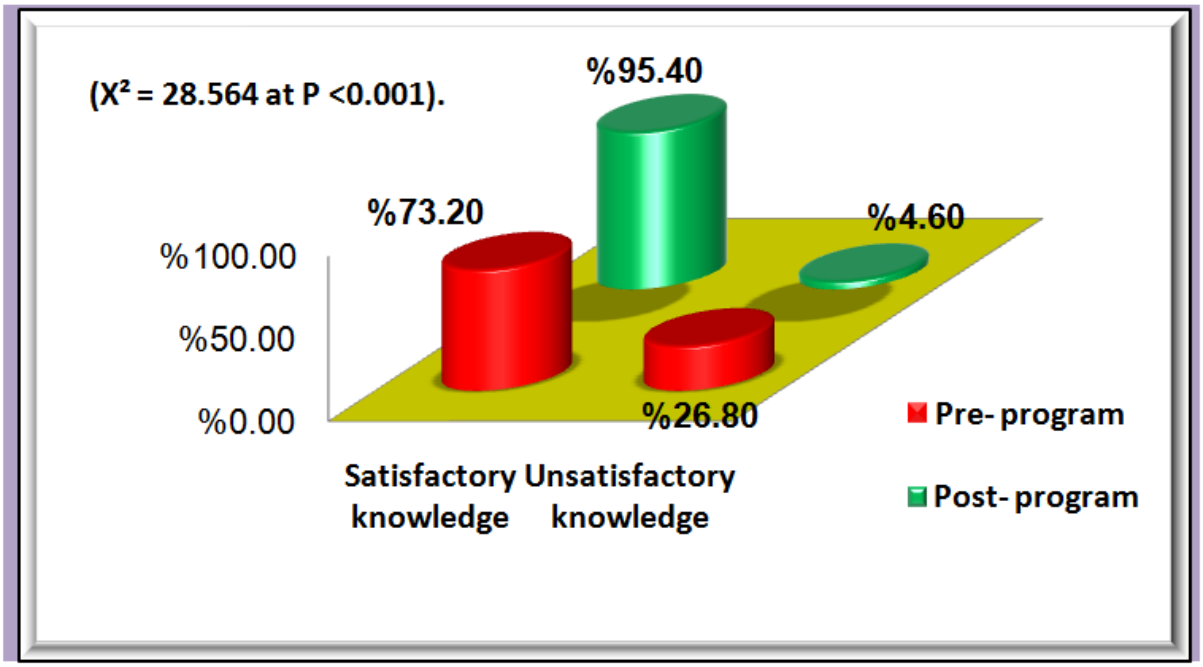

Figure (2): Total family caregivers' practices related to the care for their children with PKU (n=153).

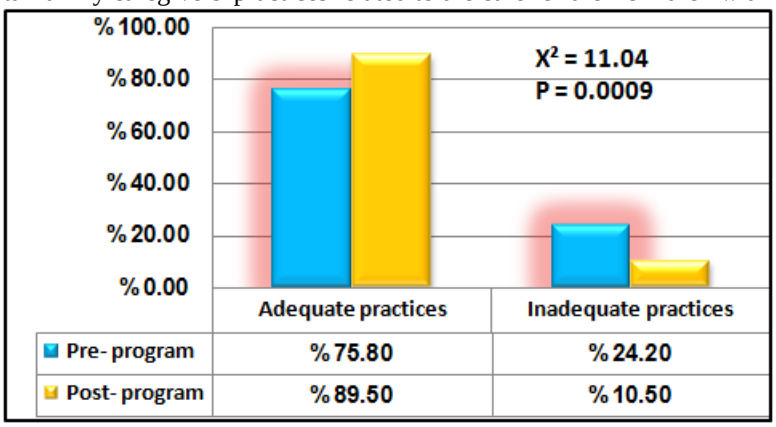

Table (6): Family caregivers' adequate practices pre/post program related to the care of their children with PKU (n=153).

