



Succinic Semialdehyde Dehydrogenase Deficiency (SSADHD): Qualitative Needs Assessment for Patients With a Rare Neurological Disorder



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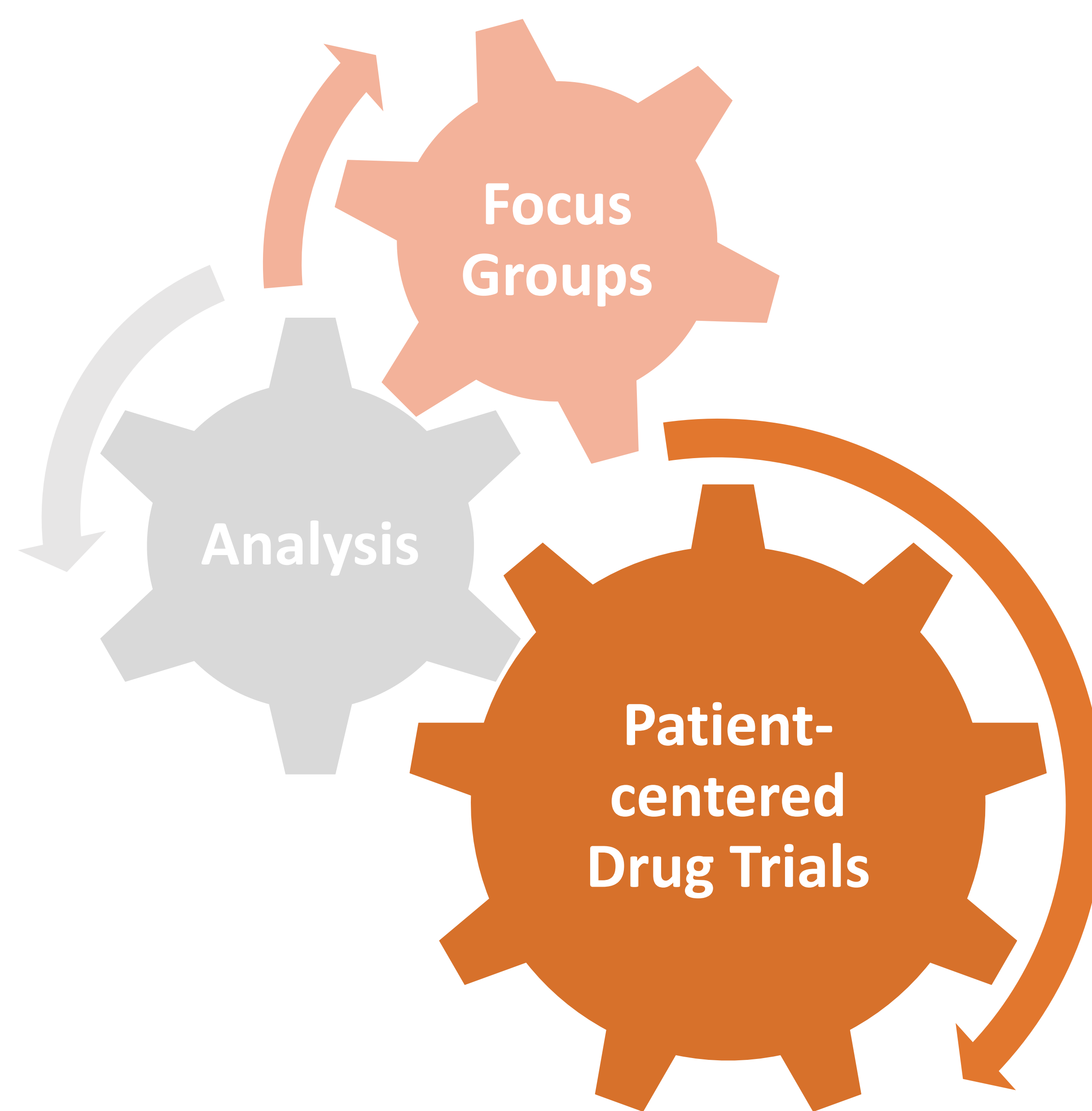
Introduction

