New Family Welcome Packet

PPP3CA HOPE FOUNDATION™

www.ppp3ca.org
info@ppp3ca.org

Welcome to the PPP3CA community! Our families come from all over the globe and our children vary in age and abilities. We are a community that continously learns from each other. We hope you find this group as helpful for you as it was for each of our families.

ABOUT THIS GROUP

This group consists of families with loved ones diagnosed with PPP3CA mutations. We provide emotional and information support as appropriate, as well as raise money to fund research projects aimed at finding a cure for PPP3CA mutations. For any questions or comments, please contact us at info@ppp3ca.org.

WHAT IS PPP3CA HOPE FOUNDATION™?

PPP3CA Hope Foundation™ is a nonprofit leading research projects to learn the impacts of genetic mutations in PPP3CA. The foundation was founded by three parents whose daughters were diagnosed with PPP3CA. You can learn more about the foundation at www.ppp3ca.org.

WHAT RESEARCH IS BEING CONDUCTED?

The research is seeking treatment to reduce the impacts of the mutation or even reverse it. A team of researchers from 3 top tier research institutions in the USA have come together to learn more about PPP3CA and find potential treatment options.
Read more about the research: www.ppp3ca.org/research
Meet the Research Team: www.ppp3ca.org/meet-the-researchers

HOW DO YOU JOIN THE RESEARCH?

You can join this research by joining Simons Searchlight. The complete process is outlined below:
1. Create an account on simonssearchlight.org
2. Create profiles for your child with the gene change and any other siblings under 18 that you wish to participate.
3. Complete the Simons Searchlight Connect Questionnaire
4. Complete consent for yourself and all children participating
5. Send Simons Searchlight your child's genetic testing results

Once steps 1-4 are completed, a number of questionnaires are posted to the portal including a medical history questionnaire. The genetic testing results do not hold up this process but are required before a family is considered enrolled

HOW DO YOU JOIN THE PPP3CA PATIENT REGISTRY?

In order to organize the information within the families, we maintain an online registry/database. You can join our database by filling the form: https://forms.gle/24kN2sBGB3NgXAis6
You can connect with families by joining the private Facebook group "PPP3CA Hope Foundation."

WHAT RESOURCES ARE AVAILABLE?

Our website (www.ppp3ca.org) and Facebook page have many resources. Please reach out if you have more questions. We also have a public social media profile on Instagram @ppp3cahopefoundation

PPP3CA CARE BEFORE THE CURE: GUIDEBOOK

This guidebook was developed by the PPP3CA Hope Foundation to help you learn important information about individuals with PPP3CA gene mutations and how to best support them. The content found within is meant for informational and educational purposes only and does not substitute professional medical advice or consultations with healthcare professionals.

DEFINITION

PPP3CA developmental and epileptic encephalopathy is a neurodevelopmental disorder caused by variants, or mutations, in genes that encode parts of a protein complex called Protein Phosphatase 3CA (PPP3CA).

DIAGNOSIS

PPP3CA developmental and epileptic encephalopathy (DEE91) is diagnosed through genetic testing, most commonly through Whole Exome Sequencing.

INHERITANCE

PPP3CA developmental and epileptic encephalopathy is typically de novo, meaning it was non inherited from either parent.

FAMILY PLANNING

When PPP3CA developmental and epileptic encephalopathy is de novo, the recurrence risk to siblings is estimated to be 1%. Individuals with PPP3CA mutations have a 50% chance of passing it on to their children.

COMMON FEATURES

The impact of PPP3CA varies in severity between individuals. The table on the following page is a summary of the main symptoms that have been reported by our community. Please note these symptoms might or might not be present for everyone with the diagnosis.

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LIVING WITH PPP3CA

Common Symptoms for People Living with PPP3CA Mutations:

Seizures
Most individuals with PPP3CA have seizures or abnormal brain activity. They tend to be very challenging to control.

Global Developmental Delays
Global developmental delays impact almost all milestones including walking and talking.

Low Muscle Tone
Low muscle tone makes all motor skills challenging. There is no way to fix muscle tone, but you can build strength with therapies. This impacts all muscles.

Language Difficulties
Learning how to speak and communicate is very challenging for individuals with PPP3CA. Communication devices are helpful.

Autism
Many individuals are diagnosed with autism and have difficulty interacting with others and controlling emotions. Some have very aggressive behaviors.

