

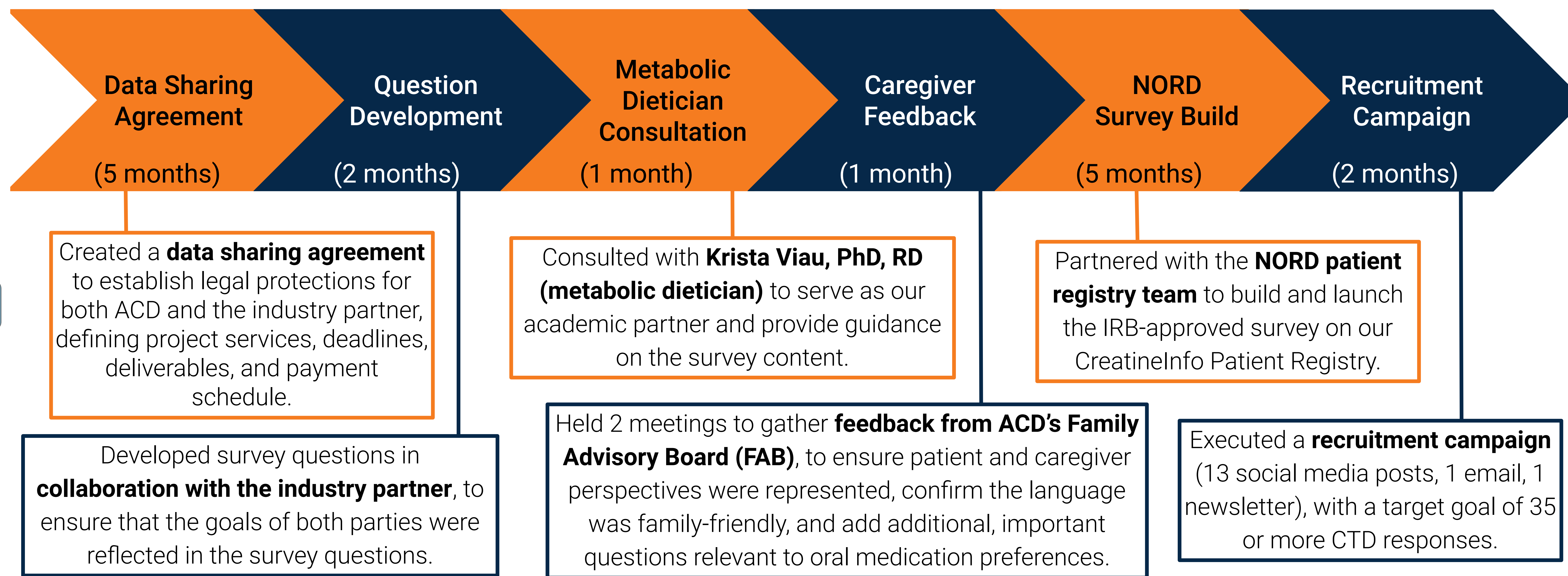
Background

- Creatine transporter deficiency (CTD) is a rare X-linked cerebral creatine deficiency syndrome (CCDS).¹
- Individuals with CTD may show intellectual and developmental delays, expressive speech and language delays, autistic-like behavior, hyperactivity, seizures, gastrointestinal issues, and movement disorders.^{2,3}
- There is currently **no treatment or cure for CTD**.

Objectives

- An **industry partner** wanted to learn about the oral medication preferences of CTD patients and caregivers to inform the development of their therapeutic, a possible treatment for CTD.
- **Goal:** Collaborate with an industry partner to **develop a custom oral medication survey** for our CreatineInfo Patient Registry and Natural History Study.

Methods



Results

Project Outcomes:

1. Industry and non-profit data sharing agreement appropriate for future use
2. 52-question custom oral medication survey
3. Successful recruitment campaign with high participant engagement surpassing our target goal, with 37 CTD participants completing the survey in the first 6 weeks
4. 55-page report containing aggregate, de-identified data from the survey
5. Valuable insights into patient oral medications preferences to share with additional stakeholders in future drug development efforts (see *Figures 1 & 2*)
6. Industry partner support of patient registry costs resulting in a self-sustaining registry
7. Better understanding of the time requirements, and therefore appropriate cost to charge, for this type of endeavour.