Adequate Reported Practices	Pre- prog. Adequate		Post- prog. Adequate		Chi-square	
	No	%	No	%	X^2	P-value
Adequate nutrition	136	88.9	147	96.1	5.68	0.0171**
Management of treatment	106	69.3	136	88.9	21.54	< 0.0001***
Home safety	108	70.6	127	83.0	6.62	0.0101**
Sleep problems	104	67.9	138	90.2	22.83	< 0.0001***
Exercise of the children	109	71.3	129	84.3	7.56	0.006**
Practice at high temperature	118	77.1	146	95.4	21.63	< 0.0001***
Protect the child from complications	112	73.2	149	97.4	35.66	< 0.0001***
Practices to meet psychological needs	98	64.1	123	80.4	10.18	0.0014**
Practices to meet social needs	119	77.8	129	84.3	2.12	0.1447*
Total practice	116	75.8	137	89.5	11.04	0.0009**

**Not Significant(NS) **Significant (S) ***Highly significant (HS)*

Fig (3): Relations between family caregivers' total knowledge and their total practices toward the care of their children with PKU pre/post program (n= 153).

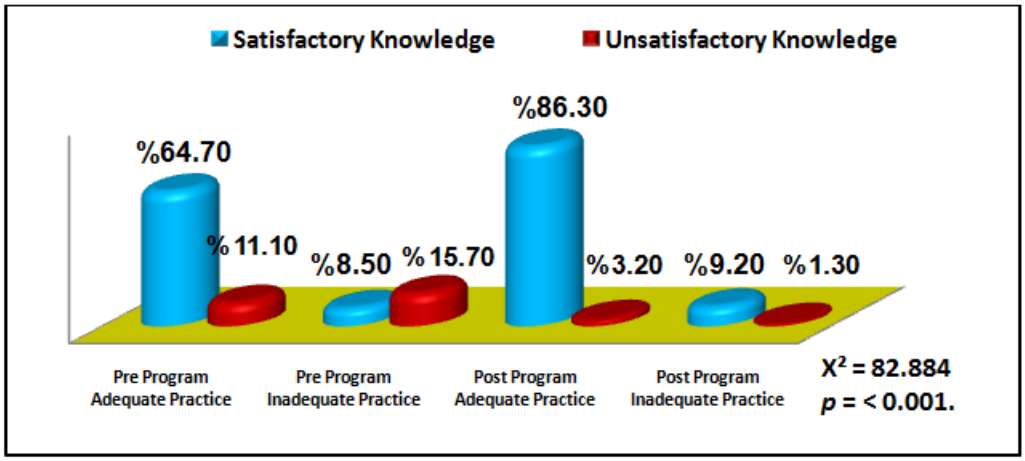

Table (7): Relations between family caregivers' total knowledge and their children's health status pre/ post program (n= 153).

Children's Health Status	Pre program total satisfactory knowledge (n=112)		Post program total satisfactory knowledge (n=146)		Chi-square	df	P Value
	No	%	No	%			
Body mass index(BMI)							
Over weight	51	45.5	67	45.9	0.147	2	0.9306* (NS)
Normal weight	42	37.5	52	35.6			
Under weight	19	17.0	27	18.5			

Height							
Tall	49	43.8	73	50.0	1.019	2	0.5897 (NS)
Normal	52	46.4	61	41.8			
Short	11	9.8	12	8.2			
Serum Phe level							
High	70	62.5	81	55.5	30.018	3	0.00000137 (HS)
Normal	42	37.5	65	44.5			

Table (8): Relations between family caregivers' total adequate practices and their children's health status pre/post program (n= 153).

Children's Health Status	Pre program total adequate practices (n=116)		Post program total adequate practices (137)		Chi-square	df	P. Value
	No	%	No	%			
Body mass index(BMI)							
Overweight	61	52.6%	59	43.1%	2.546	2	0.27999039 (NS)
Normal weight	39	33.6%	52	38.0%			
Under weight	16	13.8%	26	18.9%			
Height							
Tall	47	40.5%	62	45.3%	0.715	2	0.6994227 (NS)
Normal	56	48.3%	59	43.1%			
Short	13	11.2%	16	11.6%			
Phenylalanine levels							
High	93	80.2%	72	52.6%	21.121	1	0.00000431 (HS)
Normal	23	19.8%	65	47.4%			

V. Discussion

The current study aimed to evaluate the effect of a nursing intervention program on knowledge and practice of the family caregivers having children with PKU. The PKU is an inherited disorder in which the body lacks the enzyme needed to convert Phe to tyrosine. Excess Phe accumulates in the blood and tissues can damage the brain and nervous system if left untreated. Family caregivers must cognitively and behaviorally manage the stress of the disease comprehending condition, they were adjusting to the diagnosis and providing appropriate care to meet the health needs of the children suffering from PKU**. Charles etal.,** [18] .

