People and families affected by GSDIa: An analysis of narrative accounts written by caregivers and individuals living with GSDIa

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*at the time the research was conducted

BACKGROUND

- Glycogen storage disease type Ia (GSDIa) is a rare metabolic disorder caused by deficiency of the glucose-6-phosphatase enzyme[1]
- Affects approximately 1:125,000 births each year[2]
- Manifestations can be variable and severe:
  - Short stature, enlarged liver and kidneys (hepatomegaly and nephromegaly), and altered blood chemistry (hypoglycemia, hyperlipidemia, lactic acidemia, and hyperuricemia)[1,2]
- Daily impact of GSDIa on patients and their families is not well described in literature
  - It is important for decision-makers to understand the impact of GSDIa
- This study was designed to describe the individual experiences of those affected by GSDIa through qualitative methods by asking patients and caregivers to provide written summaries of how the condition affects them

Conventional Approach

- Existing PRO tools or bespoke questionnaires may not be suited to GSDIa
  - PROs do not capture the richness and individuality of living with a disease
  - No tools have established validity in the disease
- Taking part in interviews or focus groups is not always convenient for participants
  - Semi-structured interviews may not allow participants to describe their experiences freely and in their own words

Our Approach

- We asked people living with GSDIa and caregivers to write narrative accounts of their experience of living with or caring for someone with GSDIa

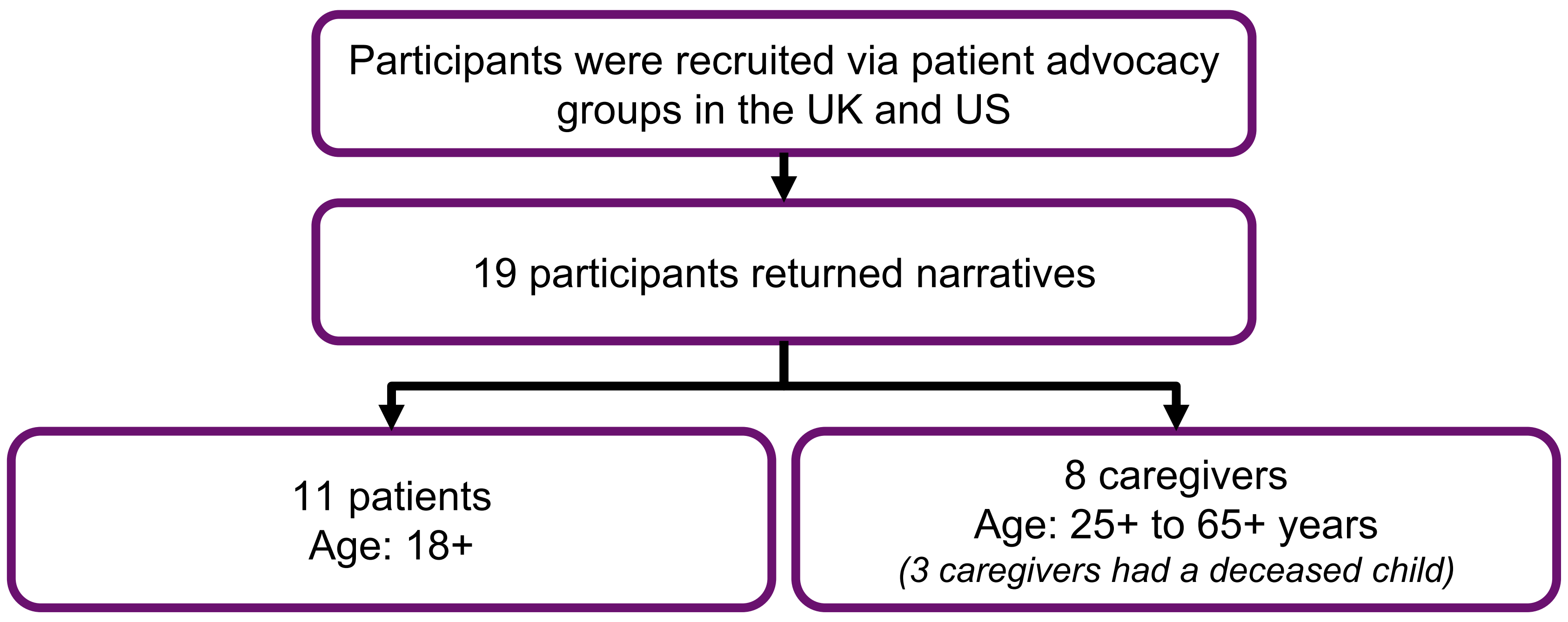
OBJECTIVE

To identify the aspects of the lives of patients and caregivers that are impacted most by GSDIa

METHODS

- People with GSDIa and caregivers were asked to write an account of how they are affected by the disease **(Figure 1)**
- Participants could:
  - Write about any aspect of their experience that they wanted
  - Write as much as they wanted
- Narratives were assessed using an inductive (data-driven) approach to identify common themes across participants