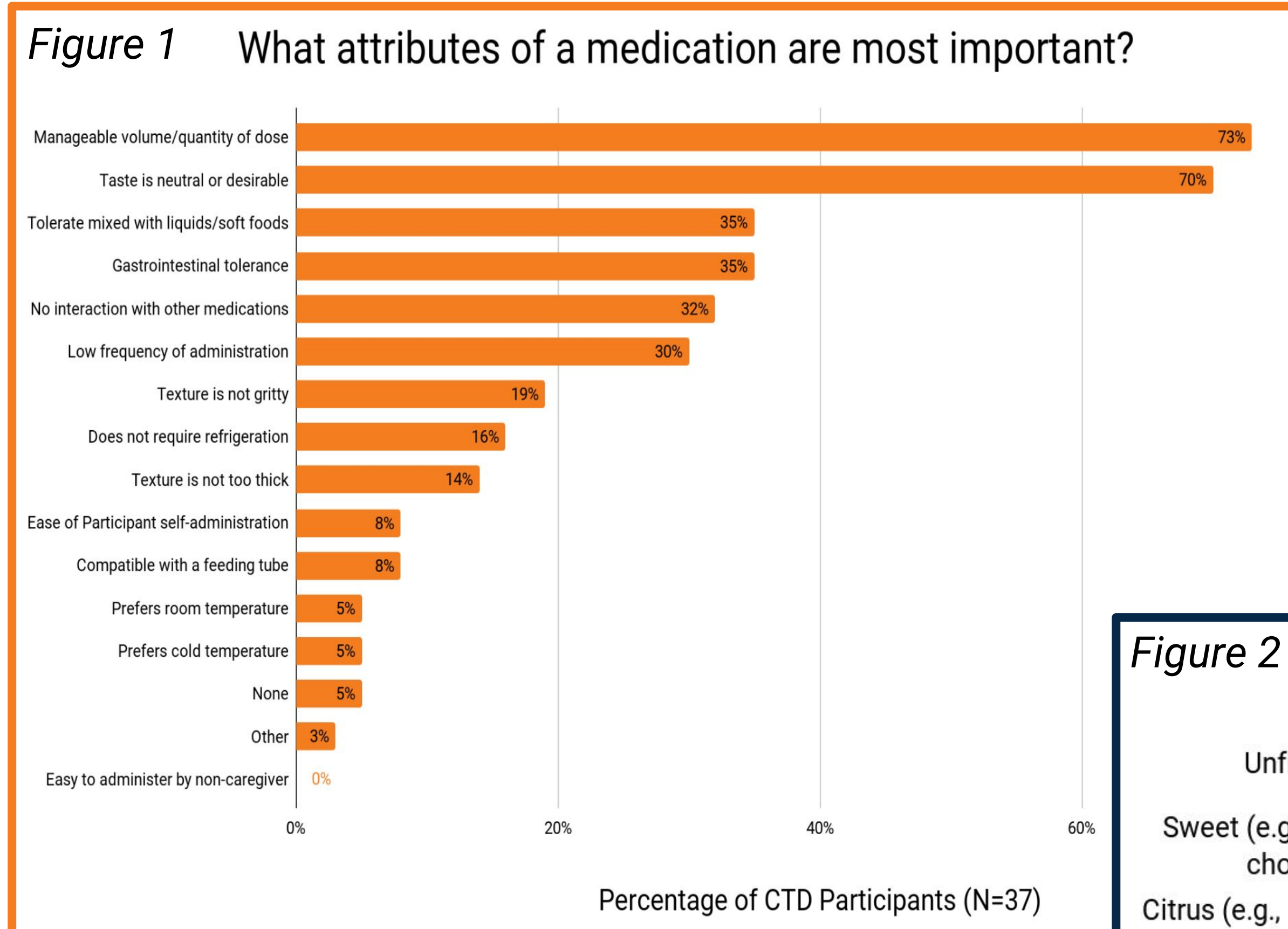


Figure 1: Percentage of CTD survey participants who included the medication attribute in their top 4 most important. Overwhelmingly, participants rated "Manageable volume or quantity of dose" and "Taste is neutral or desirable" as the top most rated attributes (N=37).