The current study results represent that, the 153 PKU children suffering from PKU were participating in this study, more than half of them were male, while less than half of them were female, and more than half of them their age ranged from 2- < 6 years. As for education more than one third of children weren't enrolled for education, while less than one fifth of them were kindergartens, and more than one tenth in special needs schools, almost one quarter in primary schools and a minority of them were in the preparatory schools. Concerning child ranking, slightly more than one third of children under the study were the third child in the family **(Table 1)**. This finding is in disagreement with many previous studies such as that of **Christel,etal.,** [19], who conducted a study about quality of life among parents of children with PKU, and found females slightly outnumbered males. As well **Marcos& Michelle,** [20], found in their study entitled "Metabolic syndrome in children and adolescents with PKU", that more than half of children suffering from PKU were females, while the rest of them were male.

Regarding to the child's educational level the current study findings are in congruent with **Akram** [21], who in a study on children suffering from PKU aged 6-18 years in the Gaza Strip found that, 85.9% of study participants were in prep/primary schools, while 14.1% were in secondary schools, and 15.2% were excellent in school performance, 30.4% were very good, 31.5% were good and 22.8% were weak. However, **Kurtis,** [22] found that, the children with PKU have learning difficulties, hyperactivity, psychological problems, all of which are reported to be more prevalent in PKU.

In the present study, most of the family caregivers were females; with a mean age of 35.3±3.2 years. As regards marital status, slightly more than three quarter of them were married, while more than one tenth of them was divorced, and more than one tenth were widowed. Concerning the caregivers relationship to the child, the majority of them were the mothers of the children, and only minorities of them were the fathergrandma or other relatives of the children. **(Table 2)**. These results are supported by several studies that of **Gallo etal.,**[23]**. Abd-Elkodoos etal.,** [24]**,** who stated that most family caregivers were female and the mothers of children are considered the most primary health care providers around the world, they take on enormous responsibilities in providing care and managing their children's conditions outside the health care institutions. In the Egyptian culture, mothers are the main caregivers of their families with or without sick persons in their families because men are always busy in their work.

Regarding caregivers' educational level, more than one tenth of them were illiterate, and more than one tenth of them can read and write, almost two fifth of them had intermediate education, while only less than one

fifth of them were having a university education. The results revealed that the majority of caregivers weren't working, which leading to that the majority of them had insufficient family income.

This finding was supported by **Ahmed** [25], whose study entitled "Home care offered by family caregivers to preschool children, suffering from hemiplegic palsy", found that, almost half of family caregivers were illiterate and the minority of them had higher education. On the other hand, **Gallo etal.,** [23] reported that most of the family caregivers completed high school, and approximately one third completed colleges or graduate schools. As well, **Lord etal.,** [26], whose study entitled "Implication of resolving the diagnosis of PKU for parents and children", reveald that half of the family caregivers, had completed tertiary education. This difference denotes that most of the females in Egypt are less educated than males as reported by the Arab Republic of Egypt, Literacy and Adult Educations **Arab Republic of Egypt** [27].

The current study results revealed that, the age at disease detection was less than one year for more than one third of the studied children, while for less than half of them it was discovered at age from 1- < 3 years, and the majority of their family caregivers reported that they discovered the disease from investigation **(Table 3)**. This finding was congruent with that of a study conducted by **Vegni etal.,** [28], who studied 40 participants between the ages of 8 to 31 years. They found that all patients had been diagnosed with PKU within the first 3 years of life, were cognitively able to participate and had no other pathologies. The researchers attribute this study findings to the constricted and obligatory follow up program for children under five years in MCH centers, affiliated to the Ministry of Health and population, in Egypt.

The results of this study showed that more than two third of family caregivers reported that their children with PKU suffered of many complications associated with the inherited disorder **(Table 3).** Such finding was in agreement with **Hamawandi etal.,** [29], who stated that, PKU is presumed to be more prevalent among the neonates of consanguineous marriages. The high incidence of relative marriages is directly associated with a high prevalence of the disease in the population.