Figure 1. Study Participants



Participants were recruited via patient advocacy groups in the UK and US

19 participants returned narratives

11 patients
Age: 18+

8 caregivers
Age: 25+ to 65+ years
(3 caregivers had a deceased child)

RESULTS

After reviewing the responses of patients with GSDIa and caregivers, several key areas of impact were identified:

Disease Burden

- Most common symptoms reported by:
  - Participants with GSDIa: **Blood sugar swings** (7/11), **lack of energy** (6/11), **short stature** (5/11)
  - Caregivers (relating to their child): **Blood sugar swings** (4/8), **short stature** (3/8), **enlarged liver** (3/8)
- Additional symptoms reported (2+ participants):
  - **Sweating, hunger, high triglycerides, breathing difficulties, pallidity, seizures, slow development**
- **Frequent admissions to hospital**

> *Many of us with GSD have wild blood sugar swings. Like, we can be at 140 and in 40-60 minutes can fall 100 points, even with eating or having cornstarch like we normally would.*
> *– Patient*

Managing Disease

- The **challenge of the continuous need for glucose supplementation**
  - All participants described **constant vigilance** and 'living by the clock' and the need to adhere to a strict eating and drinking schedule
  - Many (14/19) reported **needing cornstarch through the night**
  - Many reported issues associated with cornstarch consumption including **limiting exercise, weight gain and digestive problems**
- **Missed cornstarch or mild illness can rapidly lead to hypoglycemia, hospitalization and/or "coma and [death]"**
- Caregivers were **unable to leave their child** in the care of others

> *Simple things such as getting a good night sleep are impossible for me, as I have to wake up every four hours in order to consume cornstarch,… so I am constantly exhausted and deprived of sleep.*
> *– Patient*

Diagnosis and Healthcare

- Participants living with GSDIa were **diagnosed as infants**
- Most caregivers described the **difficult process of achieving a GSDIa diagnosis**, with symptoms appearing shortly after birth and diagnosis reported between 3-18 months of age
  - Many caregivers were **told that their child may only live 1–2 years**
- Ongoing support from HCPs was also reported to be not always good
- Patients and caregivers reported that **HCPs often lacked knowledge** of how to treat patients: one participant told how **a friend died because they did not receive the right care**, two participants described how they had to **intervene to prevent medical professionals from administering the wrong treatment**

> *I was lucky I was still conscious and able to stop the medical staff from giving me a shot of insulin… which was the extreme opposite of what I needed"*
> *– Patient*

Mental Health and Social Relationships

- The most commonly reported **emotional impacts** by:
  - Participants with GSDIa: **Desire for better life** (8/11), **worry and fear** (5/11), **frustration and anger** (5/11), **mental health, anxiety, body image issues** and **positive attitude** (4/11)
  - Caregivers (relating to self): **worry and fear** (5/8), **exhaustion, challenging, positive attitude and gratitude** (3/8)
  - Caregivers (relating to their child): **frustration and anger** (3/8**), positive attitude** (3/8)
- **People described fear**, mainly around risk of death and many expressed a **desire for new treatment**
- Two participants **reported suicidal thoughts**
- **Social and romantic relationships were impacted**

> *I am never more than 10 hours from death… It gives me extreme anxiety... – Patient*