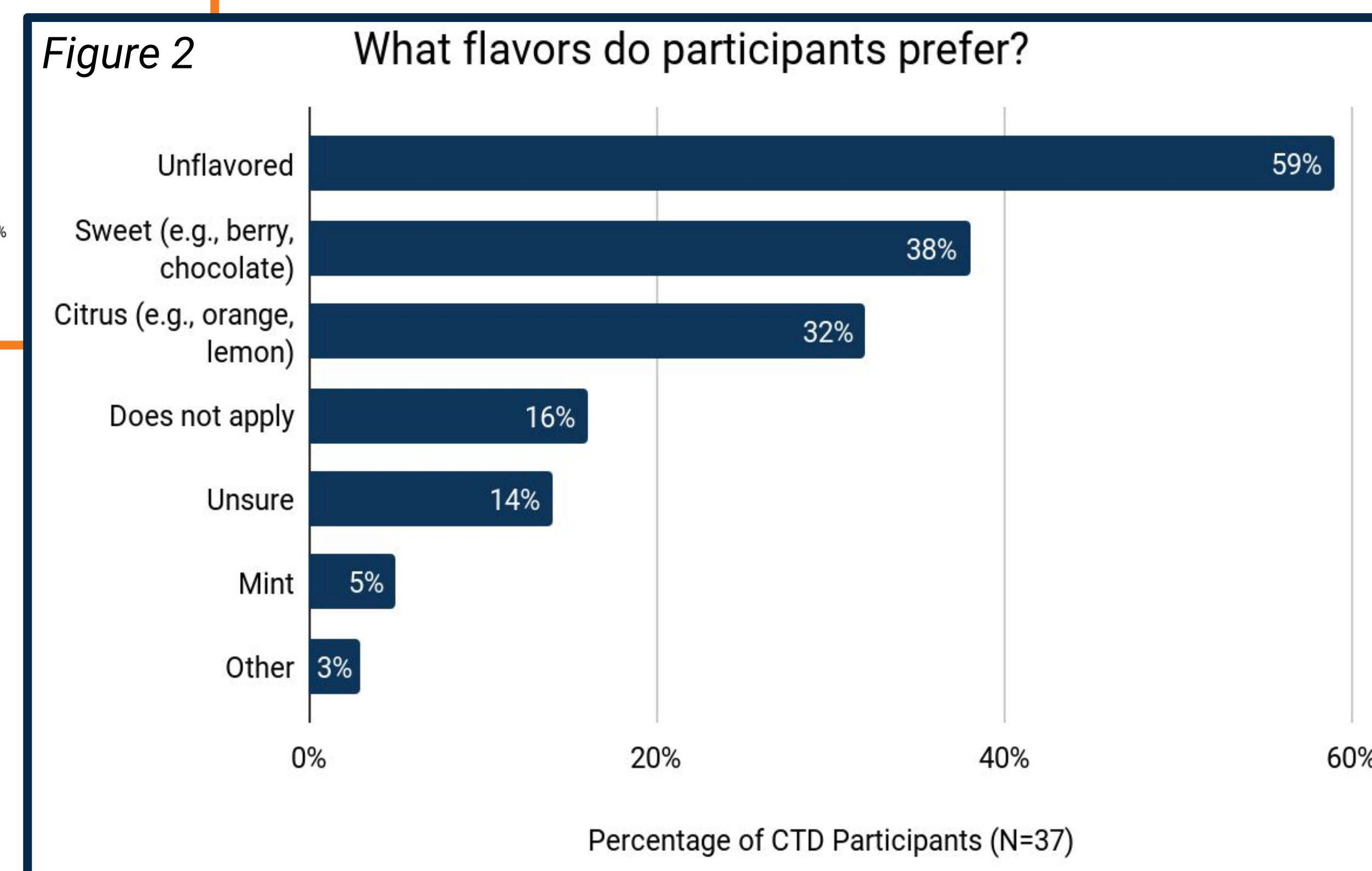


Figure 2: Percentage of CTD survey participants who selected each flavor. "Unflavored" was selected by the greatest percentage of participants, followed by "Sweet" (N=37).

Conclusions

- Co-development of a custom patient registry survey can be a mutually beneficial endeavor for both patient advocacy groups and industry partners.
- Survey participants were incentivized to complete the survey by simply knowing that their data had meaningful implications.
- Survey objectives should be well-defined and agreed upon early in the process to avoid timely delays in the development of questions.

References & Contact

1. Salomons, G. S., van Dooren, S. J., Verhoeven, N. M., Cecil, K. M., Ball, W. S., Degrauw, T. J., & Jakobs, C. (2001). X-linked creatine-transporter gene (SLC6A8) defect: a new creatine-deficiency syndrome. *The American Journal of Human Genetics*, 68(6), 1497-1500.
2. Passi, G. R., Pandey, S., Devi, A. R. R., Konanki, R., Jain, A. R., Bhatnagar, S., ... & Jain, V. (2022). Cerebral creatine deficiency disorders—A clinical, genetic and follow up study from India. *Brain and Development*, 44(4), 271-280.
3. Data collected from the CreatineInfo Patient Registry and Natural History Study (2023).

Let's Connect!
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