In relation to weight and height of the PKU children, the result of the present study indicated that pre program slightly less than half of the children with PKU were overweight **(Table 4)**. This finding is in agreement with Kanufre **etal.,** [30], who found that patients with PKU were excess weight, which potentially vulnerable to the development of metabolic syndrome. They added that excessive weight gain among the patients with PKU can be due to, that those patients are vulnerable groups of metabolic abnormalities and excess weight. Protein restriction favors and even stimulates the consumption of carbohydrate-rich foods (especially simple carbohydrate) and lipids, in particular, increasing the risk of weight gain.

The finding of the present study clarified that about one tenth of the children with PKU had short length **(Table 4).** This is in congruence with **Brumm etal.,** [31]**,** who mentioned in their research, that signs and symptoms among the children with PKU are decrease growth and development so, this study result reflected that a relatively high percentage of children with PKU are short in height.

Concerning the family caregivers' level of knowledge, there were highly statistically significant differences in their knowledge post-test compared to pre-test regarding the meaning of PKU, the causes of this disease, its signs and symptoms, the different methods of treatment, prevention of potential complications of the disease , and warning signs to visit the doctor **(Table 6)**. These differences showed significant improvement about total family caregivers' satisfactory knowledge after nursing program implementation ($X^2 = 28.564$ at P <0.001) **(Figure 1).** This finding is in agreement with **Ozel etal.,** [32]**,** who found that, the total knowledge scores were lower in most family caregivers who were living in cities. Lower knowledge scores among rural family caregivers can be attributed to that they did not have knowledge regarding the diet and this knowledge deficit may be due to their lower educational level adding to the improper health education about their children's conditions from the health care team and the limited role of health professionals including the nurses in providing the caregivers with the appropriate care for childrenwith PKU in rural health facilities.

The current study represents that, there were highly statistically significant relations between the caregivers' total knowledge and their total practices toward the care of their children suffering from PKU pre/post program ($X^2 = 82.884$ at $p < 0.001$) (**Figure 3***).* This finding goes in the same line with **Ali,** [33]**,** who mentioned that, when caregivers are provided with the basic knowledge about their child's condition, developmental prognosis and various treatment approaches, this will assist them in practicing healthy behaviors and also can change their unhealthy behaviors. As well, this opinion is congruent with **Macdonald etal .,** [9], who believed that dietary knowledge is an essential factor on dietary compliance. This finding is also consistent with the study results of **Ali,** [33]**,** who reported that, the caregiver's practice scores and dietary compliance was much lower when knowledge scores were particularly low. These results mean that, the family caregivers' knowledge had an effect on their practices, as when family caregivers have a satisfactory knowledge level regarding their children's conditions; this will improve their practices regarding the care of their children with PKU.

Highly statistically significant relations were found in this study results between family caregivers' total knowledge/total practices toward the care of their children were suffering from PKU and their children's

serum Phe level which was significantly improved post program **(Tables 7 &8).** This finding was in agreement with **Peretti& Jessica** [34], who stated that children with PKU that perceive more barriers to treating their disease, such as; lack of knowledge, difficulty planning meals, social pressures, and disliking the formula, also have elevated Phe concentrations compared to children who perceive less barriers. As well, This result is supported by that of a study using a single education intervention model, a study done in a week long camp experience by **Demydas,** [35] and showed significant improvements in plasma Phe concentrations ($p = 0.0001$) post camp but these improvements in plasma Phe concentrations were not observed in long term.

The nursing intervention program had a statistically significantly positive effect as it improved the family caregivers' knowledge and practices related to the care of their children with PKU which led to significant decrease in the Phe concentration levels in the blood of their children that consequently will lead to progress in the general health condition growth, and development for those children.

VI. Conclusion

In conclusion, the finding of this study revealed that there were highly statistically significant differences in the family caregivers' total satisfactory knowledge and adequate practices toward the care of their children with PKU after the nursing program implementation. As well, there was a highly statistically significant relation between family caregivers' total knowledge and practices and their children's serum Phe level which was significantly improved post program implementation.

