RESEARCH PAPER | Medical Science | Volume : 6 | Issue : 6 | June 2016 | ISSN - 2249-555X | IF : 3.919 | IC Value : 74.50

Nutritional Concerns Among Patients with Inborn Metabolic Disorders in The Kingdom of Saudi Arabia

KEYWORDS

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ABSTRACT *Background: Metabolic disorders are inborn errors of metabolism caused by mutant genes that result in the abnormal production of enzymes, which if not diagnosed and treated immediately can lead to mental and developmental abnormalities. Many symptoms of the metabolic disorders can be controlled by dietary intervention.Objective: This study aims to determine difficulties faced by metabolic disorders patients in regards to the nutritional resources available within Riyadh. Methods: The study design was cross-sectional, and the subject size was dependent on the number of patients visited the clinic, June to September of 2014. The participants included those diagnosed with inborn metabolic disorders at two different hospitals in the Riyadh. A questionnaire was used to evaluate the Participants awareness and problems faced regarding proper nutritional resources. Results: Participants were aware of the nutritional management of their diseases, but nutritional resources that they needed to help maintain a normal state is difficult to obtain. Conclusion: The lack of dietary choices and difficulty in getting proper diet could adversely affect the disease progress and can increase the risk of morbidity and mortality.*

Introduction

Inborn errors of metabolism (IEM) are inherited biochemical disorders in which specific enzyme defects affect the normal metabolism of protein, fat, or carbohydrate due to which certain compounds reach toxic levels, and the levels of others may become deficient. These enzyme defects lead to a host of medical and developmental consequences ranging from intellectual disability to severe cognitive impairment if left untreated. Through early diagnosis and initiation of the treatment, many of these adverse outcomes can be delayed or even prevented (Acosta & Yannicelli, 2001; Fernandes, Saudubray, Van den Berghe, & Walter, 2006). For many IEM, treatment strategies rely on the provision of specialized medical foods and dietary supplements (Acosta & Yannicelli, 2001). Inborn metabolic disorders are of many types, e.g., Protein disorders, carbohydrate disorders, urea cycle disorders and many others.

Middle Eastern cultures are tribal, and marriage between cousins has been part of the culture for ages leading to a founder effect and a significant number of autosomal recessive diseases (Aida Al Aqeel, 2004). In Saudi Arabia, just like in other Middle East countries, first, cousin marriages are very common, leading to uniquely common disorders. Many of these genetic metabolic disorders can be recognized in a clinical setting, and the treatment is either difficult or expensive or unavailable in most centers (Aida Al Aqeel, 2004). Some of the diseases can be treated or the symptoms controlled with dietary intervention; both the method of administration and the composition of feeds must be quickly considered. Oral nutrition is desirable if the condition and clinical status permits. Continuous enteral tube feeding can be temporarily useful in many Participants whose initial condition is poor, total parenteral nutrition (TPN) is the method of choice in those cases where sufficient enteral nutrition is precluded. At least, the age-related recommended daily energy should be provided (Fernandes et al., 2006)

There is a lack of research in KSA about nutritional management among metabolic disorder patients. This study aims to find out problems faced by the patients regarding their treatment using diet therapy, and the level of awareness among the Saudi population in regards to inborn metabolic disorders and newborn health screening.

METHODOLOGY

Subjects:

The study design is cross-sectional. The participants (age one to twenty-five years) were recruited between the months of June and September, in the year 2014. The data was gathered from different hospitals (King Faisal Specialist Hospital and King Fahd Medical City) within the city of Riyadh. A consent form was collected from all participants before the start of the study. Only Participants having any inborn metabolic disorder were selected for the survey.

Questionnaire:

The study tool included an interviewer administrated questionnaire. The questionnaires were distributed to all the study participants. The purpose of the survey was to gather clinical and demographic data of the participants. Moreover, to determine the available source of diet & awareness among inborn metabolic disorder Participants such as what they know about the types of food that should be avoided in diet, and what they know about types of food allowed to them, and the alternative food available for their disease. The caretakers of the Participants who were 13 and below were asked to fill the consent form and the questionnaire.

Statistical analysis:

A descriptive statistical analysis of the relevant variables was carried out to study the nutritional problems faced by the patient. Data was analyzed using Microsoft Excel 2010.

