The Importance of Diabetic Screening in Individuals with Down Syndrome

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Abstract

This study was conducted to increase diabetic screenings of patients with Down Syndrome (DS) at the Geisinger Medical Center Internal Medicine Comprehensive Care Clinic in Danville, Pa., using the Global Down Syndrome Foundation Guidelines. A retrospective chart review was conducted from March 1, 2022, through Aug. 31, 2022, to assess prediabetic and diabetic screenings performed on patients with DS. An electronic medical record smart phrase prompting the physician to screen patients was implemented and a prospective chart review was conducted from Sept. 1, 2022, through March 1, 2023. Screening for prediabetes and diabetes increased by 76.5% in adults with DS by March 2023 using the Global DS Guidelines. Future steps for this project will target (include, encompass, engage) multiple Geisinger clinics that treat patients with DS to increase sample size and corroborate our results.

Introduction

Trisomy 21, also known as Down Syndrome (DS), is the most frequent chromosomal abnormality (aneuploidy). This condition is due to the presence of an extra complete or segment of chromosome 21 caused by a nondisjunction event in embryonic cells prior to conception. It is estimated each year there are about 5,000 neonates born with this chromosomal abnormality, this being about 1 in every 691 births in the United States (1). In the last 30 years the frequency of the condition has increased by 30% and seems to be rising with increased frequency of advanced maternal age pregnancies. Clinical features associated with DS include disturbance of brain development and MSK/craniofacial abnormalities such as brachycephaly, thinning of the calvarium, small rudimentary nasal bones, and epicanthal folds. These individuals are also at an increased risk of congenital heart disease, gastrointestinal abnormalities, musculoskeletal issues, spine disorders, and endocrine disorders. With age these patients are classically at risk of development of early onset dementia sharing many commonalities with Alzheimer's disease. Some studies suggest there is an up to 70% incidence of Alzheimer's dementia in patients with DS by the age of 50 (2). These patients are also at an increased risk of developing hyperglycemia through the pancreatic beta-cell dysfunctional process seen with Type 1 diabetes (T1D) and insulin resistance seen with Type 2 diabetes (T2D).

There are few areas of research that illustrate the impact and pathophysiology of diabetes in individuals with DS. Several animal models studies have demonstrated increased inflammatory markers of autoimmunity in this population. This and the increased pancreatic B-cell dysfunction found in this population suggests an impaired auto-immune metabolic/inflammatory process of basal insulin deficiency. Insulin resistance in DS has been associated with a gene found in the extra chromosome, RCAN1, regulating hepatic glucose production and expression of gluconeogenic genes (2).

Individuals with DS have up to four times the incidence of diabetes as compared to the general population, with younger age of onset and a higher incidence of obesity. The effects of hyperglycemia and lack of insulin can have numerous deleterious outcomes. These outcomes may include retinopathy, neuropathy, nephropathy, and increased risk of vascular ischemic events. Recent studies have also investigated the role of insulin in neuronal activity and synaptic plasticity leading to a higher risk of developing age-related cognitive decline and neurodegenerative diseases. These studies suggest cognitive decline seen with DS is accelerated by insulin deficiency/resistance (2).

These risks pose a great need for preventive care in people with DS, including earlier screening and detection of diabetes, which can greatly influence their health outcomes (3). The current guideline for diabetic screening is made by the Global DS Foundation. These guidelines established in 2021 suggest BMI monitoring for individuals with DS should be done annually, and these results will dictate the frequency of screening in the population, either using HbA1c or a fasting blood glucose test. In addition to BMI, the patient age will be used to assess when to begin screening (4).

The need for screening of prediabetes and diabetes in individuals with DS is proving to be a field requiring further investigation. Research has shown that there are molecular mechanisms linking DS to both Type 1 and Type 2 diabetes (5). The prevalence of these conditions and prediabetes among people with DS is 3–5% for ages 16–30 and 5.5% for ages greater than 30. The effectiveness of the current screening protocol is poor on the national level. Missed diagnoses of diabetes or prediabetes in patients can increase the risk for retinopathies, neuropathies, nephropathies, and cardiovascular damage. While this guideline and others exist, there is more work that must be done to gain strong evidence that can help clinicians in their practice of care for the population of individuals with DS. Organizations including the American Academy of Family Physicians (AAFP) have published articles emphasizing the need for screening recommendations for diabetes in people with DS.

