

The slide features a central white circle containing the title text. The background is composed of several overlapping shapes: a solid yellow rectangle on the left, a large brownish-tan shape at the bottom, and a complex green geometric pattern on the right side.

**MSUD PATIENT  
REGISTRY WITH  
CORDS**

# Why do we need a MSUD patient registry?

- To help researchers and clinicians understand the challenges of living with MSUD
  - Medical, emotional, cognitive
- To understand how living with MSUD changes throughout the lifespan
- To identify gaps in MSUD research
- To connect patients and families with researchers
- To support applications to the FDA for new therapeutics
- To improve quality of life for our community



# WE NEED INFORMATION FROM EVERYONE!

- Everyone living with MSUD
- Experiences of those who are no longer living
- Classic, Intermittent, Intermediate, Thiamine-responsive
- Transplanted
- Worldwide

# WE HOPE TO LEARN ABOUT THE LIVED EXPERIENCE OF THOSE IN OTHER COUNTRIES

- ▶ The questionnaire has been translated into Spanish
- ▶ We are in the process of translating the questionnaire into Arabic

**ANNUAL UPDATES  
NEEDED TO DESCRIBE  
HOW LIVING WITH  
MSUD CHANGES OVER  
TIME**



# HOW HARD IS IT TO COMPLETE THE MSUD QUESTIONNAIRE?

- ▶ It will take 20-40 minutes to complete
  - ▶ You don't need to complete it all at once
- ▶ Most questions are easily answered
- ▶ If you're unsure of your mutation or your blood leucine level at diagnosis, check with your clinic
- ▶ When providing your annual update, only answer questions in which the answers have changed



**WE NOW HAVE 193  
PARTICIPANTS!**

**THANK YOU!**





## Preliminary findings of a patient registry for MSUD at Coordination of Rare Diseases at Sanford (CoRDS) Sanford Research International Patient Registry Program

Dolins K<sup>1</sup>, Elaina Jurecki<sup>2</sup>, Alexandra Larkin<sup>3</sup>

<sup>1</sup> Teachers College Columbia University, Research Lead MSUD Family Support Group, NY, NY, USA

<sup>2</sup> National Phenylketonuria Alliance, San Ramon, CA, USA

<sup>3</sup> Clinical Sciences, PTC Therapeutics, South Plainfield, NJ, USA

### BACKGROUND

Maple Syrup Urine Disease (MSUD) is an inborn error of metabolism causing an impaired ability to metabolize branched-chain amino acids (leucine, isoleucine, and valine) and has a worldwide incidence estimated at 1:190,000. A higher incidence is observed in populations with consanguinity (Mennonites, Arabs, Ashkenazi Jews). If untreated, MSUD results in severe neurological impairment, coma, and death. Treatment centers around a protein-restricted diet supplemented with a branched chain restricted amino acid fortified medical food, and monitoring of blood amino acid levels. Illness can result in metabolic decompensation and death. This registry was launched in 2020 with goal of understanding the lived experience of patients and families worldwide.

### METHODS

A registry questionnaire was developed by the MSUD Family Support Group and CoRDS.<sup>1</sup> It was launched in 2020 and promoted using direct email, phone calls, and social media. As interim analyses revealed that many questions were unanswered, the tool was revised in 2022 to address ambiguities. Responses were stratified by age group (<18 and ≥ 18 y.o.). Questions with multiple-selection answers may result in a summation of percentages greater than 100%.

