

Assessment of Knowledge and practice of Maturity Onset Diabetes of the Young (MODY) among Primary care physicians in Najran, Saudi Arabia

Mohammed Ayed Huneif

Endocrinologist at Department of Pediatrics, College of Medicine, Najran University Hospital, Najran University, Najran, Saudi Arabia.

Corresponding Author:

Dr. Mohammed Ayed Huneif

Endocrinologist at Department of Pediatrics, College of Medicine, Najran University Hospital, Najran University, Najran, Saudi Arabia, PO Box: 1988.

Email: huneif@hotmail.com

<https://orcid.org/0000-0002-0497-1029>

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Abstract

Background: Maturity-onset diabetes of the young (MODY) is a type of monogenic diabetes first described as a mild and asymptomatic form of diabetes mellitus that was observed in non-obese children, and young adults. With the development of genetic technology, many genes linked to MODY have been sequenced and described. MODY is a kind of diabetes inherited and 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 study is a cross-sectional study among primary care physicians (PCPs) in the Najran region. 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 forms link and in a printed form as well. The questionnaire has two main sections, one for knowledge and the other for practice with MODY.

Results: 12% completely responded to questions 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 aware 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 (6.8%), while overall knowledge score was 53.8%.

Conclusion: This study emphasized the importance of PCPs among individuals participating in intensive teaching programs in order to promote early detection of MODY and beginning of the proper treatment for the disease.

Keywords: Knowledge, practice, primary care physicians, Maturity Onset Diabetes of the Young (MODY).

Background

Maturity Onset Diabetes of the Young (MODY) is the most common type of monogenic diabetes that is inherited in an autosomal dominant pattern. Advances in genetic technology have emphasized that, MODY can be caused by a variety of gene mutations (1). MODY appears as a result of single gene mutation that leads to beta-cell dysfunction which impairs insulin secretion. When a family member has MODY, relatives are at a higher risk of developing the disease. Some types of MODY are associated with extra-pancreatic manifestations involving liver, intestine, kidneys, and genitourinary dysfunctions (2). MODY genes noted in some syndromes like Wolfram Syndrome 3. Earlier, MODY was recognized as six subtypes but currently it is classified by genetic defect to at least 14 different types according to the involved genes (4). The most common types are hepatocyte nuclear factor 1 alpha (HNF1A), hepatocyte nuclear factor 4 alpha (HNF4A), glucokinase (GCK), hepatocyte nuclear factor 1 beta (HNF1B) respectively (5). Less common types include: KCNJ11, ABCC8, INS, PDX1, PAX4, NEURO1, CEL, KLF11, BLK and APPL1 (4,5).

Initially, MODY was described as asymptomatic mild diabetes that was noticed in children, adolescents, and young adults (6). It is emerging before the age of 30 years mainly between the ages of 10 to 25 years old (7). Detailed medical history can provide information that help in diagnosis, as some MODY forms, as mentioned above, are associated with extra-pancreatic manifestations (8). Intrauterine growth retardation (IUGR) is noted with HNF1B (MODY 5) and slightly increase in birth weight is seen in those with HNF4A (MODY 1) (4). Depending on the neonatal history, studies showed that MODY, symptoms may include mild fasting hyperglycemia with GCK (MODY 2) or transient neonatal hyperinsulinemic hypoglycemia with HNF1A (MODY 3) (9).

The knowledge and practice of the primary care physicians (PCPs) about clinical presentation, diagnosis, type of treatment and family counseling of MODY is a vital issue regarding the medical care and control of the disease. Wherever, family history is important and supports the diagnosis of MODY, as patients with MODY usually have history of diabetes extending over 3 generations at least specially before the age of 25 years (10,11). Physical examination findings are likely to be unspecific, so basic examinations should be done for any patient with diabetes, and to screen for other associated extra-pancreatic manifestations, syndromes and expected end-organ damage complications that may develop with some types of MODY. MODY can be misdiagnosed with other common forms of diabetes mellitus either type 1 (T1DM) or type 2 (T2DM) diabetes mellitus. Establishing a correct diagnosis of MODY and identification of different subtypes should be based on molecular genetic testing (12). The diagnosis of MODY should be suspected in cases with onset before 25 years of age in a family member with at least extending over three generations, no islet

autoantibodies, stimulated C-peptide >200 pmol/L, less or no insulin therapy requirements for at least 5 years after diagnosis of diabetes (13).