Succinic Semialdehyde Dehydrogenase Deficiency is a rare, autosomal recessive disorder that is characterized by the inefficient breakdown of GABA (gamma-Aminobutyric acid), an inhibitory neurotransmitter. The lack of degradation leads to a spectrum of neurological deficits, most commonly: seizures, ataxia, hyporeflexia, and behavioral problems. There has been growing interest in the impact of clinical trials on the quality of life of the patients affected by SSADHD and their families. This study seeks to understand highlighted concerns of caregivers and will serve as a starting point to determine the focus of future of clinical trials.

Focus Group Summary

What do you feel are the greatest impacts SSADHD has on your child's day-to-day experience?	<ul style="list-style-type: none"> Communication Social interactions Independence Sleep issues Assistive Devices Special Education Impact on other family members
What would you want to see improved with treatment in your child(ren) with SSADHD?	<ul style="list-style-type: none"> Improved speech Improved focus Improved fine motor skills Improved gross motor skills Improved sleep Improved social interactions and decreased social anxiety
What concerns would you have if you were considering enrolling your child in a clinical trial for SSADHD treatment?	<ul style="list-style-type: none"> Safety information History of the research on the therapeutic Invasiveness of the procedures Timetable and planning of the roll-out of the trial
What resources would your family need if you were to enroll your child in a clinical trial for SSADHD treatment?	<ul style="list-style-type: none"> Risk v reward Contraindications with current medications Efficient communication that would allow for updates on the clinical trial, results and future research
If there were a treatment available that needed to be regularly administered to your child with SSADHD, what would be the best way to administer the treatment?	<ul style="list-style-type: none"> Preference for oral administration of treatment Injections would also be acceptable Considerations would have to be made regarding interruptions for their school day
If you were responsible for administering this new treatment to your child with SSADHD, up to how many times a day would you be willing to administer treatment?	<ul style="list-style-type: none"> Overwhelming adavance that if there was a possibility that their child's and their family's day-to-day experience would improve, "nothing was off the table."

- ❖ The project is an FDA-guided qualitative needs assessment from the caretaker community of patients with SSADHD
- ❖ Serves as a first step in patient-focused therapeutic development and evaluation for SSADHD



The objectives of these focus groups are the following:

- ❖ To identify endpoints/outcomes of interest to the SSADHD community for clinical trials
- ❖ To determine the impact of clinical trial participation in the SSADHD community

Focus Group Participants

<p>Focus Group 1</p> <ul style="list-style-type: none"> 6 parents of individuals with SSADHD 1 brother of 2 individuals with SSADHD 1 great-grandmother of an individual with SSADHD 2 of the parents were representing their son with SSADHD as the mother and father 2 mothers and 1 father were each individually representing their child(ren) with SSADHD 	<p>Focus Group 2</p> <ul style="list-style-type: none"> 3 of the participants were individually representing their child with SSADHD 1 participant was a translator representing both the mother and child with SSADHD 	<p>Focus Group 3</p> <ul style="list-style-type: none"> 2 participants were representing their son with SSADHD as the mother and father 2 participants were individually representing their child with SSADHD as their mother 1 participant was individually representing their child with SSADHD as their father
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In total, there were 17 participants of the focus groups, representing 15 individuals with SSADHD.

Conclusions

- ❖ Clinical outcomes that assess communication ability, cognitive functions, gross and fine motor skills.
- ❖ Improvements in communications, independent behavior, sleep and social interactions
- ❖ A clinical research team that collaborates with families and patients to coordinate trial procedures and clinical care
- ❖ Timing of clinical assessments planned accordingly to needs and potential trial burden of patients
- ❖ Materials about the nature of the study at the beginning (including pre-clinical and preliminary clinical data), as well as periodic updates to trial participants and their families

Acknowledgements

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