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Surveillance for the Rare Condition of Sickle Cell Disease in Wisconsin

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Abstract

Background: Despite universal newborn screening, there is no comprehensive surveillance system to understand the sickle cell disease population in Wisconsin.

Methods: We initiated the development of a sickle cell disease surveillance system by linking newborn screening data and electronic health records from 2 large tertiary health care institutions in Wisconsin: Children's Wisconsin and Froedtert Hospital.

Results: There were 1478 individuals within the 3 data sources. One hundred thirty-two (82%) of 159 identified by newborn screening from 2013 through 2019 received care at Children's Wisconsin. The majority of individuals with sickle cell disease at Children's Wisconsin and Froedtert Hospital resided in Milwaukee County.

Discussion: The new surveillance program will increase our understanding of the sickle cell disease population in Wisconsin and help improve quality of care and health outcomes.

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Appendix: Available at wmjonline.org

BACKGROUND

Sickle cell disease (SCD) is a genetic condition caused by a pathogenic gene variant in the beta-globin chain of hemoglobin. In the United States, SCD predominantly occurs among Black individuals. Despite universal newborn screening,[1] there is no national-level surveillance system to understand the care and health outcomes for the SCD population. Children and adults with SCD experience significant morbidity and have impaired quality of life.[2] These individuals face health care disparities at numerous levels, including access to specialty care and recommended treatment.[3] High quality care, assessed based on quality-of-care metrics (Table 1), is needed to improve SCD outcomes. However, due to lack of surveillance data, there is limited knowledge to inform actions needed at a systems level to effectively improve outcomes for the SCD population.

Existing Knowledge Regarding SCD in Wisconsin

Published newborn screening data show 19 to 32 infants are diagnosed with SCD each year in Wisconsin, with an overall birth incidence of 0.319 per 1000 births.[1] Previous studies from Wisconsin have assessed health care utilization for individuals with SCD. Research using Wisconsin Medicaid data show emergency department (ED) reliance (number of ED visits divided by the number of ED and outpatient visits) varies by age for those with SCD.[4,5] Data show less than 20% of children with SCD had excessive ED reliance (defined as > 0.33), whereas 40% to 50% of those in the age-group transitioning from a pediatric to adult facility (those who turned 19 years during the study) had excessive ED reliance.

Another study that used Medicaid data (years 2004–2007) looked at infection prevention strategies in individuals with SCD, including receipt of penicillin prophylaxis and pneumococcal and influenza immunizations.[5] The study showed a small proportion (18%) of eligible children received the standard of care for penicillin prophylaxis, and there was low adherence to recommended annual influenza vaccination (21%).[6] Encouragingly, 77% of eligible children in the study received pneumococcal (PCV7) vaccination. Also, with targeted quality improvement efforts at Children's Wisconsin, we were able to achieve and sustain >70% annual transcranial doppler screening rates for children with sickle cell anemia.[7] Notably, these studies are all limited to data from either 1 insurance type (Medicaid) or single facility (Children's Wisconsin). As a result, a lack of understanding of the overall epidemiology and health outcomes for the entire SCD population in Wisconsin still exists.

Surveillance for SCD in Wisconsin

The Centers for Disease Control and Prevention's (CDC) Sickle Cell Data Collection (SCDC) program is a population-based, longitudinal surveillance system. The goal of SCDC is to study long-term trends in diagnosis, treatment, and health care access for people with SCD living in the United States. In 2021, eleven states including Wisconsin, were competitively selected to participate in the SCDC program.[8]

The SCDC program in Wisconsin aims to include all individuals living with SCD in the state. We will link and aggregate data from various existing institutional and state-level

data sources to establish a SCD surveillance system for Wisconsin. We have established a multidisciplinary team that includes newborn screening program members, health care providers, policymakers, bioinformaticians, researchers, and individuals with SCD to guide the program.

In this brief report, we describe preliminary findings from the linkage of data from the newborn screening program and electronic health records (EHR) from 2 large tertiary-care facilities with SCD clinics, Children's Wisconsin (CW) for children and Froedtert Hospital (FH) for adults. We also discuss how Wisconsin's participation in SCDC could improve care and outcomes for those with SCD in the state.

METHODS

We used a combination of deterministic and probabilistic linkages to deduplicate and link records based on patient name, sex, and date of birth. The string of names were compared and matched using Levenshtein's edit distance.[9] Sex and date of birth were compared as exact matches. A varying range of weights for the specific identifiers were used to identify overlapping patients, along with chart review as needed.