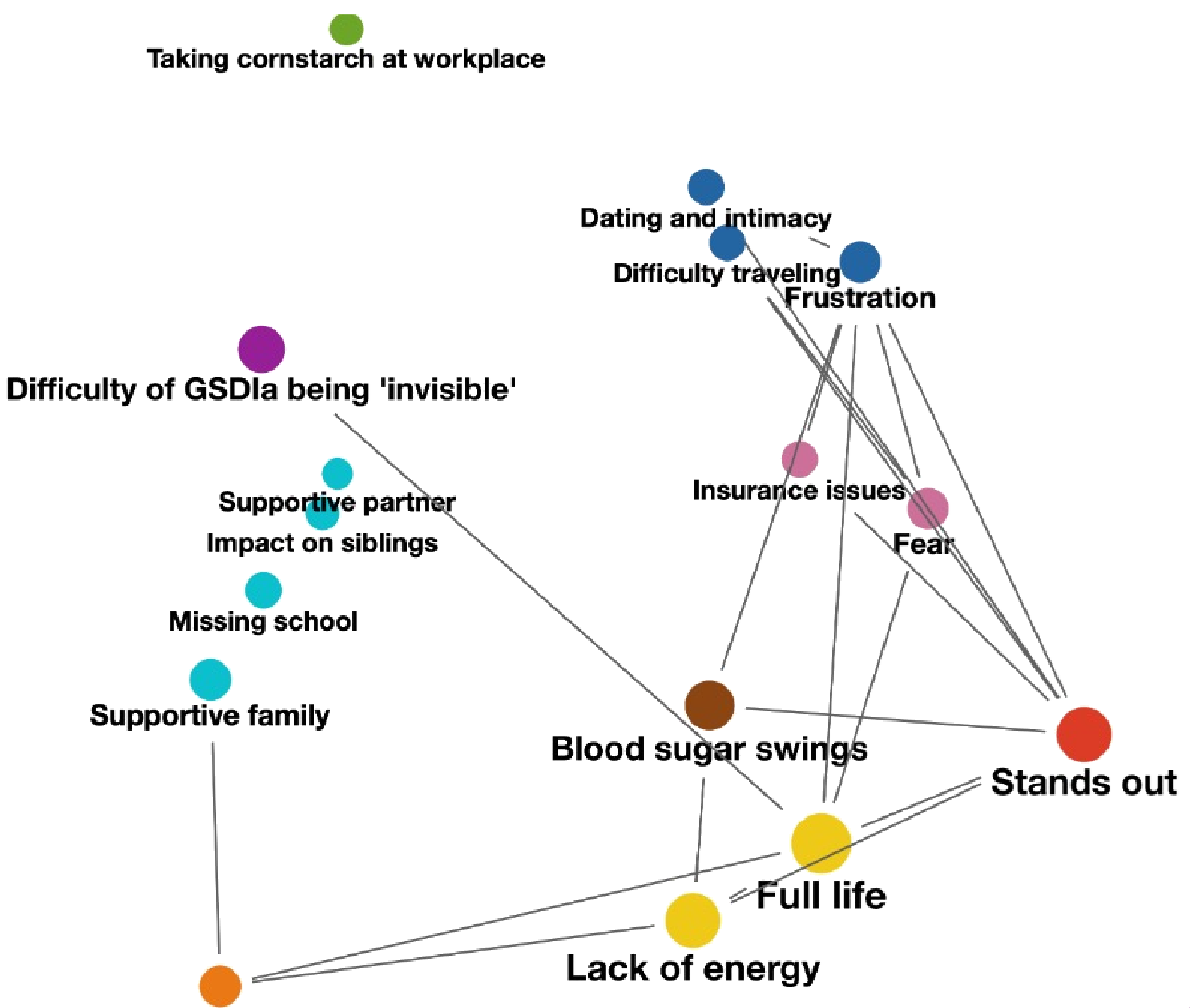
Daily Activities, Work, and Family Life

- The mostly commonly reported **social impacts** by:
  - Participants with GSDIa: **invisibility of GSDIa** (6/11), **standing out from peers** (5/11), **stigma** (4/11), **supportive friends** (4/11)
  - Caregivers (relating to their child): **unable to sleep over** (4/8), **missing out socially** (2/8), **social awkwardness** (2/8)
- **Daily living was impacted** by the need to limit physical activity (7/19) and the inability to travel (8/19)
- **Work/school impact**:
  - Participants with GSDIa/Caregivers (relating to their child): **taking cornstarch** (5/19), **missing school** (2/19) **and/or limited job opportunities** (2/19)
  - Caregivers (relating to self): **Reduced hours** (2/8), **career side-lined** (3/8)

> *I have personally had to put my career to one side, it is almost impossible to have a career with a child that needs specialist care.*
> *– Caregiver*

Network of Concepts

- Diagram illustrates how concepts co-occurred
- Connecting lines show where concepts were co-reported 4+ times
- Proximal codes were frequently co-reported
- Colors represent clusters



Word clouds of the emotions spontaneously reported by participants living with GSDIa **(Figure 2)** and caregivers **(Figure 3)** were generated from responses. The larger the word appears, the more frequently it was reported.

Figure 2: Participants living with GSDIa

Figure 3: Caregivers of individuals living with GSDIa

CONCLUSIONS

- The descriptive data from this study reflect the main concerns of people with GSDIa and caregivers
- The narrative approach was flexible, which allowed for:
  - A rich dataset
  - A good number of participants for an ultra rare condition
- This study provides a useful approach for capturing the patient voice in rare diseases such as GSDIa

DISCLOSURES AND ACKNOWLEDGMENTS

EK is an employee and stockholder of Ultragenyx Pharmaceutical Inc. The Odyssey Program is sponsored by Ultragenyx Pharmaceutical Inc. Medical writing assistance was provided by Jack Pike, PhD of Ultragenyx Pharmaceutical Inc.

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1. Kishnani PS, et al. *Genet Med.* 2014; 16(11):e1.
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Thank you to the participants for sharing their experiences, and to the Association for Glycogen Storage Disease in the UK and the USA and The Children's Fund for Glycogen Storage Disease Research in the USA for their support.

Presented at the *International Society for Pharmacoeconomics and Outcomes Research* Annual Meeting*;* 6–9 November 2022; Vienna, Austria