Knowledge and practice among primary care physicians in Najran (south west region), Saudi Arabia regarding Maturity Onset Diabetes of the Young (MODY)

Mohammed Ayed Huneif

Correspondence:
Dr. Mohammed Ayed Huneif
Department of Pediatrics, College of Medicine
Najran University
Najran , Saudi Arabia
Email: huneif@hotmail.com

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Abstract

Background: Maturity Onset Diabetes of the Young (MODY) is a disease that is caused by a single gene. This indicates that it is the result of a single gene mutation. MODY can be caused by a variety of gene mutations. When a family member who has MODY, relatives are at a higher risk of developing the disease. Maturity Onset Diabetes of the Young is a kind of diabetes mellitus that is inherited. A mutation in one of eleven genes causes it. MODY could be the cause of up to 5% of all diabetes cases. MODY patients, like other diabetics, have difficulty controlling their blood sugar levels.

Methods: This was a cross-sectional study among primary care physicians (PCPs) in the Najran region (13). The questionnaire was created following a series of conversations between a panel of experts, which included subject specialists, researchers, and language experts. Pilot study with 15 respondents was also conducted to observe the clarity of the content of the questionnaire and its validity. The questionnaire's Cronbach alpha was computed. It was created in English and disseminated through Google as well as on printed forms. The forms had two main sections, one for knowledge and the other for practice regarding MODY.

Results: 12% responding completely regarding definition of MODY, 55.5% have knowledge regarding mode of inheritance of Mody, 9% were aware about the type of the MODY,60% were about typical symptoms of MODY, Diagnostic tools of MODY (45%), Possible presentations of MODY (6%), Therapeutic modalities of MODY (39%), Complications of MODY (20%)Differences between Mody and T1DM and T2DM 6.80 while overall knowledge score was 53.8%

Conclusion: Clinicians should maximize alternative therapy in the era of CF modulators and correctors to improve outcomes and prevent long-term morbidity and mortality.

Key words: knowledge, practice, primary care physicans, Maturity Onset Diabetes of the Young (MODY)

Background

Maturity Onset Diabetes of the Young (MODY) is a disease that is caused by a single gene. This indicates that it is the result of a single gene mutation. MODY can be caused by a variety of gene mutations. When a family member who has MODY, relatives are at a higher risk of developing the disease. Maturity Onset Diabetes of the Young is a kind of diabetes mellitus that is inherited. A mutation in one of eleven genes causes it. MODY could be the cause of up to 5% of all diabetes cases. MODY patients, like other diabetics, have difficulty controlling their blood sugar levels(1-3).

MODY is a type of familial diabetes with an early onset (in childhood, adolescence, or young adulthood) and an autosomal-dominant mode of transmission (shown by the existence of three generations of the same afflicted lineage) linked to anomalies in the insulin secretion sphere (2-5).

When compared to type 1 and type 2 diabetes, MODY is extremely rare; scientists believe that only 1–2% of people with diabetes (20–40,000 people) in the UK have it. Because MODY is so uncommon, doctors may be unaware of it, and it is estimated that 90% of patients with it are initially misdiagnosed with type 1 or type 2 diabetes (6-8).

The two most common types of monogenic diabetes are neonatal diabetes mellitus (NDM) and maturity-onset diabetes of the young (MODY). NDM is a condition that affects neonates and infants. MODY is far more frequent than NDM, and it usually begins in adolescence or early adulthood (9-10).

Stefan S. Fajans, an American researcher, described for the first time in 1960 a group of non-obese children and adolescents with a strong family history of diabetes mellitus who had mild diabetes mellitus and achieved good metabolic control with the use of sulfonylurea after several years of observation. MODY has been confirmed through the use of genome scanning strategies, carried out in several families with a clinical diagnosis of MODY (9).

A study reported that MODY accounts for up to 2% of all diabetes cases in people aged 20 and under in the United States. MODY is caused by a variety of distinct gene mutations, all of which restrict the pancreas' ability to make insulin. This causes high blood glucose levels, which can harm bodily tissues such as the eyes, kidneys, neurons, and blood vessels over time. GCK-MODY (MODY2) and HNF1A-MODY (MODY3) are the two most prevalent causes of MODY, accounting for 30 percent to 60 percent of all MODY cases. GCK-MODY is expected to affect one out of every 1,000 people (11-12).

The prevalence of MODY has not yet been defined, but it is estimated that 2-5% of individuals considered to have DM2 are actually MODYs. The prevalence of GCK-MODY is higher in some countries (the United States, Germany, Italy, France, and Spain) than in others (most likely due to biased assessment of children versus adults) (11-12).

