Identification of Atypical Pediatric Diabetes Using Electronic Medical Records (EMR)

Marcela, Astudillo MD; Mustafa Tosur MD; Alejandro F. Siller MD; Ashok Balasubramanyam MD; Maria J. Redondo MD.
Department of Pediatrics, Diabetes and Endocrinology, Baylor College of Medicine, Texas Children's Hospital, Houston, TX 77030 USA

Introduction

- Atypical forms of diabetes that challenge the diagnoses of "type 1 diabetes (T1D)" or "type 2 diabetes (T2D)" are emerging.
- Molecular diagnostic capabilities and deeper phenotyping techniques have confirmed a heterogeneous spectrum of diabetes based on multiple etiologies.[1]
- It is important to classify these atypical forms accurately because the diagnosis of specific subtypes carries implications for disease progression, prognosis, optimal treatment and counselling of family members regarding heritability of the disease.[2–4]

Objectives

- To identify cases of rare or atypical pediatric diabetes patients who may be candidates for studies to understand their etiology and pathogenesis

Methods

- We tested two strategies in a large pediatric hospital in Southwestern USA.
- **STRATEGY 1:** We designed a questionnaire that would allow a health-care provider to rule out typical diabetes types by manual EMR review. This questionnaire was first applied to the EMR of 50 youth (0-21 years old [y/o]) consecutively seen in the diabetes outpatient clinic, then modified to address problems identified, and tested in a second iteration, and on 50 additional cases (n=100).
- **STRATEGY 2:** In collaboration with local EMR analysts, we built three queries to generate periodic, automated reports of possible pediatric atypical diabetes cases: unknown type, type 2 diabetes diagnosed <10 y/o, and autoantibody-negative type 1 diabetes. Diabetes type (i.e. unknown, T1D or T2D) was determined by the treating pediatric endocrinologist, through a customized flowchart embedded in the EMR.

Results

- **STRATEGY 1:** We found 6 cases (6%) of atypical diabetes (mean age at diagnosis 11 y/o [SD 2.64], 16.6% male, 33% non-Hispanic White [NHW] and 66.6% Hispanic [Hisp]).
- **STRATEGY 2:** Unknown diabetes type: 68 cases (1%, out of 6676 total diabetes cases), with mean age at diagnosis 12.6 y/o (SD 3.3), 32.8% male, 23.8% NHW, 47.6% Hisp, 25.4% African-American (AA), 3.2% other. Out of 1142 children with T2D, 64 (6.6%) cases were diagnosed <10 y/o, with mean age at diagnosis 8.6 y/o (SD 1.6), 20.3% male, 4.7% NHW, 65.6% Hisp, 28.1% AA, 1.6% other. Out of 680 cases of new onset T1D, 38 (5.6%) were negative for islet autoantibodies (IAA, GAD, IA-2), with mean age at diagnosis 11.3 y/o (SD3.8); 57.9% male, 50% NHW, 19.4% Hisp, 22.3% AA, 8.3% other. (Table 1)

Table 1.- Characteristic of patients identified with possible atypical diabetes using EMR

	Strategy 1. Questionnaire	Strategy 2. EMR Queries		
	Atypical Diabetes (n=6/100)	**Unknown type** (n=68/6676)	**T2D < 10 y/o** (n=64/1142)	**Autoantibody Negative T1D** (n=38/680)
Age at diagnosis (y/o)	11 ±2.64	12.6 ±3.3	8.6 ±1.6	11.3 ±3.8
Gender				
Male	17%	33%	20%	58%
Female	83%	67%	80%	42%
Race/ethnicity				
Non-Hispanic White	33%	24%	5%	50%
Hispanic	67%	48%	66%	19%
African American	-	25%	28%	22%
Other	-	3%	1%	8%

- We next conducted a manual review of the 68 cases with unknown diabetes type to further identify their type and characteristics, and thus, estimate the yield of this approach (Table 2).

Table 2. Characteristic of patients with "Unknown diabetes"

Cluster	N (%)
Suspected monogenic, negative MODY panel	4 (6%)
Suspected monogenic, VUS	1 (1%)
Suspected monogenic, MODY panel-Not Done	23 (34%)
T1D vs T2D vs monogenic-MODY panel-Not Done	1 (1%)
Syndromic	2 (3%)
T1D vs T2D	3 (4%)
Gestational diabetes	11 (16%)
Likely T2D	13 (19%)
Under study (missing tests)	3 (4%)
Known MODY diagnosis	2 (3%)
Drug-induced or post-transplant vs T2D	4 (6%)
Stress induced hyperglycemia	1 (1%)

Conclusion

- We designed and tested two strategies that use EMR to identify atypical pediatric diabetes cases for further studies that may shed light on the heterogeneity of pediatric diabetes, possibly opening new opportunities for classification, diagnosis, treatment and prevention.

Bibliography

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