Knowledge and Awareness of Metabolic Inborn Errors among Male and Female Students at King Abdulaziz University – Rabigh.

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ABSTRACT

***Context**: Inborn errors of metabolism are a large group of rare genetic diseases. The incidence of inborn metabolic errors, collectively, as high as 1 in 2500 live births, but it varies greatly and depends on the population. Presentation is usually in the neonatal period or infancy but can occur at any time. Diagnosis does not require extensive knowledge of biochemical pathways or individual metabolic diseases. **Aims:** To assess the students' knowledge about IEM and to plan future improvement in the curriculum if needed and to plan and organize awareness campaigns. **Settings and Design:** A cross-sectional study was conducted on 400 undergraduate medical and non-medical students. **Methods and Material:** The study included 400 medical and non-medical students (including 202 males & 198 females) from the Rabigh campus. The questionnaire was constructed and translated into Arabic. The frequencies of different categories were evaluated for their statistical significance. **Results:** Among male students, trivial differences were observed between the answers of medical and non-medical students, both groups agreed on the fact that metabolic disorders can be caused by an enzyme deficiency. However, 36% of medical students and 43% of non-medical students did not know that metabolic disorders can be diagnosed prenatally. As regards the female students, 43% of non-medical students accepted that diagnosis usually needs invasive procedures, while 43 % of medical students refused that concept. On comparison of male and female students, 50 % of male students had no idea of the usefulness of gene therapy as a tool for management, yet 43 % of females agreed on the importance of gene therapy. **Conclusion**: More efforts should be exerted to raise the level of awareness and improve the knowledge among both medical and non-medical students and the community in Saudi Arabia by increasing the taught material regarding genetic diseases in universities and even introduce it in school.*

Introduction

In 1908, Sir Archibald Garrod was the first to describe Inborn Errors of Metabolism (IEM) [1]. IEM are a group of more than 750 molecular disorders [2] with multi-organ manifestations [2, 3]. They are mostly inherited as autosomal recessive[1, 2, 4, 5]. It could be inherited on rare occasions as autosomal dominant or X-linked [1, 4, 5]. Other factors like infections and environmental factors may play an important role in IEM [4]. IME should be

suspect in families with a history of IEM [5]. They may present at any age and affect any organ [1, 2, 6]. They could cause complete, partial, or incomplete dysfunction in the pathway of metabolizing proteins, fatty acids, and carbohydrates [4].

IEM collectively affects 1 in 2500 births [4], which makes them common but rare diseases when discussed individually [2-5]. Saudi Arabia has a high rate of IEM reported to be around 1:667 birth due to relative marriage [1]. IEM causes errors in the pathway of metabolizing proteins, fatty acids, and carbohydrates or in glycogen storage [3, 4], which creates a wide range of symptoms and signs.

IEM can be divided into three categories: Energy deficiency, intoxication by accumulating of the product before the deficient enzyme in the pathway, or it could be an issue in metabolizing complex molecules [1, 5]. Unspecific symptoms may appear on the infant with IEM any time from hours to weeks after delivery [5].

The patient could present with hypoglycemia, anemia, vomiting, failure to thrive, seizures, hypotonia, coma, recurrent infection, psychomotor retardation, or even dead. On examination and investigation, the patient could have hepatomegaly, splenomegaly, heart failure, dilated hypertrophic cardiomyopathy, acid/base imbalance, hypoglycemia, or hyperammonemia [2, 4, 5]. Routine newborn screening is a handy tool to identify IEM even before developing symptoms by simple urine or blood tests [1, 2, 5]. Expecting mothers should know that some IEM can be diagnosed prenatally [1, 2, 5]. The general practitioner plays an essential role in diagnosing and start managing IEM [6].

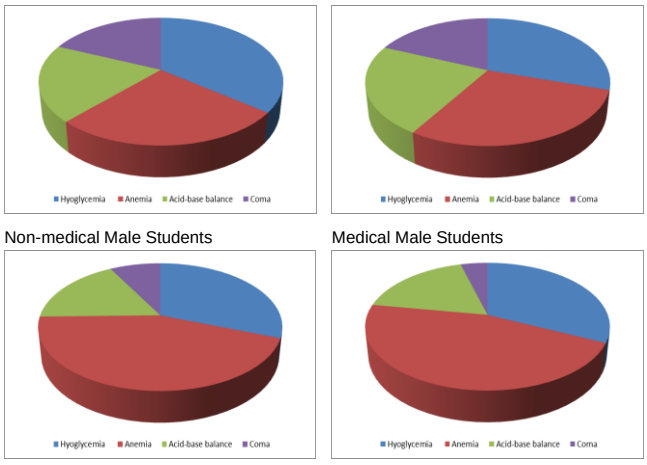
Fast and effective interventions are essential in treatable IEM to avoid irreversible complications [2]. In families with a history of IEM, management can be started before delivering the baby, and it should be in a prepared center that can manage IEM immediately [5]. Some IEM can be managed by medications and enzyme replacement and other only by avoiding certain types of foods and supplements [1, 5]. A liver transplant can be the only way to treat some types of IEM [1, 5]. Some types of IEM can be treated by hematopoietic stem cell transplantation [7]. One of the strategies to treat IEM is gene therapy [8]. Unfortunately, some IEM still don't have an appropriate therapy [5]

Subjects and Methods

By conducting a cross-sectional study included 400 medical and non-medical students (including 202 males & 198 females) recruited from the Rabigh campus. The questionnaire was constructed and translated into Arabic. After a brief explanation of the purpose of the study to the participants, they filled in the survey separately. The frequencies of different categories were evaluated for their statistical significance.