RESULTS

Table 1 shows the social demographics of the subjects. A total of 48 patients took part in the study. Their ages ranged from 0 to 25 yrs. There were 35.14% children from the age group 1 to 6 yrs, 27.03% from age group 7 to 13yrs, 35.14% from the age of 14 to 24 yrs and there was only 2.70% from the age 25 and above. The subjects were coming from all

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parts of the kingdom, with 32.43% being the central part of the country while only 5.41% were from the south of KSA. There were 62.16% Saudi nationals among the subjects while 37.84% were of other nationalities. 54.05% males took part in the study and 45.95% females.

Table 1. Socio-demographics of the subjects

Characteristics	Classification	Frequency in %
Age	0-6 7-13 14-24 >25	35.14 % 27.03 % 35.14 % 2.70 %
Nationality	Saudi Non-Saudi	62.16% 37.84%
Gender	Female Male	45.95 % 54.05 %

When asked about whether they had any information regarding metabolic disorders before diagnosis, most (70.27%) answered in the negative while only 29.73% knew about these disorders. On the other hand, when asked about newborn health screening, 32.43% had not heard about it while 67.57% agreed to have heard about the screening.

Figure one shows the distribution of different metabolic disorders among the participants. Phenylketonuria was seen to be most common among the diseases with 53% of the participants having PKU while 18% had organic academia and 13% had Tyrosinemia. Only 9% and 7% had Galactosemia and Glycogen storage disorder respectively. The disorders were not further specified according to the enzyme defect but was defined under a general group disorder.

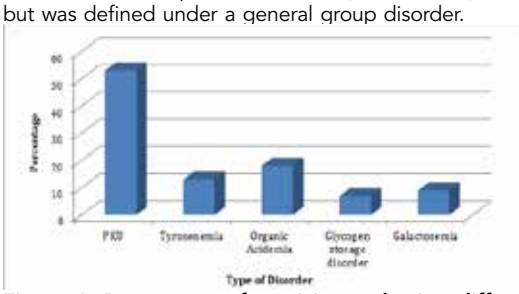

Figure 1. Percentage of participants having different inborn metabolic disorders

Figure 2 demonstrates the percentage of when the diagnosis of the metabolic disorder was made for the Participants. Approximately 35% were diagnosed at birth while 40.54% were diagnosed after a year of birth. Around 13.5% were diagnosed two weeks and 10.81% were diagnosed after four months up to 1 year. Those diagnosed at birth, screening was done because of the knowledge of the parents about genetic disorders in the family.

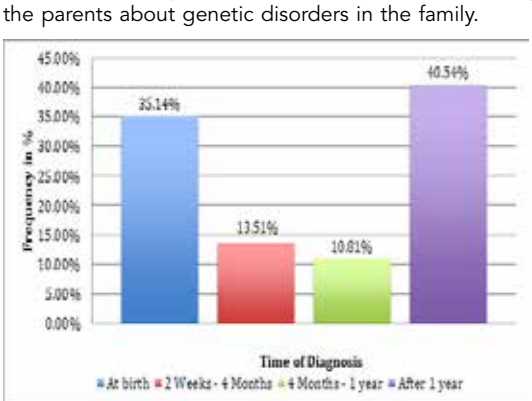

Figure 2. The period during which the diagnosis of the metabolic disorder was made.

When asked about the main caretaker for the patients, the majority (86.49%) of the subjects had their mother as the main caregiver, while 8.11% had the mother together with the maid to take care of them. Those who responded by saying they took care of themselves was only 2.70% and were adults. Around three percent also agreed to have both their parents care for them along with themselves.

When the participants were asked about whether their metabolite levels (depending on their disease) remained in control, only 10.81% agreed while 59.46% sometimes said the levels fluctuated. Twenty-four percent rarely had their levels under control while 5.41% never had it under control (Figure 3).

