Healthcare Resource Utilization Among Pediatric and Adult Patients with Glycogen Storage Disease Type Ia (GSDIa): A Case Series from the GSDIa Odyssey Natural History Study

Manpreet K. Sidhu,[1] Erru Yang,[1] Omid Khazaie Japalaghi,[1] Daniel Garcia[1]

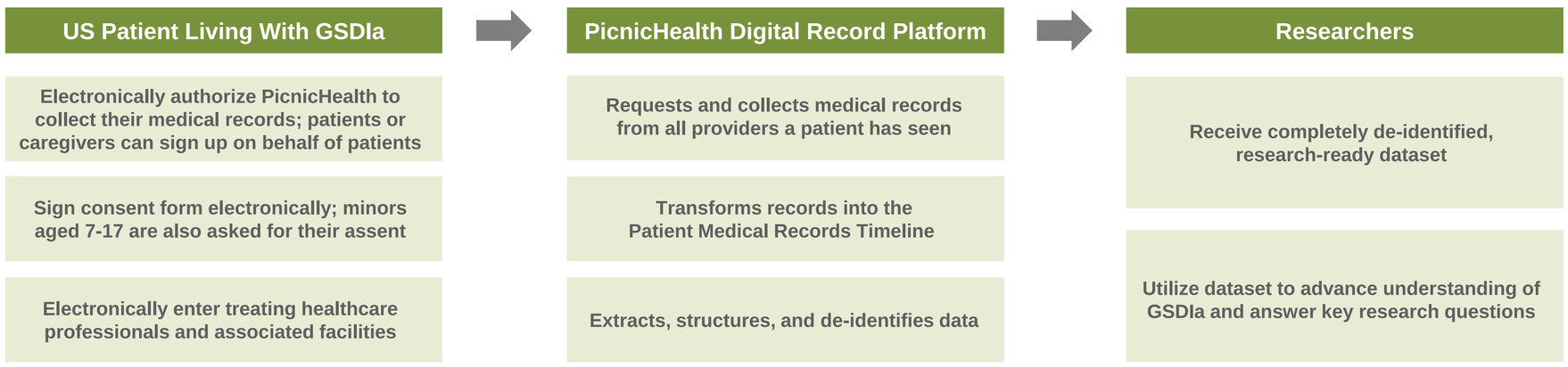
[1]Ultragenyx Pharmaceutical Inc., Novato, CA, USA

INTRODUCTION & OBJECTIVE

- GSDIa is a rare, progressive, inherited metabolic disorder of carbohydrate metabolism caused by a deficiency of glucose 6-phosphatase, which results in potentially life-threatening hypoglycemia, metabolic abnormalities, and complications often requiring hospitalization[1,2]
- Patients with GSDIa experience a substantial burden associated with the condition and its treatment, which requires exogenous glucose replacement, strict dietary adherence, and frequent monitoring[1,2]
- Real-world data (RWD) regarding GSDIa management and patient outcomes are limited
- The GSDIa Odyssey study collected RWD to better understand the natural history of GSDIa and identify areas to improve patient care

METHODS

- Non-interventional cohort study that collected retrospective data with the PicnicHealth digital record platform using a novel human-in-the-loop machine learning system to structure US medical records (clinical notes, medications, laboratory/imaging results, diagnostic reports)
- Healthcare resource use was analyzed in children (5–13 years old) and adults ≥ 18 years old
- The study was HIPAA-compliant and IRB-approved; all patients or caregivers of patients <18 years provided written informed consent, after which medical records were requested, transformed to a timeline, and data were anonymized