VII. Recommendations

Based on the results of the present study, it can be recommended that:

- Publication and dissemination of the nursing intervention program in all maternal and child health care centers for families having children with PKU to raise their awareness about the disease management to prevent its complications.
- Encouraging the importance of regular follow-up and regular investigations of children with PKU to ensure proper serum Phe level and early detection of complications.
- Further research study to be done to investigate the other social and psychological health needs of family caregivers that are evidence–based in order to prevent health problems of children with PKU.
- Further longitudinal study to be done to evaluate the effect of the researchers' intervention program on the general health condition of the children suffering from PKU and their growth and development.

References

[1]. Anderson, PJ, & Leuzzi, V. (2014): White matter pathologyin phenylketonuria.(3rd ed). Mol Genet Metab; 99(Suppl 1):S3-9.
[2]. Marcason, W. (2014): Is there a standard meal plan for phenylketonuria (PKU)? Journal of the Academy of Nutrition and Dietetics; 113:S1.
[3]. Ghosh-Jerath, S., Thompson S., & Rocca, A. (2013): Phenylalanine hydroxylase deficiency, type I hyperphenylalaninaemia; Patient Plus Articles (v24) 28/03/2013.
[4]. Acosta, P.B.,& Matalon, K.M. (2010): Nutrition management of patients with inherited disorders of aromatic amino acid metabolism; In: Acosta PB, editor. Boston: Jones and Bartlett Publishers; pp. 119-174.
[5]. Steiner, R.D. ; (2011): Phenylketonuria, Medscape, Nov 2011.
[6]. Camp, K.M. (2014): Phenylketonuria Scientific Review Conference: State of the science and future research needs. Genetics and Metabolism; 112:87.
[7]. Screening, Technology, & Research (2016): Screening and genetic testing in Genetics is a multi-state project to improve information about the financial, ethical, legal, and social issues surrounding expanded newborn. Available at: http:// www.newbornscreening.info Disorder name: Phenylketonuria Acronym: PKU 1 Update Date: 2/16/2016.
[8]. Somaraju U.R., & Merrin, M. (2015): Sapropterin dihydrochloride for phenylketonuria. Cochrane Mar 27;(3):CD008005. Doi: 10.1002/14651858.CD008005.pub4.
[9]. Macdonald, A., Davies, P., Daly, A., Hopkins, V., Hall, S. K., Asplin, D., Hendriksz, C. & Chakrapani, A. (2012): Does maternal knowledge and parent education affect blood phenylalanine control in Phenylketonuria? Journal of Human Nutrition and Dietetics. 21: 351-358.
[10]. National Alliance for Caregiving. (2010): Care for the Family Caregiver: A place to start, fifth anniversary, The hite House Conference on Aging, pp.12:24.
[11]. Patney S. (2013). Textbook of community health nursing, (3rd ed). Dynamic rint Grafix: New Delhi pp. 275-300.
[12]. Georgianne, W. (2014): Phenylketonuria and epidemiology from wikipedia, the free encyclopedia Updated: Apr 28, 2014.
[13]. Rodenburg, R., Meijer, A.m. Dekovic, M. & Aldenkamp, A.P. (2010): Parents of children with enduring: predictors of parenting stress and parenting. Accepted on May 3, 2007, from www. Elsevier. Com / locate / ybeh.
[14]. http://www.rightdiagnosis.com/p/phenylketonuria/stats-country.htm [Accessed on 13th August, 2015 (1:50pm)].
[15]. Abdelrahim, A.S., Ahmed, M. E., &Mostafa, Y.A. (2013): Clinicolaboratory profile of phenylketonuria (PKU) in Sohag University Hospital-Upper Egypt, Egyptian Journal of Medical Human Genetics, July; 14(3): p. 293–298.
[16]. Theodore, H., Tulchinsky, A., and Varavikova E., (2014): textbook new public health, 3rd ed, Oxford, New York, Tokyo, Oxford Uni.Press. pp. 436-442.
[17]. Centers for Disease control and Prevention (CDC) (2016): BMI Categorization for children and Teens between 2 and 20. Accessed at http://www.calculator.net/pdf/chart-2016.pdf.
[18]. Charles, S.C., & Connor-Smith, (2010): "Personality and coping". Annual Review of Psychology: 61: 679–704.