A 3-tiered SCD case definition was applied where (1) individuals identified with SCD through the Wisconsin newborn screening program were considered confirmed cases, (2) individuals who did not meet the confirmed case definition but had 3 or more SCD-associated encounters at CW and/or FH were considered probable cases, and (3) individuals who did not meet the confirmed case definition but had 1 or 2 SCD-associated visits at CW and/or FH were considered possible cases. SCD-associated encounters are those with a SCD International Classification of Disease [ICD] code present at admission, discharge, or final diagnosis. (See Appendix for SCD ICD codes.)

In this report, we describe the overlap between data from the newborn screening program and the EHR at CW and FH for years 2013–2019. We also describe demographic characteristics (age, sex, race, ethnicity, known vital status) for individuals who met either the confirmed or probable case definition and received care at CW and/or FH. Age was calculated as of December 31, 2019, or on the date of death if the individual was deceased. Vital status was determined from the EHR. We used the Federal Office of Rural Health Policy eligible file to determine the rural/nonrural category. Any ZIP code where more than 50% of its population resides in a nonmetro county and/or a rural census tract was classified as rural.

RESULTS

A total of 1478 individuals met 1 of the tiers of the SCD case definition (Figure). There were 159 infants diagnosed by the Wisconsin newborn screening program during 2013–2019 who were considered confirmed cases; 83% (N=132) of these had at least 1 SCD-associated encounter at CW during 2013–2019. There were 793 and 797 individuals at CW and FH, respectively, who had at least 1 SCD-associated visit. Upon linking and deduplicating EHR data sources with newborn screening data, there were 1451 unique individuals across the 2 sites (1450 had an SCD-associated visit at CW/FH plus 1 who did not have a SCD-

associated visit at CW but received care at the facility). Of these, 68% (N = 993) met the confirmed or probable case definition, and 31% (N = 458) met the possible case definition. Overall, there were 1020 confirmed or probable (993 + 27 from newborn screening but not in EHR) cases in the program. The overlap between data sources for cases by age group is shown in Table 2.

Fifty-three percent of confirmed or probable cases at CW and/or FH were female (N = 525), and the median age was 22 (range: 0.10–87.8) years. Ninety-five percent (N = 942) were Black, and 98% (N = 971) were neither Hispanic nor Latino. Fifty-five (6%) patients died during 2013–2019. Of those in Wisconsin and with available ZIP code data, 83% resided in Milwaukee County, and 99% were classified as living in a nonrural area.

DISCUSSION

Our preliminary linkage of 3 data sources shows the majority of children diagnosed with SCD through Wisconsin newborn screening (82%) had at least 1 visit at CW. Combined data from CW and FH show a large proportion of patients receiving care at these institutions reside in 1 county (Milwaukee County). There is ongoing work to include individuals with SCD who receive care within Wisconsin outside of these health care systems by incorporating other statewide data sources, such as administrative claims data from Medicaid and the Wisconsin Health Information Organization (all payers claims). This comprehensive assessment intends to identify to the best of our ability the majority of individuals living with SCD in Wisconsin. Further work on this surveillance program will facilitate systematic data collection and provide a platform to engage various stakeholders to understand the needs of the SCD population. Specific examples below discuss how the program can be leveraged to improve the lives of those with SCD living in Wisconsin.

Understanding demographics of the SCD population in Wisconsin:

The knowledge of demographics (age, sex, race/ethnicity, geographic distribution) of individuals with SCD is essential to understanding this population’s needs. This program will help determine if certain subgroups of the SCD population reside in areas with few or no hospitals, EDs, or subspecialty care so efforts can ensure equitable access to care for all individuals with SCD.

Quality of care delivered to individuals with SCD:

Our program will help determine if individuals with SCD in Wisconsin are receiving recommended care to manage their disease. Sharing data on quality-of-care metrics (Table 1) with clinicians and third-party payers will help identify and fill the gaps in care.

Health care service utilization and health-related outcomes for individuals with SCD:

Individuals with SCD experience severe acute pain events that often require ED visits and hospitalizations. They are also at risk of chronic end organ damage. This surveillance program can help identify the frequency of acute care utilization for pain using prior validated algorithms[10] and determine the prevalence of complications so an informed care approach can be adopted.

Communication platform for individuals with SCD:

We will develop communication plans to share the findings of our program with the SCD community in Wisconsin. This will help establish a platform to share research findings and SCD resources with those who might otherwise be unaware of the complexities of the disease.