Currently, there is no protocol or emphasis regarding the screening for prediabetes and diabetes in patients with DS at Geisinger. Development and implementation of a tool to aid Geisinger physicians in the diagnosis of prediabetes and diabetes in these individuals will not only enhance the care we are providing our patients, but also minimize the disease burden within these individuals that may be more susceptible to T1D or T2D. Our aim was to improve the diagnosis of prediabetes and diabetes in adults with DS at Geisinger Medical Center (GMC) Internal Medicine Comprehensive Care Clinic by 50% by March 2023 by using the Global DS Guidelines.

Methods

A project aim was developed and defined: "To improve the diagnosis of prediabetes and diabetes in adults with DS at the GMC Internal Medicine Comprehensive Care Clinic (by 50%) by March 2023 utilizing the Global Medical Care Guidelines for Adults with DS." An electronic health record SMART phrase, "Diabetes: asymptomatic adults screen at 30 yo and if obesity, screen at 21 yo," was then implemented into each comprehensive care visit in patients with DS of 18 years or older starting on Sept. 1, 2022. A retrospective chart review which focused on screenings before utilization of the more recent Global DS Guidelines and SMART phrase was performed between March 1, 2022, and August 31, 2022, recording age, gender, hemoglobin A1c, and BMI. This was subsequently followed by a prospective review performed between Sept. 1, 2022, and March 1, 2023. Results including patients who were screened for prediabetes and diabetes pre- and post-intervention, as well as identification of prediabetic or diabetic status according to HbA1C was recorded (Normal Range <5.7%, Prediabetic 5.7% - 6.4%, Diabetic 6.5%+). The study was piloted at the Department of General Internal Medicine at GMC in Danville, Pa.

Results and Discussion

Prior to implementation of the Epic SMART phrase intervention, only 3 of 17 patients who qualified for screening (17.6%) were properly screened for diabetes and prediabetes (Figure 1). After intervention was implemented into each comprehensive care visit, 16 out of 17 qualifying patients (94.1%) were screened (Figure 2). Therefore, screening for prediabetes and diabetes increased by 76.5% in adults with DS by March 2023 using the Global DS Foundation Guidelines. The 3 patients (of the total possible 17) who were screened during the retrospective chart review prior to intervention were determined to be prediabetic (Figure 3). However, of the 14 remaining individuals who did not receive diabetes screening during their comprehensive care visit prior to intervention (March 1, 2022 – Aug. 31, 2022), 100% of these patients who had appointments during the prospective chart review period (Sept. 1, 2022 – March 31, 2023) were properly screened (Figure 4). 21.4% of these individuals who were not screened during the pre-intervention period but were properly screened post-intervention were found to be prediabetic. Of the 16 patients who were screened during the prospective chart review, 5 were found to be prediabetic and 11 were determined to have HbA1C levels within the normal range.

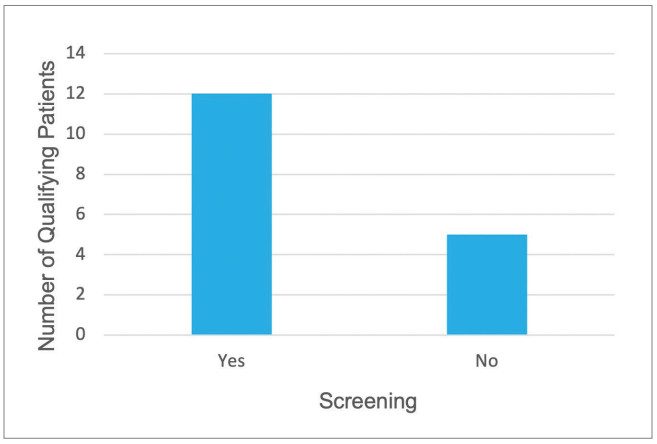

Figure 1. Bar graph representing HbA1c screening performed in qualifying patients with DS at GMC Internal Medicine Comprehensive Care Clinic from March 1, 2022, to Aug. 31, 2022, prior to intervention. 17.6% of qualifying patients were screened.