The best treatment for MODY depends on the involved gene in the mutation process. In MODY 2 (GCK), patients need lifestyle and dietary modification alone. MODY 1 (HNF1A) and MODY 3 (HNF4A), at the beginning, managed with dietary changes alone, progressively worse over time and to be managed with sulfonylureas (14). MODY 5 (HNF1B) patients require insulin to control their diabetes (15).

The aim of this study was to find out the knowledge and practice of PCPs about clinical presentation, diagnosis, type of treatment and family counseling of MODY as it is important to make recommendations for proper treatment.

Methods

Study setting

This study represents a cross-sectional study conducted throughout March 2021 in Najran city, which is a city in Saudi Arabia's southwest, close to the Yemeni border. It is the provincial capital of Najran Province. The health services are delivered by many districts that include governmental and private to different degrees.

Inclusion criteria

The study population consists of all physicians who involved in the care of patients with diabetes at the level of primary health services in Najran city, Saudi Arabia at primary health care centers, emergency departments, outpatient clinics and private polyclinic centers outpatient clinics, emergency departments of hospitals, and private polyclinic centers during the study period who accepted to participate in the research study.

Data collection

A questionnaire was designed and formulated following a series of conversations with experts in the same research field. It was created in English and disseminated through Google forms link as well as printed forms. The questionnaire has two main sections: the first part included socio-demographic data determined by age, gender, nationality, Qualification degree, medical specialty, job title, qualification, workplace, healthcare facility, and experience duration. the second part was about the physician's knowledge and practice with MODY. There were 8 knowledge questions and 3 practice questions that examined various parts of MODY. The knowledge questions include Definition, Mode of inheritance, types, clinical presentation, Diagnostic tools, Therapeutic modalities, Complications, Differences between MODY and common types of diabetes mellitus (T1DM, T2DM). The practice questions included timelines of referral of suspected cases with MODY to diabetes centers, action in emergency situations, and follow up care for the proved cases of MODY.

Knowledge assessment

Pilot study with 15 respondents was conducted with their feedback to observe the clarity of the questionnaire and its validity. The questionnaire's Cronbach alpha was computed. Questionnaire template was scored as each question had correct and incorrect answers, 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 when the responder selected all correct answers, incomplete means 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.

Data analysis

We used a convenient sampling method. After collection of data, data was coded, entered in the SPSS ver.20 software for analyses and 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.

Ethical consideration

The study was approved by the research ethical committee of the Najran University. Research purpose and objectives were explained to participants in clear simple words. Informed consent was obtained from the respondents, questionnaire was anonymous.

Results

Demographic characteristics of the participants

Forty-five respondents were successfully completed the questionnaire out of fifty-four, PCPs were enrolled in this study, so the response rate was 83.3%, Cronbach alpha of the questionnaire was 0.79. Out of 45 respondents 25 (55.5%) having age group less than 40 years. The male doctors' respondents were 88.8% and females were 11.2%, out of 45 respondents 77.7% were performing their duties in the primary health care center (PHCC) from less than 5 years while 13.3% working since more than 10 years professional responsibilities. 77.7% MODY were working as a general practitioner while others are 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 sulfonylurea (Table 1).

Participants' knowledge toward MODY

Table 2 showed that, 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 aware 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 (6.8%) while overall knowledge score was 53.8% (Table 2).

Attitude of the participants and management of MODY

The overall practice score percent 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 MODY (Table 3).

Table 1. Demographic characteristics of participants (n = 123).

