Increasing Primary Care Provider-Initiated Geneticist Referrals in 22q11.2 Deletion Syndrome

Bavette Miller, Ph.D.
School of Healthcare Administration
Oklahoma State University Center for Health Sciences, Tulsa, Oklahoma, USA

Michael A, Kayser, D.O., FACMG
TrueFit Medical Laboratories, LLC, Tulsa, OK, USA

Melody Wang, M.S.
School of Healthcare Administration
Oklahoma State University Center for Health Sciences, Tulsa, Oklahoma, USA

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Abstract

As one of the most common multiple anomaly genetic syndromes in humans is 22q11.2, primary care providers should have the knowledge to properly assess the presenting abnormalities that may be found in a patient. Without a recognizable anomaly such as a heart disorder or palate deficiencies, individuals affected with 22q could live years without answers to critical health conditions. The lack of this diagnosis may cause additional medical conditions, even premature death. The purpose of this study was to measure primary care provider knowledge on 22q. Next, it was to examine the effect of education on the number of primary care initiated referrals when two or more known 22q anomalies are present.

Introduction

One of the most common multiple anomaly genetic syndromes in humans is 22q11.2 Deletion Syndrome (22q). Second only to Downs Syndrome.[1,2] 22q deletion is the most frequently seen microdeletion syndrome, impacting an estimated one in 2000 people[1] but could easily be as prevalent as 1:1,600.[2] There are many different symptoms, such as congenital anomalies, immunodeficiency, developmental delays, psychiatric disorders, and other abnormalities. Research identifies more than 180 clinical features in affecting almost every organ may be present, but no single anomaly occurs in all cases.[3,4] There is no documented case of 22q deletion that has all or most of the clinical features present.[2] The most common clinical feature found in patients is congenital heart disease.[5] Other predominate clinical manifestations may include facial dysmorphism, cellular immunodeficiency, palate abnormalities, and psychiatric and developmental disorders.[6,7]

This study examined the knowledge gained by primary care providers with 22q deletion education. Primary care providers in this research are defined as those physicians, medical residents, medical students, and other healthcare providers who attended the annual 2016 family practice spring conference in Tulsa, OK. Two specific questions were asked to guide this study:

Q1: Is there a significant difference in the knowledge of primary care providers once 22q education is provided?

Q2: Is there a difference in the tendency of primary care providers to refer to a geneticist when two or more known prevalent 22q anomalies are present after 22q education is introduced?

This study serves as a small step in addressing the lack of awareness of this common yet under diagnosed disorder while encouraging healthcare providers to consider all options in multi-faceted disorders.

Methods

The data for this study was collected and analyzed by researchers at the Oklahoma State University School of Healthcare Administration. Data collection occurred at the annual 2016 OSU Center for Health Sciences Family Medicine Spring Fling Continuing Medical Education (CME) Conference held at the DoubleTree in Tulsa, OK. Attendees participated in a survey aimed to assess content learned and considered useful for future patient cases regarding 22q deletion genetics.

Population: The Kaiser Family Foundation states that there are currently a little more than 425,000 active practicing primary care physicians in the United States with approximately 10% of them in Oklahoma.[8] The population for this study was 400 primary care physicians who attended the spring conference. The sample size was 107 participants who attended the 22q seminar session. The selection process was a nonprobability sample. Creswell describes this process as a less desirable approach than a random sample[9] but the goal of this study is to assess knowledge learned from the training offered at the 2016 OSU Spring CME Conference. A 22q genetics education session for CME credit was held for the sample population. Immediately following the session presentation, a survey was distributed.

Instrument: Data was collected in a self-reported eight question five-point Likert scale paper survey with an additional four open-ended questions that allowed for free-form answers. The question results reported in this study are questions one, three, and six, which can be found in table 1.

Table 1: Selected Training Assessment Questions

Question	Scale				
	None	Some			Extensive
1. To what extent was your knowledge of 22q11.2 Deletion Syndrome prior to this session?	1	2	3	4	5
3. How much did this session contribute to your general knowledge regarding 22q11.2 Deletion Syndrome?	1	2	3	4	5
6. In the future, if you treat a patient with two or more known 22q11.2. Deletion Syndrome anomalies that are highly prevalent in 22q11.2 DS patients, how likely is it that you would consider referring the patient to a geneticist?	1	2	3	4	5

The survey was disseminated prior to the beginning of the CME session and collected at the end, and results of the eight questions were averaged and compared using descriptive statistics. The difference in means of questions one and three assessed the change in knowledge from baseline to post-CME. Answers to question six are reported as a mean.