RESULTS

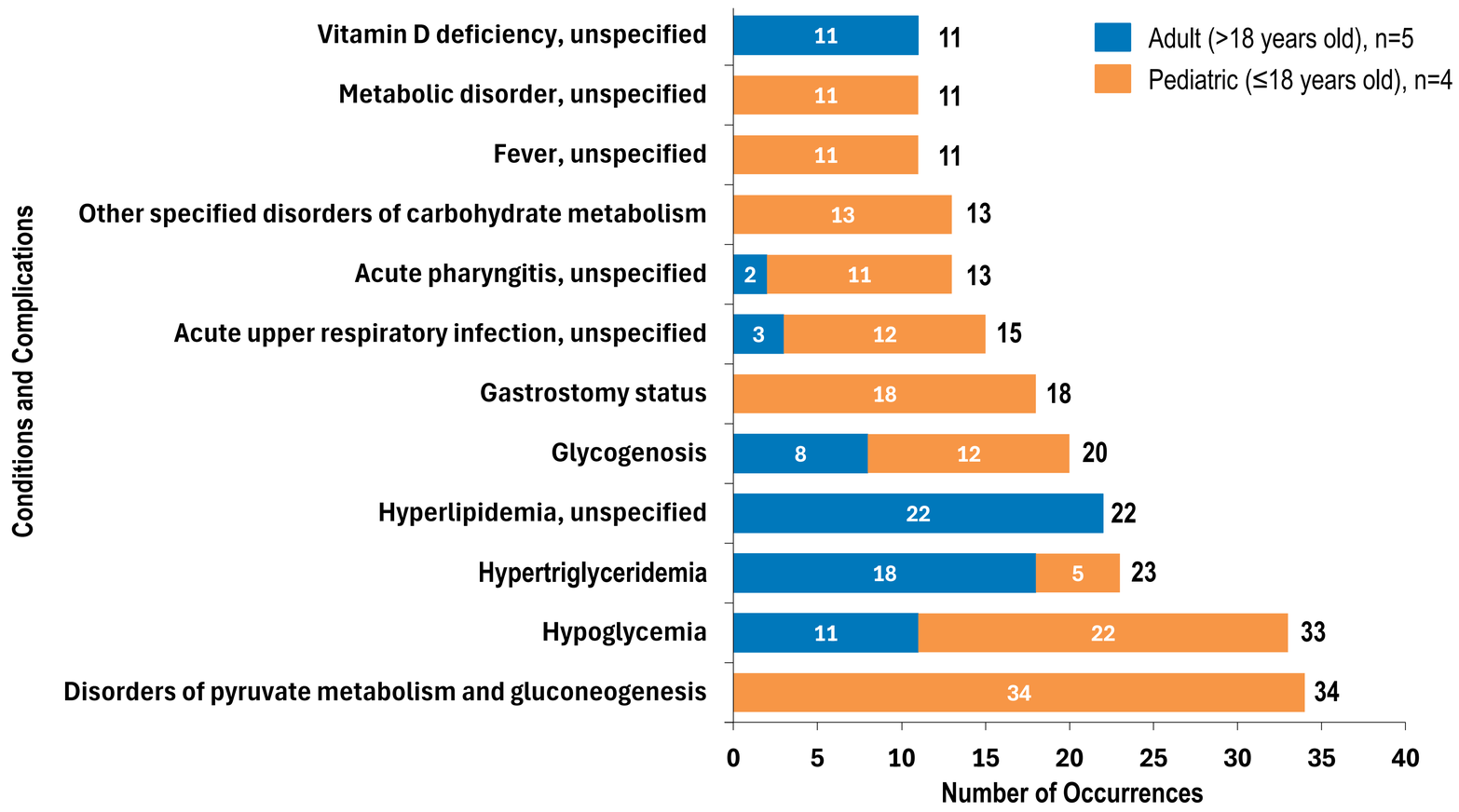
Table 1. Nine patients with GSDIa were enrolled from December 2020 through February 2023.

Baseline Characteristic	Total (N=9)[a]
Sex	
Female	6 (67)
Male	3 (33)
Race	
Unknown	1 (11)
White	8 (89)
Ethnicity	
Hispanic or Latino	1 (11)
Not Hispanic or Latino	7 (78)
Unknown	1 (11)
Age	20 (8, 28)
Pediatric (range)	8 (5–13)
Adult (range)	32 (19–48)
Total Years of Clinical Documents	12 (8, 16)
Total Years of Visits	14 (9, 20)
Number of Providers	22 (17, 36)
Number of Care Sites	7 (4, 10)
Number of Hospitalizations	6 (4, 16)
Total Hospital Days	31 (10, 40)

[a]n (%) or median (interquartile range) unless otherwise noted.