For illness prevention, early diagnosis and treatment, and public knowledge of genetic disorders is critical. Furthermore, institutions must be made aware of the importance of providing the finest possible healthcare, social, and environmental amenities for patients, their families, and care managers. Due to a lack of knowledge and practice and likely a lack of awareness of the disease at the level of primary care physicians, any disease including MODY will increase.

The aim of this study is to determine the knowledge and practice of primary care physicians about MODY and to make recommendations.

Methods

This was a cross-sectional study among primary care physicians (PCPs) in the Najran region of Saudi Arabia. Primary healthcare centers (PHCCs) are regarded as the patient's primary point of contact with the healthcare system. Many countries use health indicators to assess the quality and operation of their primary healthcare systems. Saudi Arabia has been working to integrate preventative and primary curative healthcare services into PHCs, which now provide a wide range of treatments. Najran is a city in Saudi Arabia's southwest, close to the Yemeni border. It is the provincial capital of Najran. Najran, which has been designated as a new town, is one of the kingdom's fastest-growing cities, with a population that has increased from 47,500 in 1974 to 90,983 in 1992 to 246,880 in 2004 and 505,652 in 2017 (13). The questionnaire was created following a series of conversations between a panel of experts, which included subject specialists, researchers, and language experts. A pilot study with 15 respondents was also conducted to observe the clarity of the content of the questionnaire and its validity. The questionnaire's Cronbach alpha was computed. It was created in English and disseminated through Google as well as on printed forms. The form had two main sections, one for knowledge and the other for practice with MODY. There were 8 knowledge questions and three practice questions that examined various aspects of MODY. Each question had correct and incorrect responses, as well as the possibility of selecting more than one option. For each question, there were three options: complete answer, incomplete response, and incorrect answer. Complete implies the respondent selected all correct answers, incomplete meant the responder selected some correct and some incorrect answers, and wrong means the responder selected all incorrect answers. The total knowledge score was computed based on all correct answers to all questions, and each respondent's total score was calculated based on his or her correct responses. Age, gender, years of experience after graduation and in a primary care setting, nationality, job nature (resident, specialist, family medicine consultant), and whether the responder had ever attended the yearly educational pediatric club meeting were all included in the first section of the questionnaire. We used a convenient sampling method. After collection of data, data was coded and entered in the SPSS Ver. 20 software for analysis; descriptive statistics were computed. The

median and percentage out of the total scores were used to compute the total knowledge and practice scores. The sum of all correct answers was used to calculate the total knowledge score, while the practice score was out of five points.

The study was approved by the research ethical committee of the Najran University; informed consent was obtained from the respondents, and the questionnaire was anonymous.

Results

45 respondents successfully completed the questionnaire out of 54, so the response rate was 83.3%. Cronbach alpha of the questionnaire was 0.79. Out of 45 respondents 25 (55.5%) were of age group less than 40 years. The male doctor respondents were 88.8% and females were 11.2%, out of 45 respondents. 77.7% had been performing their duties in the PHCC for less than 5 years while 13.3% had been working for more than 10 years witth professional responsibilities. 77.7% were working as a general practitioner while others were specialists (11.11%) and family medicine consultant (11.1%); 13.4% attended a training course or a conference on management of MODY while only 12.2% prescribed sulfonylureas (Table 1).

Table 2 shows that 12% responded completely regarding definition of MODY, 55.5% have knowledge regarding mode of inheritance of MODYy, 9% were aware about the type of the MODY, 60% were aware of typical symptoms of MODY, diagnostic tools of MODY (45%), possible presentations of MODY (6%), therapeutic modalities of MODY (39%), complications of MODY (20%), differences between MODY and T1DM and T2DM 6.80, while overall knowledge score was 53.8% (Table 2).

The overall practice score percentage was 48% with a mean of 5.4 and maximum and minimum scores were 5 and 1, respectively. Regarding the practice of PCPs about when to refer suspected

MODY patients, 18.6% responded correctly and the remaining either answered incorrectly or incompletely. More than 40% of the responders answered correctly about treatment of MODY. On the other hand, 47.8% knew the standard of care to follow up with children who are known to have the diagnosis of CF (Table 3).

Discussion

This is the first study in Najran to analyze PCPs' knowledge and practice about MODY in Saudi Arabia's southwest region. Misconceptions, gaps, and inaccuracies in MODY knowledge could lead to inadvertent non-adherence to treatment, which could affect the disease's course and outcome.

MODY was found to be responsible for 2.4 percent of diabetes cases in children under the age of 15 in Saxony, Germany. The most frequent type of monogenic diabetes is MODY. Adults have a prevalence of about 1/10,000, whereas children have a prevalence of about 1/23,000. There has been no mention of any ethnic preferences. It is estimated that about 80% of cases are misdiagnosed as type 1 or type 2 diabetes, confounding estimates of prevalence and incidence (14-16).

