

Background

Kovler Diabetes

Center

- Monogenic diabetes is a group of diabetes subtypes caused by a single genetic abnormality.
- These forms represent $\sim 5\%$ of diabetes diagnosed < 35 years of age, yet they are often misdiagnosed and treated inappropriately.
- The UChicago Monogenic Diabetes Registry is a longitudinal study that collects medical and family history from those with known or suspected monogenic diabetes.

Specific Aims

We aim to describe the demographic and clinical characteristics of participants with known or suspected monogenic diabetes in the UChicago Monogenic Diabetes Registry, as well as their Registry referral source.

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The University of Chicago Monogenic Diabetes Registry: Precision Medicine in Diabetes

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- 4057 participants were enrolled and 1203 had a known genetic cause as of 6/29/2022.
- The majority identified as Female (56%), Non-Hispanic (86%), and White (85%).
- The majority were referred to our studies through a physician or diabetes provider (55%), with the majority of provider referrals from the past 12 months representing specialists such as endocrinologists and diabetes care and education specialists (83%)

Table 1: Results		
Total enrolled:	4057	
Known Cause-n (%) GCK, HNF1a, HNF4a, KCNJ11, ABCC8 Other	1203 986 (82%) 217 (18%)	
Provider Referrals Past 12 months-n (%) Generalist Specialist Did not specify	47 4 (8.5%) 39 (83%) 4 (8.5%)	
Diagnoses participants with known monogenic diabetes were told- n(%) Prediabetes Type 1 diabetes Type 2 diabetes	959 responses 144 (15%) 224 (23%) 260 (27%)	

Results

- Of those with a known genetic cause, previous misdiagnoses included prediabetes (15%), type 1 diabetes (23%), or type 2 diabetes (27%)
- Of those with a known genetic cause, 82% have a type that is typically amenable to precision treatment, such as oral therapies or no treatment.

Table 2: Demographics		
Total responses-n (%)	4020	
Male	1751 (44%)	
Female	2269 (56%)	
Total responses-n (%)	1533	
Hispanic	194 (13%)	
Non-Hispanic	1316 (86%)	
Prefer not to answer	9 (0.6%)	
Unknown	14 (0.9%)	
Total responses-n (%)	1550	
American Indian or Alaska Native	32 (2%)	
Asian	127 (8%)	
Black or African American	140 (9%)	
Native Hawaiian or other Pacific Islander	1 (0.1%)	
White	1312 (85%)	
Unknown	13 (0.8%)	
Prefer not to say	5 (0.3%)	

Methods

- Enrollment begins by completing a secure web-based registration form: monogenicdiabetes.uchicago.edu (UChicago IRB #6858 and #15617B).
- After consent, participants may:
 - provide a saliva sample via mail for researchbased genetic testing
 - complete longitudinal surveys
 - participate in gene-specific discussion groups
- Data were collected and managed using Research Electronic Data Capture (REDCap) hosted at UChicago (1,2).

Conclusion

Diabetes Care and Education Specialists are critically important in recognizing patients with known or suspected monogenic diabetes to ensure that patients receive an accurate diagnosis and precision treatment.

Fig. 2 Individuals with a Known Cause

