Elevating patient and caregiver voices: How CCDS families are taking a patient-centered approach to prepare for clinical trials

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Introduction

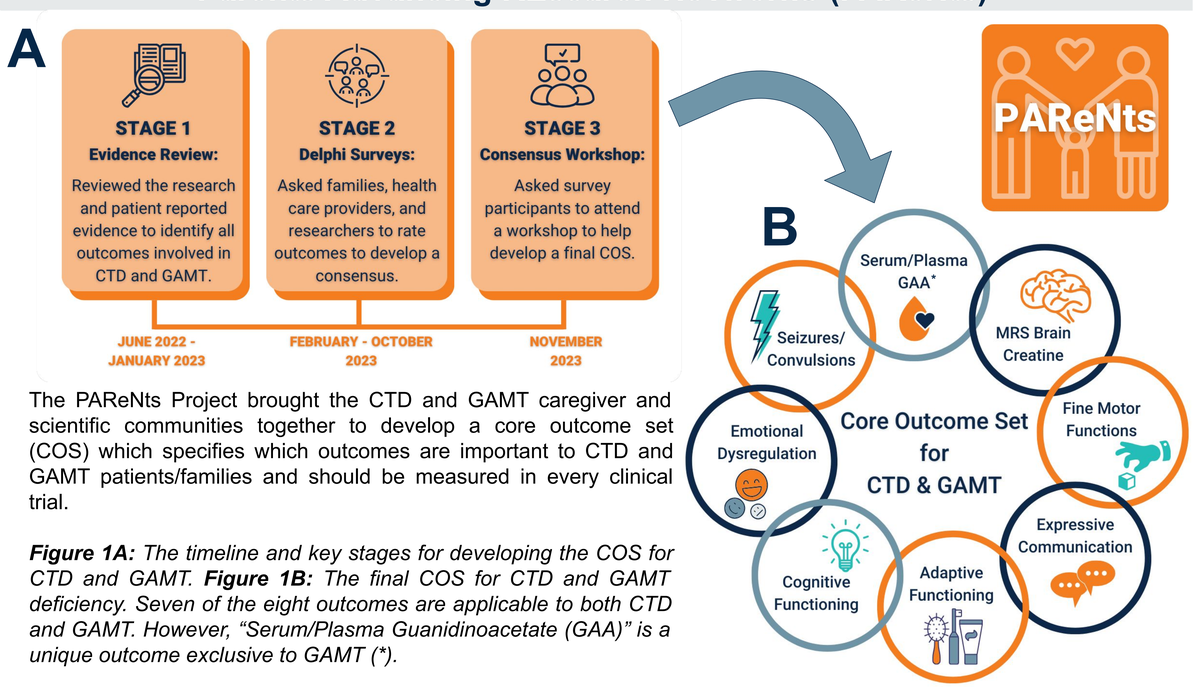
- Creatine transporter deficiency (CTD) and guanidinoacetate methyltransferase (GAMT) deficiency are cerebral creatine deficiencies (CCDS) that cause intellectual and developmental disabilities, behavior problems, speech delay, seizures, and motor impairments.[1]
- No FDA-approved treatments exist. Caregivers frequently express a desire for a better long-term solution, ideally an FDA-approved treatment that leads to improved symptoms and quality of life.[2]
- Clinical trials often differ in how they select and measure outcomes, posing challenges to their interpretation and real-world application.[3] Additionally, research outcomes often do not fully reflect patient priorities nor are the tools used to measure the outcomes sensitive enough to capture meaningful changes in patients.[4,5]

Objectives

- Engage patients and caregivers in preparation for clinical trials.
- Develop a patient-centered core outcome set (COS) for CTD and GAMT deficiency to ensure consistent evaluation of therapies which are meaningful to patients and caregivers.
- Identify a set of patient-centered considerations for researchers as they select tools to measure the outcomes in the COS during clinical trials.

Methods

Parents Advancing REsearch NeTworkS (PAReNts)



The PAReNts Project brought the CTD and GAMT caregiver and scientific communities together to develop a core outcome set (COS) which specifies which outcomes are important to CTD and GAMT patients/families and should be measured in every clinical trial.