### CONCLUSIONS

These descriptive analyses provide preliminary data describing the experiences of those with MSUD. Missing data continues to hamper analysis and result interpretation. Future efforts will include calling respondents to clarify responses, requesting assistance from clinicians in guiding patients as they complete the questionnaires, encouraging greater participation in the registry, and translating questionnaires into other languages such as Arabic to achieve a more diverse demographic. Self-report is an additional limitation and may result in inaccuracies; however, responses reflect the perceptions of the participants. A patient registry is essential to understanding the lived experience and needs of those impacted by MSUD and informing future research directions. This registry represents the largest collection of MSUD patient responses to date. A more robust dataset will enable us to provide an in-depth analysis aimed at identifying the key needs of individuals and will help improve their quality of life. We gratefully acknowledge the contributions of those individuals who have participated in this

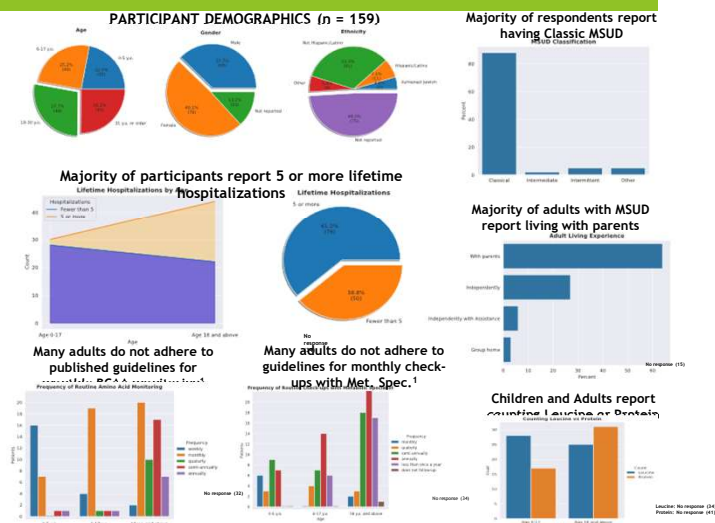
### REFERENCES

<sup>1</sup>MSUD CoRDS Patient Registry <https://msud-support.org/patient-registry/>

<sup>2</sup>SERN GMDI MSUD Nutrition Management Guidelines v1.58 June 2021

<https://managementguidelines.net/guidelines.php/105/MSUD%20Nutrition%20Guidelines/Version%201.54> accessed 3/5/24