MODY is the final diagnosis in 1%–2% of patients who have been diagnosed with diabetes. The prevalence is 70–110 persons per million. The identical mutation will be inherited by 50% of first-degree relatives, putting them at a greater than 95% lifetime risk of getting MODY. As a result, proper diagnosis of this illness is critical (17-19).

Apart from glucokinase, all types of MODY carry a risk of long-term diabetes complications, so patients should eat a healthy balanced diet and stay physically active to help maintain good blood glucose and cholesterol levels, which reduces the risk of complications (19).

This research emphasized the importance of guidelines that are clear and defined across the country for diagnosis of the problem and initiating the necessary treatment. However, we need a true and reliable baseline before creating such treatment centers for MODY (20).

Affected individuals and families should receive genetic counseling to learn about the nature, mode(s) of inheritance, and implications of genetic abnormalities so they can make informed medical and personal decisions. According to one study conducted in Kenya, the total prevalence of MODY is estimated to be 1–5 per 10,000 people, accounting for 1–5% of all diabetes mellitus cases. Among Western Australians, however, the prevalence of MODY in diabetic individuals under the age of 35 years is 0.24 percent, equating to an estimated minimum prevalence of 89 instances per 1,000,000 for the entire Australian population (22-23).

In our study we have observed the lack of information and misconceptions regarding MODY. We need a series of seminars and educational activities to educate doctors regarding MODY.

Conclusion

This study emphasizes the importance of PCPs participating in intensive teaching programs in order to promote early detection of MODY and begin proper treatment of MODY. Clinicians should maximize alternative therapy in the era of CF modulators and correctors to improve outcomes and prevent long-term morbidity and mortality.

Defective transcriptional regulation, aberrant metabolic enzymes, protein misfolding, malfunctioning ion channels, and impaired signal transduction are all involved in the etiology of MODY. In order to effectively identify patients, individualize patient therapy and follow-up, and screening of family members of afflicted individuals for diabetes mellitus is required. Clinicians need to have a complete grasp of the epidemiology and etiology of MODY.

Table 1: Demographic characteristics of the responding physicians	
• Age in years	
• Less than 30 years old	25 (55.5%)
• 30-40 year	15 (33.3%)
• 41-50 years	2 (4.3%)
• More than 50 year	3 (6.67%)
• Gender	
• Female	5 (11.2 %)
• Male	40 (88.8 %)
• Qualification	
• MBBS	40 (88.8%)
• Diploma	0 (0%)
• Master	4 (8.8%)
• Doctorate/PhD/Fellowship	1 (2.4)
• Others	0 (0.0%)
• Years of experience in the primary health care field	
• 5-10 years	4 (8.8%)
• Less than 5 years	35 (77.7%)
• Over 10 years	6 (13.3%)
• Nationality	
• Arabic other than Saudi	2 (4.4%)
• Non-Arabic	8 (17.7%)
• Saudi Arabia	35 (77.7%)
• Job nature	
• Consultant	5 (11.11%)
• General practitioner	35 (77.7%)
• Specialist	5 (11.11%)
• Have you ever attended conference meeting in Diabetes?	
• No	20 (45%)
• Yes	25 (55%)
• Attending a training course or a conference on management of MODY ?	
• No	39 (86.6%)
• Yes	6 (13.4%)
• Have you ever prescribed sulfonylurea to your patients during the last year?	
No	40 (88.8%)
Yes	5 (12.2%)

Table 2: Responses about MODY knowledge among primary care physicians

Items	Responding completely	Responding Incompletely	Responding wrongly
Definition of MODY	12%	45%	33%
Mode of inheritance MODY	55.50%	10%	34.50%
Awareness regarding types of MODY	09%	40%	51%
Typical symptoms of MODY	60%	10%	30%
Diagnostic tools of MODY	45%	19%	36%
Possible presentations of MODY	6%	45%	49%
Therapeutic modalities of MODY	39%	40%	21%
Complications of MODY	20%	25%	55%
Differences between MODY and T1DM and T2DM	6.80%	25%	68.20%
Overall knowledge score (percent)	53.80%		

Table 3: Responses about MODY practice among primary care physicians

Items	Responding completely	Responding Incompletely	Responding wrongly
Referring suspected cases of Mody to specialized center	18.6%	20.0%	61.4%
Treating of Mody	42.0%	32.0%	26.0%
Regular follow-up of patients with Mody	47.8%	32.0%	20.2%
Overall practice score	48.0%	20.0%	32.0%

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