The initial inquiry was to determine a baseline of knowledge on 22q prior to the CME session. Previous education related to 22q was solicited as well as an assessment of the knowledge gained from the CME session. Based upon this self-reported data, the researchers asked if a future geneticist referral would be considered. The purpose of this study was to determine the impact of education on the increased probability of geneticist referrals. IRB approval was requested and the study was found to be exempt. The results were descriptively analyzed through Microsoft Excel.

Results

Training Contribution to General Knowledge: Out of the 400 healthcare providers registered for the two-day conference, 107 participants attended the session. Sixty-eight family medicine providers made up the largest medical specialty. Three medical students participated. All 107 participants reported that training contributed to their general knowledge regarding 22q Deletion Syndrome. One-hundred and five participants reported that they gained some to extensive knowledge from the CME session compared to their previous level of knowledge, and two individuals did not answer the question. The

difference score of 1.963 (Table 2) indicates that there was an increase of knowledge of 22q gained after the CME training.

Table 2: Knowledge of 22q Deletion Syndrome (1-5 Likert Scale)

Mean General Knowledge Prior to Training (Question 1)	2.0
Mean General Knowledge Gained Through Training (Question 3)	3.963
Difference of Mean Scores	1.963

Table 2 reveals the difference in 22q knowledge post training as nearly doubling from the training provided.

Referral Consideration: The second phase of the study was to analyze whether the increase in knowledge concerning 22q would lead to an increased probability of referral to a geneticist. On the five-point scale, the mean rate of increased probability of referrals to a geneticist for participants when patients have two or more known 22q anomalies after the training is 4.43. Ninety-seven participants suggested the training session directly contributed to the increased likelihood of a geneticist referral while three submitted the training had no effect on their decision to refer. Seven participants did not answer the question.

Discussion

Early Suggestion of 22q: Congenital heart conditions associated with newborn hypocalcemia are the most common clinical characteristics leading to a 22q diagnosis.[10] An earlier detection of 22q deletion is possible with increased education leading to healthcare provider suspicion. In recent studies, heart disease was detected in roughly 77% of individuals diagnosed with 22q deletion while 71% had palate abnormalities.[5] Caregivers are often left to manage educational and clinical treatments necessary for those with 22q. Consequently, in order to successfully prepare, one has to have a proper and prompt diagnosis. An appropriate and timely diagnosis can only be obtained with educated healthcare providers. These providers must have an understanding and suspicion of clinical presentations that could be the result of something more in-depth.[11] Our research supports the notion that CME better prepares a physician to give a more timely and accurate diagnosis of 22q when such patients are encountered.

Perceived Impact of Healthcare Providers' Experience and Education: In circumstances in which a healthcare provider has only limited information or a poor understanding of the 22q disorder, diagnosis of this condition can be an extremely stressful and negative experience for both the patient and the caregiver.[12] Most caregivers feel they become the expert due to a lack of knowledge on the part of the healthcare provider.[13] Healthcare professionals must have the education necessary to appropriately treat these patients and effectively communicate with caregivers.

Limitations: The purposeful sampling used in this study was specifically designed to target a population of healthcare providers that has some experience with the 22q microdeletion condition. Due to geographical and cultural differences, this study may not fully represent the 22q knowledge of all healthcare providers. An issue that could negatively impact the study was the fact that only those who have strong perceptions or opinions on the subject may have chosen to participate.

While this study may not reflect the understanding and learning opportunities of all healthcare providers within the larger population, the benefit the larger population is that this small study offers a template that can be replicated elsewhere.

Recommendations and Implications: This study was an introductory look at the impact of continuing medical education on 22q Deletion Syndrome for healthcare providers. The findings of this study underscore the increasing need for continuing research that assesses the current knowledge of 22q in the broader population of healthcare and educational providers. Future research should focus on the perceptions of caregivers regarding the knowledge provided to them by providers concerning a 22q deletion diagnosis. Follow-up research should be conducted further into the clinical characteristics found in those with a delayed 22q diagnosis. Additionally, further opportunities for providing healthcare provider education should be explored.

The findings of this study have the potential to create a positive experience for healthcare providers and patients when education is provided, leading to a more timely and appropriate diagnosis. Findings contribute to the existing literature that implies healthcare providers need a deeper understanding of 22q deletion and the common clinical manifestations that should lead one to suspect it as a possible diagnosis.

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