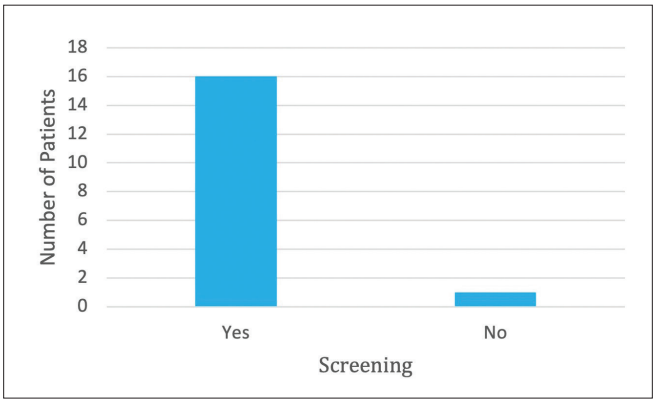

Figure 2. Bar graph representing HbA1C screening performed in qualifying patients with DS at GMC Internal Medicine Comprehensive Care Clinic from Sept. 1, 2022, to March 1, 2023, post intervention. 94.1% of qualifying patients were screened.

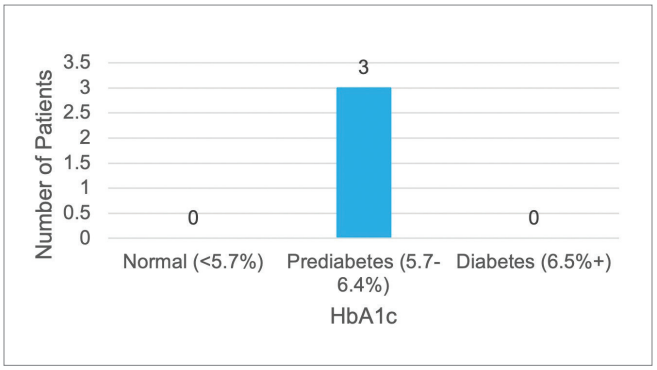

Figure 3. Bar graph representing prediabetes and diabetes status of patients with DS at GMC Internal Medicine Comprehensive Care Clinic who were screened from March 1, 2022 to Aug. 31, 2022. Prior to intervention, only 3 members of a possible 17 in this patient population were screened.

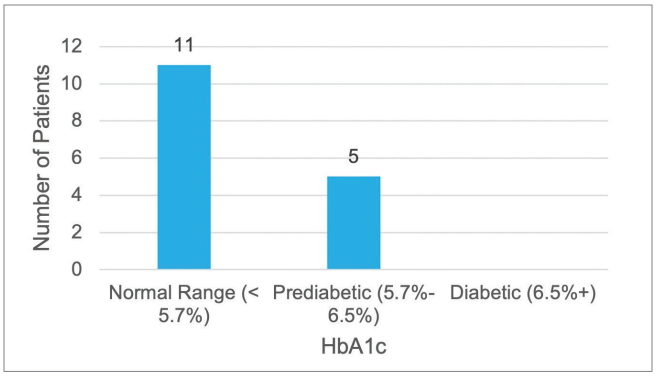

Figure 4. Bar graph representing prediabetes and diabetes status of patients with DS at GMC Internal Medicine Comprehensive Care Clinic who were screened from Sept. 1, 2022 to March 31, 2022. Post intervention, 16 members of a possible 17 in this patient population were screened.

Conclusion

While we believe this study can provide insight into appropriate management plans for metabolic diseases such as diabetes mellitus in patients with DS, there were some important limitations to this study that influence the quality of the interpretation of the data, such as the small sample size due to piloting this study at only one clinic within the larger Geisinger system. In the future, it will be important to institute this intervention in clinics within the system to increase the patient population and ultimately the number of patients screened. The study can further be expanded to more clinics across multiple systems in order to increase the confidence in our hypothesis.

The increased detection for prediabetes in patients with a DS diagnosis will leave room for early detection of metabolic disease which will improve both quality of life for these patients and overall cost of care associated with complications that can be endured secondary to late detection and treatment.

Disclosures

The authors have nothing to disclose.

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