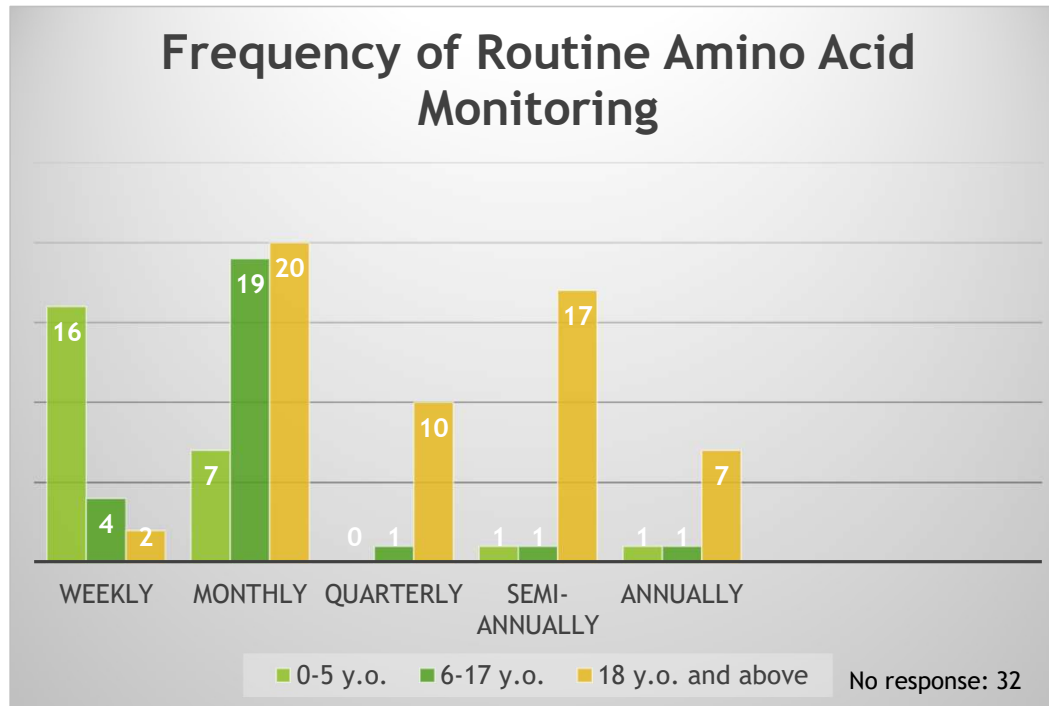
### RESULTS



Your information has been presented at professional conferences

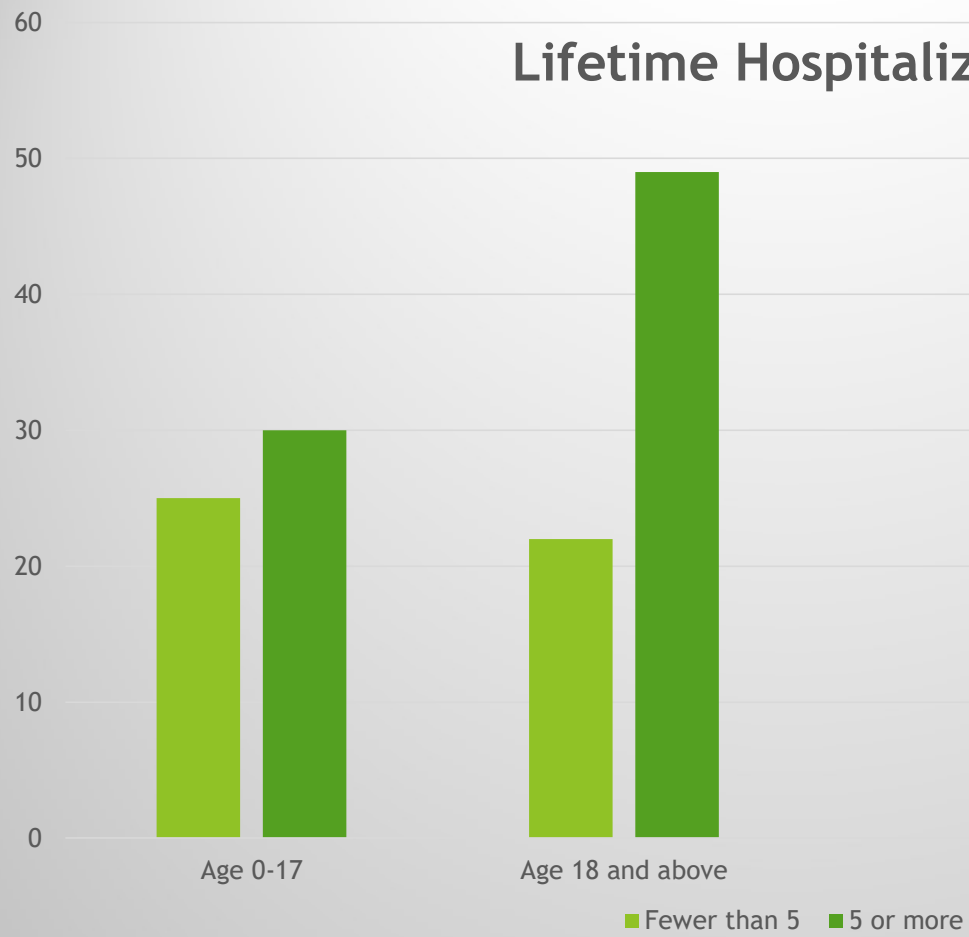


# SAMPLE DATA



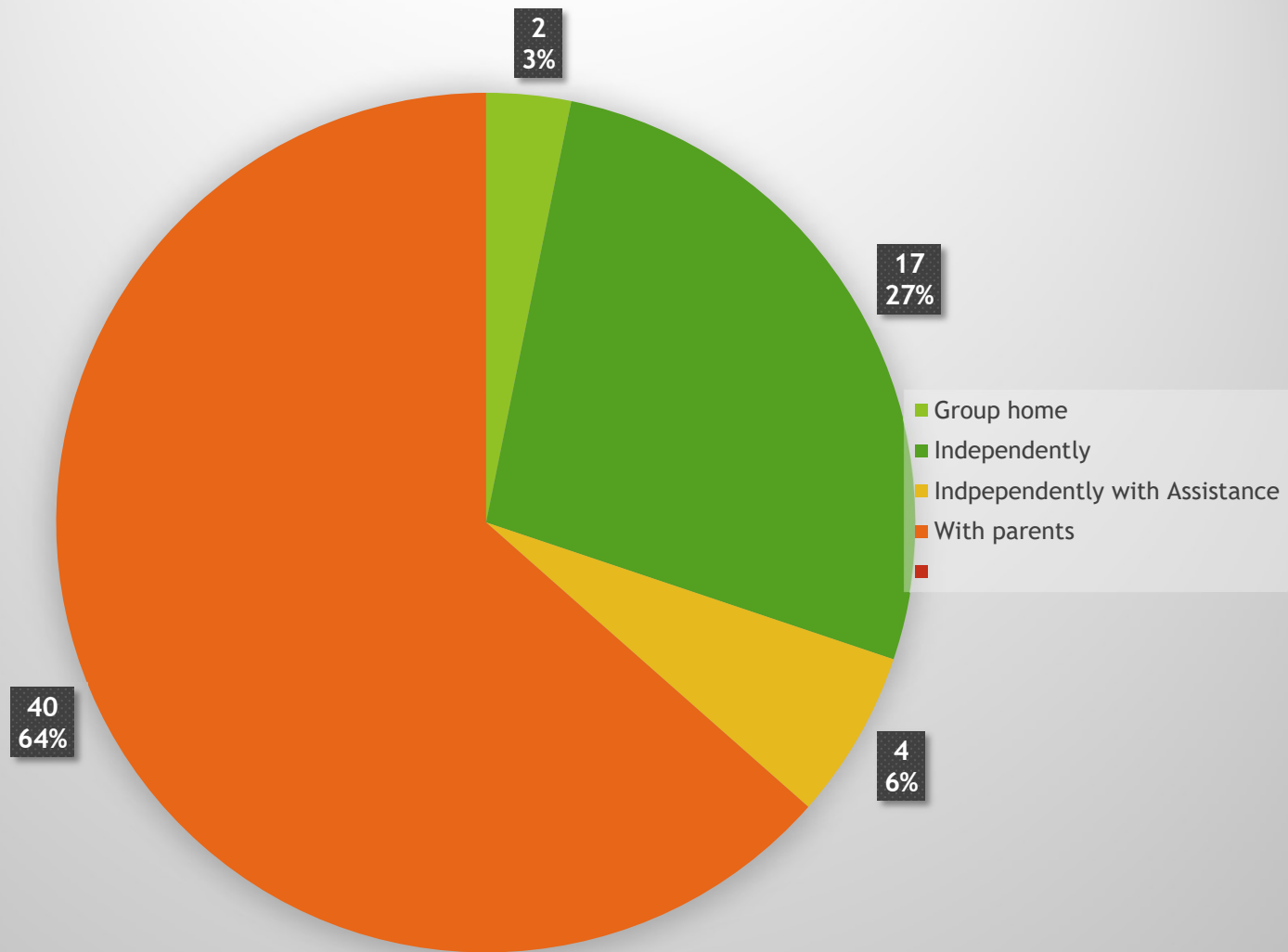
SERN GMDI Recommended Guidelines: Daily or weekly until 2 years, monthly for other age groups

## Lifetime Hospitalizations



No response: 33

# Adult Living Experience



# HOW YOU CAN HELP STRENGTHEN OUR REGISTRY

- ▶ If you haven't yet done so, enroll today!
- ▶ If you are unsure how to answer some questions, ask us!
  - ▶ Call CoRDS with questions about the general questionnaire
  - ▶ Call or email Karen with questions about the MSUD-specific questionnaire