Results

In our sample, 400 students from Rabigh campus, included 202 males (50.5%) and 198 females (49.5%). Of all the sample, only 59 (14.75%) students from medical school and the rest 341 (85.25%) students from other schools. The majority believe that IEM can be inherited (57% females, 58% males). Regarding infection as a causative agent of IEM, there was a split. 40% of females and 41% of males said it could be a cause. While 49% of females and 39% of males disagree with them. IEM are common in both genders, as noted by 48% of females and 42% of males. 34% of females don't see a relation between family history and IEM, and another 34% of them don't know if there is a connection while 41% of males aware of the role family history plays in IEM. Females were divided pediatric and adult age groups (38% each), and 34% of males choose the adult age group as the age group when IEM symptoms appear.



Non-medical Male Students Medical Male Students

Non-medical Female Students Medical Female Students

Figure 1: Students' response about the most common symptoms of metabolic inborn errors.

In the male sample, medical students make about 17.8% of the sample, and the non-medical make the rest 82.2%. The majority of the male medical students (69%) agreed that infection my cause IEM, while 43% of non-medical disagree. Almost all the medical students (97%) aware that IEM can be caused by enzyme deficiency and that enzymes play an essential role in body metabolism while more than half (59% and 70% respectively) of the non-medical students aware of that. 45% of non-medical sample chose that IEM occur equally in both genders, and 41% knows that family history plays an important role. On the other hand, 36% of the medical students chose that IEM occurs more in females, and 39% of them aware of the importance of family history, and surprisingly 31% don't know if family history has any role. Among the non-medical sample, the majority (37%) believes that IEM symptoms appear in adults while medical students were divided between neonatal and pediatric ages (28% both). While 36% of medical students don't know if some IEMs can be diagnosed with a blood sample only, 31% of medical and 42% of non-medical agrees that it could be that simple. 44% of medical and 39% of non-medical thinks that there is no available treatment for most of IEM.

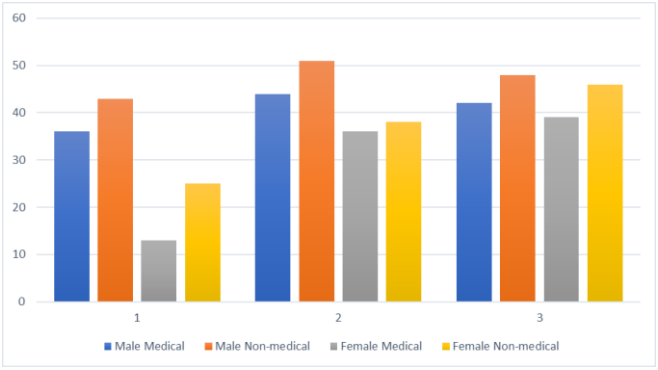


Figure 2: shows lack of knowledge among some students (percentage of students who answered by "Don't know") for the following questions:

1- Diagnosis of some metabolic disorders can be done prenatally?
2- Gene therapy can be a helpful tool for treatment of some metabolic disorders?
3- Liver transplantation is an option in management of some metabolic disorders

In the female sample, 11.6% of the sample are medical students, and 88.4% are non-medical students. Of medical students, 61% said that it could be caused by infection, and 100% agreed that it could be caused by an enzyme deficiency. Of non-medical students, 51% disagree that it could be caused by infection, and 66% aware that it could be caused by an enzyme deficiency. 43% of medical student believe that IEM are more common in females, while 50% of non-medical students believe that it affects both genders equally. The majority of medical students aware of the role of family history in IEM. On the other hand, 35% of non-medical students don't see a connection between IEM and family history, and another 35% don't know about it. 43% of medical students said pediatric age is when symptoms start, and 41% of non-medical students choose adult age. 43% of medical students don't believe that making the diagnosis needs invasive procedures, while the same percentage of non-medical students believe the opposite. 52% of medical and 41% of non-medical students agree that most IEM have an available treatment.

Discussion

We couldn't locate any previous publication discussing the awareness of IEM among university students in Saudi Arabia.

In our study sample, the numbers of male and female students were almost equal. Around 15% of the sample were medical students. In the female sample, medical students had better knowledge of the causes of IEM, especially that the infection can cause IEM and family history is one of the IEM risk factors. Also, medical students had better awareness of the method of diagnosing IEM. Neither the medical student nor non-medical students had enough knowledge about IEM management.

Regarding the male sample, medical students had better awareness of the etiologies of IEM than non-medical. Surprisingly, non-medical students know more about family history than medical students. As for diagnosing IEM, both medical and non-medical students need more information. Another part that should get more attention in the future for both medical and non-medical students is managing IEM patients.

Comparing knowledge of all males and females in our sample, both had great information on the causes of IEM. Both answered that IEM is equally common in males and females. Males know more about the association between IEM and family history. Both believe that IEM are more common in pediatric and adult age groups than the infant age group. Although there were more critical and severe symptoms for IEM, anemia, and hypoglycemia were the most chosen answers. In spite of the fact that both males and females don't have enough information about IEM diagnosing methods, females are more aware than males, especially in the part of prenatal diagnosis. Liver transplant and gene therapy are some areas where awareness campaigns should focus on both males and females.

Conclusion

Even though medical students had more knowledge than non-medical students, they have some misconceptions and missing information. In general, females are more aware of IEM than males; it could be because females are the one who gets pregnant or they heard about it from other pregnant ladies. IEM are a big group of diseases that need more attention and awareness campaigns directed to the general public as well as to medical students. This can be achieved by establishing a unique education program that focuses on the inheritable nature of these disorders and the importance of routine neonatal screening program with a final emphasis that appropriate treatment is available in the Kingdom.

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Disclaimer

The article has not been previously presented or published and is not part of a thesis project.

Conflict of Interest

There are no financial, personal, or professional conflicts of interest to declare.

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