Figure 1A: *The timeline and key stages for developing the COS for CTD and GAMT.* ***Figure 1B:*** *The final COS for CTD and GAMT deficiency. Seven of the eight outcomes are applicable to both CTD and GAMT. However, "Serum/Plasma Guanidinoacetate (GAA)" is a unique outcome exclusive to GAMT (*).*

Methods

Parents Advancing REsearch NeTworkS 2.0: Expand Community, Tools, & Engagement (PAReNts 2.0: ExCiTE)

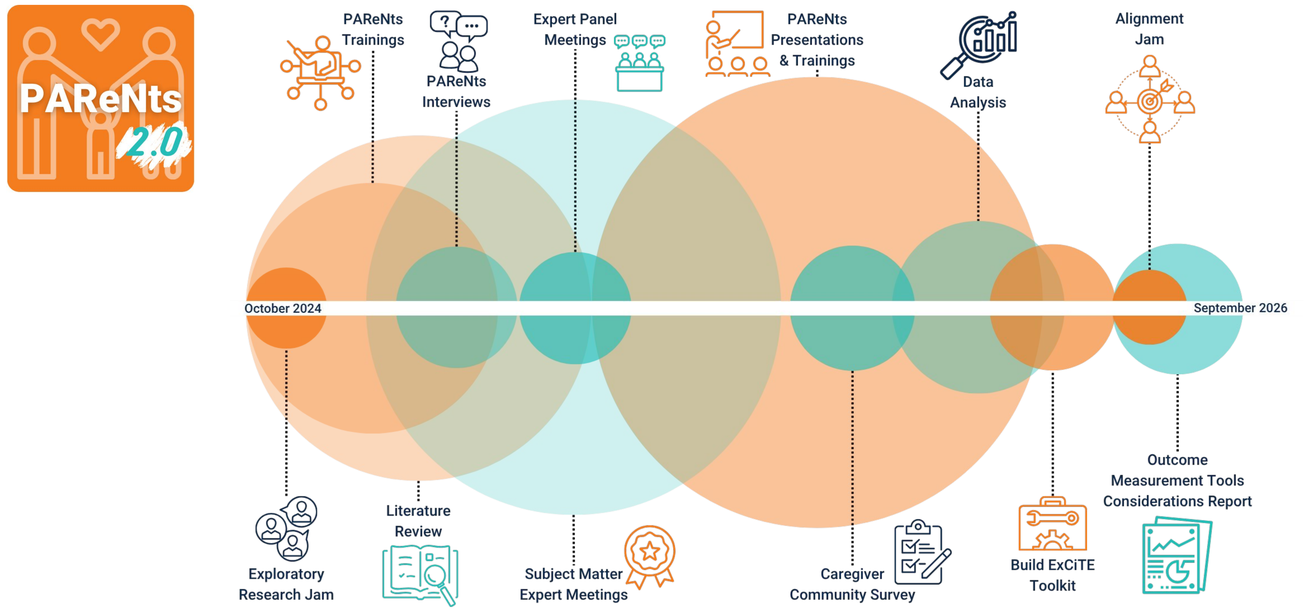


Figure 2: *The PAReNts 2.0 Project timeline and key stages for developing "Considerations for CTD & GAMT Outcome Measurement Tools".*

While identifying which outcomes to measure was an important first step in our first PAReNts Project, it is just as important to consider the tools used to measure those outcomes and their sensitivity to capture subtle but meaningful changes in patients. Therefore, the PAReNts 2.0 Project was designed as a follow-up to again bring together the CTD and GAMT caregiver and scientific communities to identify a set of patient-centered considerations for selecting tools to measure the outcomes in the COS during clinical trials

Conclusions

- Engaging tools, such as Delphi surveys and community surveys, allow patients and caregivers to be partners in clinical trial preparations.
- Focus groups, exploratory meetings, and caregiver surveys complement traditional literature reviews to gather data and help shape valuable resources for patient-centered trial design.
- Patients and caregivers are better prepared to share their expectations in future clinical trials when engaged in patient-centered trainings.

References & Funding

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