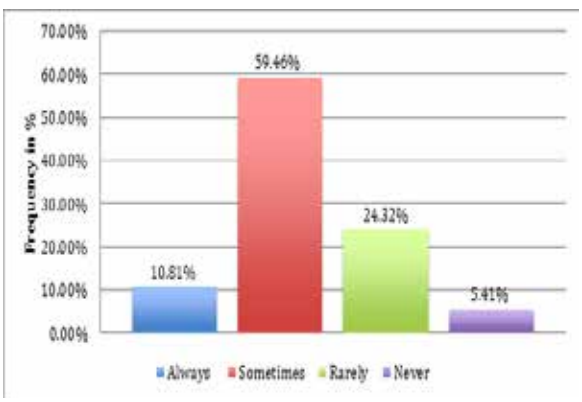

Figure 3. Illustration of the blood serum level of the metabolite in concern under control in the patient.

From among all the subjects, 78.38% said they got their special diet food from the hospital while 16.22% prepared at home following the recipe books provided to them by the hospital. Only 5.41% brought what was convenient from the local supermarkets.

When asked about their dietary restriction, 78.38% agreed to stick to their diet strictly while 10.81% each, denied and sometimes stuck onto their diet.

As for the liking of the taste of the medical food, 62.17% participants liked the taste of their medical dietary food while 27.03% sometimes liked the taste and 10.81% did not like the taste of the medical food at all. This meant that 10.81% most probably did not restrict their diet according to their condition thus leading to fluctuations in the metabolite levels in their system.

When asked about the number of meals per day each patient took, 81.08% said they took their meal 3-4 times per day, while 16.22% took their meal up to 3 times per day and 2.70% took their meal 5 to 6 times per day. Medication was not used often by the participants; only 29.73% used medication while the other 70.27% relied on diet therapy.

The pie chart in Figure (4a) shows that 81% of the participants were interested in reading food labeling before buying any product. While 8% of the participants were not interested in reading food labeling before buying any product at all. Figure (4b) shows 65% of the participants sometimes found enough information on the food labeling, whereas those who said they never found enough information as per their needs on food labels was only three percent. Twenty-one percent always found enough infor-

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mation that they need on the food labels while 11% rarely found enough information.

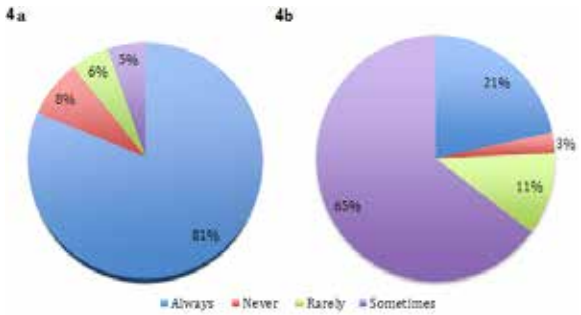

Figure 4. The pie chart (4a) shows the interest among Participants in food label reading. The pie chart labeled (4b) shows how many Participants have actually found the food labels to be of value.

Table 2 illustrates the evaluation of different food consumption among the participants. The majority of the subjects avoided the foods that were to be strictly avoided while a few did eat them occasionally. The foods that were to be limited to some extent and the foods to be eaten freely were consumed more regularly. Even the limited foodstuffs were taken periodically by the majority of the participants. Fruits, fruit juices, and vegetables that were to be taken in limited amounts were being consumed by all the subjects without any restriction.

Table 2. Assessment of consumption of foods to be avoided, limited or free taken by the participants.

Category	Food Item	Yes	No
Food to be avoided	Meat	8%	92%
	Turkey	3%	97%
	Fish	5%	95%
	Chicken	8%	92%
	Milk	14%	86%
	Cheese	22%	78%
	Ice-cream	46%	54%
	Yogurt	27%	73%
	Egg	5%	95%
	Beans	8%	92%
	Nuts	22%	78%
	Peanut butter	8%	92%
Food to be Limited	Fruits	100%	0%
	Fruit Juice	100%	0%
	Vegetables	100%	0%
	Bread	59%	41%
	Chips	89%	11%
	Pop-corn	73%	27%
Food to be taken freely	Low protein food	84%	16%
	Soda drink	54%	46%
	Lollipops	76%	24%
	Honey	73%	27%
	Tang	62%	38%
	Coffee	41%	59%
	Tea	68%	32%

DISCUSSION

The aim of this study was to define the nutritional problems faced by inborn metabolic disorder patients. A newborn screening test done at birth protects against many complications associated with metabolic disorders. In this study, 35.14% were diagnosed at birth, and this may be due to having a family history of the same disease, and 54.05% among the participants already knew the presence of the disorder in their family. One study found that the death of new born infants associated with bacterial sepsis may occur in about 30% of those with untreated classical galactosemia. In most of these deaths, diagnosis and treatment were delayed until the second week of life (Eggertsen, Schneeweiss, & Bergman, 1980). Galactosemia screening should be routine for all new born infants to prevent the complications with simple and inexpensive screening. Since most of the participants had phenylketonuria, we focused our discussion on that group mainly, but the problems can be generalized to the other disorders as well.