- Mean age at enrollment was 20 years (range, 5–48), with 4 children aged 5–13 years old and 5 adults >18 years old
- The majority of patients were female (67%) and White (89%)
- Median years of retrospective data was 12 years, with 22 providers and 7 care sites per patient

Figure 1. Most common conditions and complications in children and adults with GSDIa.



- The most common conditions and complications in children that resulted in healthcare visits included hypoglycemia, gastrostomy status, and hepatic glycogenosis (hepatomegaly)
- The most common conditions and complications in adults that resulted in healthcare visits were hypoglycemia, isolated hypertriglyceridemia, and hyperlipidemia (unspecified)

Table 2. Healthcare resource use among pediatric and adult patients with GSDIa.

Healthcare Resource Utilization[a,b]	Pediatric (5–13 years old) (n=4)	Adult (>18 years) (n=5)
Hospitalizations[c]		
Total hospital episodes (range)	51 (3–24)	29 (3–21)[d]
Median days in hospital (range)	2 (0–33)	1 (0–8)
Total days of inpatient visit (stays)	98 (51)	37 (29)
Total days of inpatient visit (non-ER) (stays)	42 (36)	27 (25)
Median days of inpatient (non-ER) visit (range)	1 (0–6)	1 (0–8)
Annualized inpatient visits	4	2
Emergency Room Visits		
Total ER visits (range)	22 (1–9)[e]	8 (1–5)
ER visit that resulted in an inpatient stay (range)	15 (1–7)	2 (1–3)
Total inpatient days (range)	56 (1–33)	10 (1–5)
Annualized ER visits with an inpatient stay	2	1
Outpatient Visits		
Total outpatient visits (range)	205 (26–68)	266 (11–94)
Annualized outpatient visits	21	22
Labs Visits		
Total lab visits (range)	38 (7–14)	122 (17–35)
Annualized lab visits	4	8

[a]Total refers to the total episodes, days, or visits for either pediatric (n=4) or adult (n=5) patients; range refers to the range per episode; [b]Median days in hospital and inpatient (non-emergency room [ER]) visit are per episode and the episodes are linked to acute events. [c]Includes inpatient and ER visits that resulted in a hospital stay. [d]Adult patients likely missing hospital episode data. [e]Two pediatric patients only visited the ER without an inpatient stay (7 visits).

- All patients had an inpatient visit, and an emergency room visit that resulted in an inpatient stay
- Pediatric patients with GSDIa had 1.8 times more inpatient stays and 2.8 times more emergency room visits than did adult patients with GSDIa
- Adult patients with GSDIa had 1.3 times more outpatient visits than did children with GSDIa

Table 3. Total number of hypoglycemia events resulting in emergency room/inpatient visits.

Pediatric patients (<8 years at encounter)	**56% (10/18)**
Emergency room visit	1
Emergency room and inpatient visit	7
Inpatient Visit	2
Outpatient Visit	8
Pediatric patients (<18 years at encounter)	**100% (4/4)**
Emergency room visit	0
Emergency room and inpatient visit	1
Inpatient Visit	3
Outpatient Visit	0
Adult patients (>18 years at encounter)	**10% (1/11)**
Emergency room visit	0
Emergency room and inpatient visit	1
Inpatient Visit	0
Outpatient Visit	10

- Hypoglycemia leading to emergency room/inpatient visits was more commonly seen in pediatric versus adult patients with GSDIa

CONCLUSIONS

- GSDIa is a chronic and progressive inherited metabolic disorder that is associated with numerous clinical complications and high healthcare resource utilization in both children and adults
- In this real-world analysis of nine patients, children had more emergent (acute) health-care interactions, which continued into longer-term (chronic) based healthcare visits
- Despite strict dietary management, there remains a high unmet need for new therapies that target the underlying cause of GSDIa to improve patient outcomes and minimize the impact on healthcare systems

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DISCLOSURES

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