CONCLUSIONS

Our results show the potential for learning more about the SCD population by combining data sources. Future work incorporating additional SCDC data sources is required to fully assess the prevalence of SCD throughout the state and inform policy and practice. This program will also serve as a model to understand other rare chronic conditions.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

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REFERENCES

1. Therrell BL Jr, Lloyd-Puryear MA, Eckman JR, Mann MY. Newborn screening for sickle cell diseases in the United States: a review of data spanning 2 decades. Semin Perinatol 2015;39(3):238–251. doi:10.1053/j.semperi.2015.03.008 [PubMed: 25979783]
2. Brandow AM, Brousseau DC, Panepinto JA. Postdischarge pain, functional limitations and impact on caregivers of children with sickle cell disease treated for painful events. Br J Haematol 2009;144(5):782–788. doi:10.1111/j.1365-2141.2008.07512.x [PubMed: 19055663]
3. Lee L, Smith-Whitley K, Banks S, Puckrein G. reducing health care disparities in sickle cell disease: a review. Public Health Rep 2019;134(6):599–607. doi:10.1177/0033354919881438 [PubMed: 31600481]
4. Hemker BG, Brousseau DC, Yan K, Hoffmann RG, Panepinto JA. When children with sickle-cell disease become adults: lack of outpatient care leads to increased use of the emergency department. Am J Hematol 2011;86(10):863–865. doi:10.1002/ajh.22106 [PubMed: 21815184]
5. Singh A, Yan K, Brandow AM, Panepinto JA. Longitudinal trend in emergency department reliance for pain among sickle cell disease patients in Wisconsin. J Pediatr Hematol Oncol 2019;41(7):e438–e442. doi:10.1097/MPH.0000000000001533 [PubMed: 31219908]
6. Beverung LM, Brousseau D, Hoffmann RG, Yan K, Panepinto JA. Ambulatory quality indicators to prevent infection in sickle cell disease. Am J Hematol 2014;89(3):256–260. doi:10.1002/ajh.23627 [PubMed: 24779032]

7. Singh A, Danda V, Van Swol L, Scott JP, Brandow AM, Panepinto JA. Recommendation to reality: closing the transcranial Doppler screening gap for children with sickle cell anemia. Pediatr Blood Cancer 2021;68(2):e28831. doi:10.1002/pbc.28831 [PubMed: 33283465]

8. Sickle Cell Data Collection (SCDC) Program. Centers for Disease Control and Prevention Updated April 2, 2021. Accessed June 3, 2021. https://www.cdc.gov/ncbddd/hemoglobinopathies/scdc.html

9. SAS Viya Programming Documentation Accessed December 20, 2022. https://documentation.sas.com/doc/en/pgmsascdc/v_034/ds2ref/n13dc4wnakmwj3n1uop2hkilww2d.htm

10. Singh A, Mora J, Panepinto JA. Identification of patients with hemoglobin SS/Sβ0 thalassemia disease and pain crises within electronic health records. Blood Adv 2018;2(11):1172–1179. doi:10.1182/bloodadvances.2018017541 [PubMed: 29792312]