Diet therapy is the mainstay in the treatment of PKU (MacDonald, Rocha, Van Rijn, & Feillet, 2011; MacDonald, Evans, Cochrane, & Wildgoose, 2012) and if started within the first weeks of life and continued throughout life, it can prevent intellectual disability and other clinical presentations (Acosta & Yannicelli, 2001). Diet therapy has improved in many countries both regarding nutritional quality and palatability of specialist dietary products (MacDonald et al., 2004; MacDonald et al., 2006; MacDonald et al., 2011). In this study, a percentage of 62.16% of the participants like the taste of the formula and this because of the improvements in the protein substitute formula and this, in turn, helped in dietary compliance as mentioned by Giovannini et al. (2012); where 78.38% followed their diet strictly (Giovannini, Verduci, Salvatici, Paci, & Riva, 2012).

Mean plasma phenylalanine level is considered as a sign of dietary adherence in PKU patients (Alaei, Asadzadeh-Totonchi, Gachkar, & Farivar, 2011). We found from among the participants having tyrosinemia or PKU, 10.81% & 59.46%, were either not complying to the blood levels or were sometimes off the range. Studies have reported it is common for blood phenylalanine level in PKU patients to be above the recommended range especially in teenagers and adults, and adherence to the diet is often poor especially when the patients reach adolescence (Beckhauser et al., 2011; MacDonald et al., 2004).

Caregivers could be a factor that affects the dietary adherence in participants. A high percentage of participants follow their diet restriction count about 78% and from our point of view, this proportion is due to a large number of Participants whom their mothers are their main caregiver.

Low-protein cookbooks offer another option for increasing the variety of foods available for inclusion in PKU diet (Casey, 2013) and other protein disorders. In this study, a percent of 16.22% of participants cooked their food at home. Moreover, that is attributed to the fact that King Faisal Specialist Hospital offers low-protein cookbooks to their patients free of charge. In this study, 81% of the participants consumed 3-4 meals per day, because if meals are distributed throughout the day, the more controlled PHE concentration in each meal.

The intellectual impairment is also much more closely linked with the quality of metabolite control than previously recognized and there appear to be two components to this association. General ability is closely associated with phenylalanine control in the preschool years and to a lesser extent in the preadolescent years. Also, performance on executive

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tasks depends on phenylalanine control (Smith et al., 1993). In our study, 56% of the participants suffered from a variety of complications when they eat food they should be avoiding. Moreover, some of the caregivers said that their children may suffer from seizures, mode fluctuation, nervousness and hair color change. The other Participants who do not have complications when they eat avoided food may be due to having the mild PKU of non-PKU-PHA.

Most of the participants faced problems in following their diet (62.16%). Moreover, some of them said that they do not like the formula's taste. They also seemed to get bored when repeating the same food. Moreover, the common reason was difficulty in finding suitable food during traveling, family gathering, restaurants, and wanting to eat like their peers.

Several studies approved that the diet therapy is also the mainstay of the treatment for Amino Acid Metabolism disorder, glycogen storage disease, tyrosinemia, and galactosemia and the treatment should be started in the first week and continued throughout life (Acosta & Yannicelli, 2001; Fernandes et al., 2006; MacLeod & Ney, 2010). Yet many in this survey avoided strict dietary restrictions leading to abnormal metabolite elevations and side effects.

When asked the participants "If you could ask the Ministry of Health for improvement regarding nutrition and diet therapy for your disorder, what would it be?" their recommendations included; create more food alternatives and make it available everywhere and not just in hospitals and, provision of appropriate food choices in the schools and universities. The positive comment from the participants was the treatment, and the medical food was available for free for Saudi and non-Saudi patients in King Faisal Specialist Hospital.