Demographic variables	Frequency	Percent
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 years	3	6.67
Gender		
• Female	5	11.2
• Male	40	88.8
Nationality		
• Arabic other than Saudi	2	4.4
• Non-Arabic	8	17.7
• Saudi Arabia	35	77.7
Qualification degree		
• MBBS	40	88.8
• Diploma	0	0
• Master	4	8.8
• Doctorate/PhD/Fellowship	1	2.4
• Others	0	0
Job nature		
• Consultant	5	11.11
• General practitioner	35	77.7
• Specialist	5	11.11
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
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	20	45
• Yes	25	55
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	55.5%	10%	34.5%
Types of MODY	09%	40%	51%
Clinical presentation 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 common types of diabetes mellitus (T1DM, T2DM)	6.80%	25%	68.2%
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%

Discussion

This study was conducted to assess the knowledge, attitude, and practices of MODY among PCPs in Najran governorate of Saudi Arabia. To the best of our knowledge, no studies have been done, hitherto, in this region. Hence, this represents the first study in Najran region to analyze PCPs' knowledge and practice about MODY in Saudi Arabia's southwest region. An insight into the awareness among PCPs, of an important problem like MODY, would be a guide to devise effective management strategies.

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 is the most frequent type of monogenic diabetes, was found to be responsible for 2.4 percent of diabetes cases in children under the age of 15 in Saxony, Germany 16. 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's estimated that, about 80% of cases are misdiagnosed as T1DM or T2DM diabetes, confounding estimates of prevalence and incidence 17.

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 18.

According to a previous 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 Australia, 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.19-20

Apart from glucokinase, all types of MODY carry the risk of long-term diabetic complications, so patients with MODY should be kept under diet control with healthy balanced diet and stay physically active to help maintain good blood glucose and cholesterol levels, which reduces the risk of complications.1 Moreover, individuals and families should receive genetic counseling to learn about the nature, mode(s) of inheritance of MODY, and implications of genetic abnormalities, so they can make informed medical

In our study we have observed the lack of information and misconceptions regarding MODY. There is a significant deficiency of knowledge among PCPs in this part of Saudi Arabia, with slightly less than half (46.2%) having poor knowledge. Only 55.5% have knowledge regarding mode of inheritance of MODY and 9% were aware about the different types of the MODY. In addition, 60% of respondents were aware about the symptoms of MODY, 45% were aware by diagnostic tools of MODY, only 6% were aware by the possible presentations of MODY, 39% were aware by the therapeutic modalities of MODY, 20% aware by complications of MODY and only 6.8% were aware by the differences between MODY and T1DM and T2DM. Physicians working at PHC level are at the forefront of healthcare delivery system. The low knowledge score among a significant number of such healthcare workforce is a matter of serious concern.

Regarding attitude towards MODY among PHC Physicians is an important indicator of overall approach in the management of MODY. The overall practice score was 48 % with a mean of 5.4. Concerning the practice of PCPs about when to refer suspected cases of MODY, only 18.6% of respondents responded correctly, more than 40% of the responders answered correctly about treatment of MODY. In addition, 47.8% knew the standard of care to follow up with children with MODY. MODY has been recognized as an important clinical problem that could have serious health repercussions 2-4. The key in preventing such adverse outcomes is early detection of the disease with an effective management 14-15. Primary care physicians work at grassroots and are extremely crucial in detecting such cases at the earliest. With Saudi Arabia having one of the highest prevalence rates of diabetes including MODY, a proactive approach is required to combat this scourge. Hence, series of seminars and medical educational activities should be conducted to improve physician's information, attitude, and practice and to update their knowledge about MODY. This research work emphasized the importance of guidelines that are clear and defined across the country, for proper diagnosis of MODY and initiating the necessary treatment. However, we need a true and reliable baseline before creating such treatment centers for MODY.

Conclusion

This study emphasized the importance of PCPs for participating in intensive teaching programs in order to promote early detection of MODY and beginning the proper treatment of the disease. In order to effectively identify patients, individualized patient therapy and follow-up, and screening of family members for afflicted individuals with diabetes mellitus, clinicians need to have a complete grasp of the epidemiology and etiology of MODY.

Declaration of patient consent

The authors certify that they have obtained all appropriate participants consent forms. In the form the participant(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in

the journal. The participants understand that their names and initials will not be published, and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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