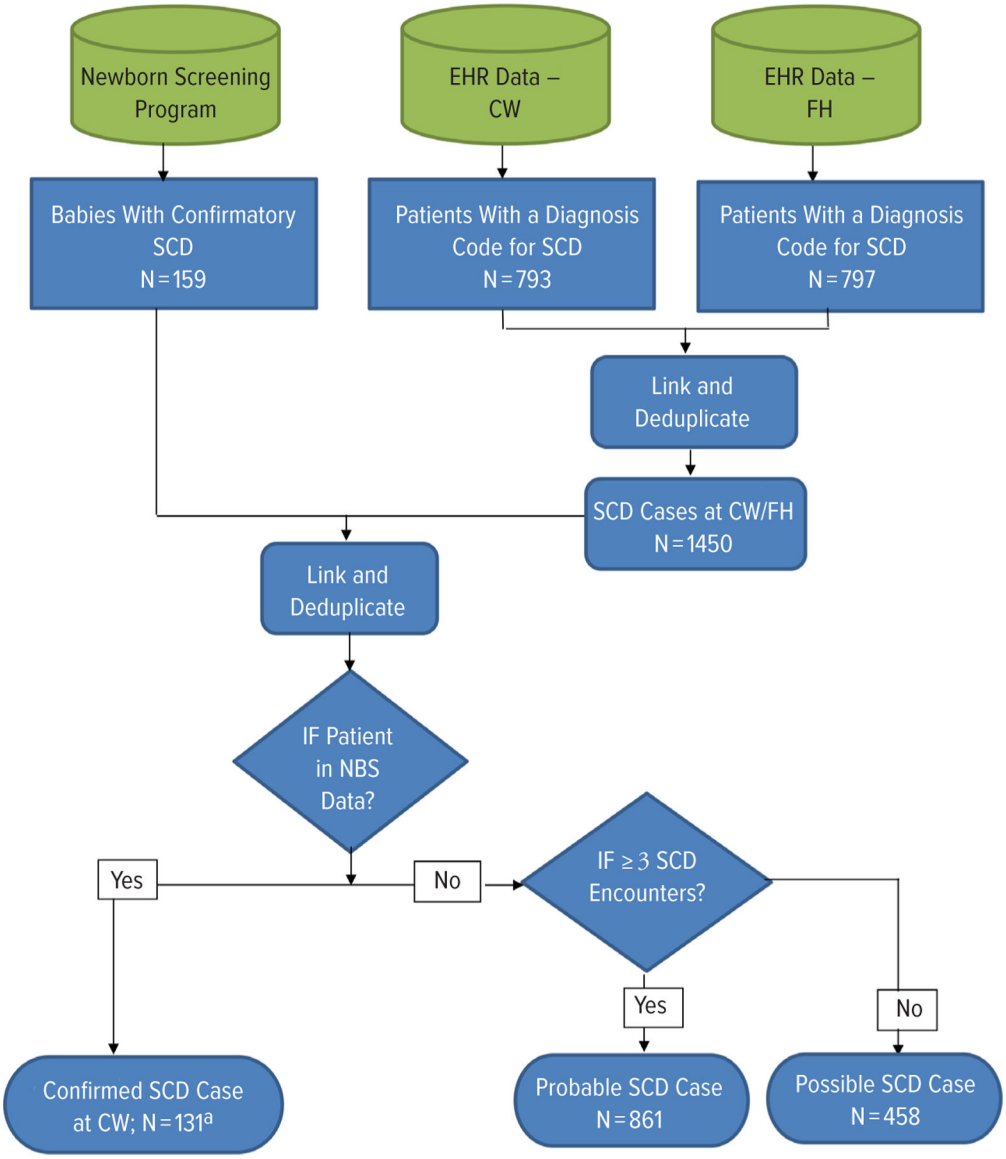

Figure.

Flow Chart to Describe the Identified Sickle Cell Disease Cases in This Report Using Newborn Screening and Electronic Health Record Data for Years 2013–2019

Abbreviations: EHR, electronic health record; NBS, newborn screening; SCD, sickle cell disease; CW, Children's Wisconsin; FH, Froedtert Hospital.

[a]There was 1 more child diagnosed by NBS as having SCD in 2019 who received care at CW but did not have a SCD-associated visit during the study period. Therefore, total confirmed SCD cases at CW = 132.

Table 1.

Examples of Recommendations in Guidelines Issued by American Society of Hematology and National Heart, Lung, and Blood Institute

Recommendation	Eligible Sickle Cell Disease Population
Annual transcranial doppler ultrasound	2–16 years of age with sickle cell anemia (HbSS/HbS beta^0 thalassemia)
Brain magnetic resonance imaging (at least 1 time)	Early-school-age children with sickle cell anemia
Antibiotic prophylaxis	< 5 years of age with sickle cell anemia
23-valent polysaccharide pneumococcal vaccine	> 2 years of age with sickle cell disease

Table 2.

Overlap of Probable or Confirmed Cases Between EHR and NBS sources in the Wisconsin Sickle Cell Data Collection Program

Age Group (in Years)	N	In NBS Only	In CW Only	In FH Only	In NBS and CW	In CW and FH
< 10 years	255	27	92	0	132	4
10–19 years	232	0	179	1	0	52
20–29 years	188	0	8	25	0	155
30–39 years	153	0	0	44	0	109
40–49 years	90	0	0	54	0	36
50–59 years	64	0	0	42	0	22
60+ years	36	0	0	29	0	7
Total	1020[a]	27	279	195	132	387[a]

Abbreviations: EHR, electronic health record; NBS, newborn screening; CW, Children's Wisconsin; FH, Froedtert Hospital.

[a]2 subjects from EHR had age missing.