[19]. Christel, S., Astrid, F., & Lutz, G. (2013): Quality of life among parents of children with phenylketonuria (PKU) Fidika ; licensee Bio Med Central Ltd. DOI: 10.1186/1477-7525-11-54.
[20]. Marcos, V. C., & Michelle, R. A. (2015): Metabolic syndrome in children and adolescents with phenylketonuria. J Pediatr (Rio) J. ; 91(1):98-103 Brazil
[21]. Akram, M.A. (2013): Impact of dietary adherence on blood L-phenylalanine levels among Phenylketonuria children aged 6-18 years in the Gaza Strip; Al-Azhar University, Gaza Faculty of Pharmacy.
[22]. Kurtis, S.M.D. (2015): PKU: A parent's case report: University of Virginia, at web site http://pkunews.org/personal/parent.htm[Accessed July 21, 2015].
[23]. Gallo, A.M., Knafl, K.A., & Angst, D.B. (2009): Family information management patterns in childhood genetic conditions. Journal of Pediatric Nursing; june, 24: 3.
[24]. Abd-Elkodoos, R. F., Gharaei J.M., Mostafavi, S., & Alirezaei, N. (2011): Quality of life and the associated psychological factors in caregivers of Children with PKU. Iran J. Psychiatry, Spring; 6 (2): 66-69. PMCID: PMC3395945.
[25]. Ahmed, S. (2009): Home care offered by family caregivers to preschool children, suffering from hemiplegic palsy. Minia University p. 81.
[26]. Lord, B., Ungerer, J., & Wastell, C. (2008): Implications of resolving the diagnosis of PKU for parents and children, Journal of Pediatric Psychology, 33, (8): 855-866, Published by Oxford University Press on behalf of the Society of Pediatric Psychology. Journals. Permissions@oxfordjournals.org. [Accessed on 20 December, 2014].
[27]. Arab Republic of Egypt (2008): Literacy and Adult Education. Online at: http://www.unesco.org/ fileadmin/ multime-dia/institutes/uil/ confintea/ pdf/ National_ Reports/ Arab%20States/Egypt.pdf, 2008 [Accessed on 3 march,2015].
[28]. Vegni, E., Fiori, L., Riva, E., Giovannini, M., & Moja, E.A. (2010): How individuals with phenylketonuria experience their illness: An age-related qualitative study. Child: Care, Health & Development, Jul; 36(4):539-48.
[29]. Hamawandi, M.H., Rashid, J.A., Raof, S. H., & Hawrami, O.M. (2015): Annual incidence of phenylketonuria in Sulaimani City, Merit Research Journal of Medicine and Medical Sciences (ISSN: 2354-323X) 3(9): 427-431, Available online at: http://www.meritresearchjournals.org/mms/index.htm [Aaccessed on 15 March 2015].
[30]. Kanufre. C.V., Marcos J.B., & Michelle, R. A. (2014): Metabolic syndrome in children and adolescents with phenylketonuria. J Pediatr (Rio J). 2015; 91(1):98-103 Brazil.
[31]. Brumm, V.L., Bilder, D.,& Waisbren, S.E. (2010): Psychiatric symptoms and disorders in phenylketonuria Mol. Genet. Metab., 99 : pp. S59–S63 at wib site http://dx.doi.org/10.1016/j.ymgme.2009.10.182 [Accessed on 3 Februery 2012]
[32]. Ozel, H.G., Kucukkasap, T., Koksal, G., Kal-Kanoglu Sivri, H.S., Dursun, A., Tokatli, A. & Coskun, T. (2008): Does maternal knowledge impact blood phenylalanine concentration in Turkish children with Phenylketonuria? References to electronic databases: PKU: OMIM 261600. HPA: OMIM 261600. SSIEM and Springer.
[33]. Ali, N. (2008): Needs of the physically handicapped adults in the vocational rehabilitation centers. Master Thesis, Faculty of Nursing, Alexandria University, p. 25.
[34]. Peretti, A. & Jessica, R. (2012): Group education for children with phenylketonuria. Thesis presented to the graduate programs in Human Nutrition and the Oregon Health & Science University School of Medicine, in partial fulfillment of the requirements for the degree of Master of Science, pp.10-68.
[35]. Demydas, T., (2011): Consumer segmentation based on the level and structure of fruit and vegetable intake: An empirical evidence for US adults from the national health and nutrition examination survey (NHANES) 2005-2006. Public Health Nutr. Jun; 14(6):1088-95.