This study was basically done to determine the nutritional problems faced by inborn metabolic disorder patients, and the result shows that nearly all participants get their treatment and medical food at no cost which is a good thing to help them follow the diet more conveniently and reduce the complications associated with these diseases. The most common problems they faced were the lack of support in the surrounding environment, lack of food variety, and unfavorable taste of the dietary formula.

These patients have special needs, and they need help to integrate into the community. So their health and food need in all aspects of life; schools, universities, hospitals, and restaurants should be taken into account. Provision of newborn health screening test at least for the more common of the inborn metabolic disorder should regularly be made in all hospitals. As of now, most patients were referred to only King Faisal Specialist hospital for treatment as well as for dietary management of the disorder. Once the problems faced are recognized, they can be brought to notice to the higher authorities to make diet source and therapy more readily available for the patients in all hospitals.

Acknowledgement
I am grateful to Ms. Eman Mohammed Megdad, who guided my students in data collection. I am also thankful to my students, Hadeel Al-Dubaikhi, Haneen Al-Madany, Razan Al-Gadiby for helping in the collection of data. I also thank King Faisal Specialist Hospital and Research Center and King Fahd Medical City for their cooperation.

Ethical approval
The study was approved by the IRB and ethics committee of the College of Applied Medical Sciences, King Saud University, Riyadh, Saudi Arabia.

Conflicts of interests
None declared.

Reference List

1. Acosta, P. B. & Yannicelli, S. (2001). The Ross metabolic formula system nutrition support protocols. Abbott Laboratories, 4th Edition.2001. [Links].
2. Aida Al Aqeel, M. D. (2004). Common genetics and metabolic diseases in Saudi Arabia. Middle East Journal of Family Medicine, 6.
3. Alaei, M., Asadzadeh-Totonchi, G., Gachkar, L., & Farivar, S. (2011). Family social status and dietary adherence of patients with phenylketonuria. Iranian journal of pediatrics, 21, 379.
4. Beckhauser, M. T., Peruchi, M. M., de Luca, G. R., Lin, K., Esteves, S., Vilarinho, L. et al. (2011). Neuroradiological findings of an adolescent with early treated phenylketonuria: is phenylalanine restriction enough? Clinics and practice, 1.
5. Casey, L. (2013). Caring for children with phenylketonuria. Canadian Family Physician, 59, 837-840.
6. Eggertsen, S. C., Schneeweiss, R., & Bergman, J. J. (1980). An updated protocol for pediatric health screening. J Fam.Pract., 10, 25-37.
7. Fernandes, J., Saudubray, J. M., Van den Berghe, G., & Walter, J. H. (2006). Inborn metabolic diseases: diagnosis and treatment. Springer Science & Business Media.
8. Giovannini, M., Verduci, E., Salvatici, E., Paci, S., & Riva, E. (2012). Phenylketonuria: nutritional advances and challenges. Nutrition & metabolism, 9, 1.
9. MacDonald, A., Davies, P., Evans, S., Daly, A., Hall, S. K., Hendriksz, C. et al. (2006). □ÇÿReady to drink□ÇÖprotein substitute is easier is for people with phenylketonuria. Journal of inherited metabolic disease, 29, 526-531.
10. MacDonald, A., Evans, S., Cochrane, B., & Wildgoose, J. (2012). Weaning infants with phenylketonuria: a review. Journal of Human Nutrition and Dietetics, 25, 103-110.
11. MacDonald, A., Lilburn, M., Cochrane, B., Davies, P., Daly, A., Asplin, D. et al. (2004). A new, low-volume protein substitute for teenagers and adults with phenylketonuria. Journal of inherited metabolic disease, 27, 127-135.
12. MacDonald, A., Rocha, J. C., Van Rijn, M., & Feillet, F. (2011). Nutrition in phenylketonuria. Molecular genetics and metabolism, 104, S10-S18.
13. MacLeod, E. L. & Ney, D. M. (2010). Nutritional management of phenylketonuria. Annales Nestl+¬ (English ed.), 68, 58-69.
14. Smith, I., Cockburn, F., Barwell, B., Brenton, D., Chapple, J., Clark, B. et al. (1993). Recommen