Gross Motor Delays
Crawling, walking and other gross motor skills are often very delayed and/or may never be reached.

Fine Motor Delays
Fine motor skills are often delayed resulting in difficulty to self-feed, get dressed and play with toys.

Sleep Challenges
Some individuals may have difficulty falling asleep or staying asleep. Medications such as melatonin may help with this.

Vision Difficulties
These include strabismus, cortical visual impairment, amblyopia and far sightedness. Glasses, surgery and vision therapy are recommended.

Feeding Challenges
Due to low tone, fine motor delays and overall developmental delays, some individuals will have difficulty eating and may need a feeding tube and/or feeding therapy.

COMMON FEATURES OF PPP3CA

SYMPTOM	IMPACT	RECOMMENDED EVALUATION	RECOMMENDED INTERVENTION
Hypotonia (low muscle tone)	• Gross motor delays • Fine motor delays • Mobility • Speech • Feeding/swallowing	Neurology	• Physical Therapy • Occupational Therapy • Orthotics
Speech Delays	• Issues with communication	Speech Therapy	• Speech Therapy • Communication devices
Vision Difficulties	• Strabismus/esotropia • Astigmatism • Far sighted • CVI	Ophthalmology	• Glasses • Vision Therapy • Surgery
Feeding Challenges	• Trouble chewing and swallowing	Speech Therapy Occupational Therapy	• Feeding Therapy • Occupational Therapy
Gastrointestinal Challenges	• Constipation • Diarrhea	Gastrointestinal Specialist	• Specialized diet
Seizures	• Varying types	Neurology/Epilepsy (EEG, MRI)	• Medications • Specialized diet • Surgery
Global Developmental Delay	• Delay in achieving developmental milestones	Neurology Developmental Pediatrics	• Frequent Therapies (eg: PT, OT, Speech)
Sleep Challenges	• Trouble falling asleep or staying asleep	Neurology (EEG) Sleep Specialist (sleep study)	• Medications
Learning Disabilities	• Challenges at school	Special Education Team	• Special accommodations • Communication support
Challenging Behaviors	• Mood swings • Self injurious	Psychologist Psychiatrist	• Medications
Sensory Challenges	• Autism • Sensory Processing Disorder	Psychologist Neurologist Pediatrician	• Medications • Occupational Therapy

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RECOMMENDED INTERVENTIONS FREQUENCY

Maximizing on therapies especially through early development has been proven beneficial. Families are encouraged to access the therapies without sacrificing their mental health and the quality of life of the entire family. We recommend that you connect with local disability groups to get a better understanding of available resources.

DEVELOPMENTAL DELAYS MANAGEMENT

Consultation with a developmental pediatrician is recommended to ensure families are supported in maximizing quality of life for their children.
The following information represents typical management recommendations for individuals with developmental delays in the United States; standard recommendations may vary from country to country.
Ages 0-3 years: Referral to an early intervention program is recommended for access to occupational, physical, speech, and feeding therapy. In the US, early intervention is a federally funded program available in all states.
Ages 3-5 years: In the US, developmental preschool through the local public school district is recommended. Before placement, an evaluation is made to determine needed services and therapies and an individualized education plan (IEP) is developed.
Ages 5-21 years: In the US, an IEP based on the individual's level of function should be by the local public school district. Affected children are permitted to remain in the public school district until age 21. Discussion of transition plans including financial, vocation/employment, and medical arrangements should begin at 12 years. Developmental pediatricians can provide assistance with transition to adulthood.

SPECIAL ACCOMODATIONS

Communication: Speech delays are particularly common in individuals with PPP3CA mutations. We recommend alternative ways of communicating such as sign language, pictures, and a communication device to support general communication and promote speech development. We also recommend working with a speech therapist.
Assistive Technology: The field of assistive technology has come a long way and can expand abilities. Some examples include communication devices, communication buttons, speech to text, typing, and read/write programs. A speech therapist can help with these.
Motor support: While gross motor skills vary across individuals with PPP3CA, support such as a stander or a walker might be required until walking is reached. Some families may use adaptive strollers or wheelchairs.
Special Diet: Some families have been successful in improving seizure activity through the Keto diet. Others have reported improved overall health and cognition through the gluten free and dairy free diet especially when food sensitivities were detected.

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ALTERNATIVE THERAPIES

These are interventions that our families have tried and recommended.

For Motor Skills	For Speech Development	Other
Horse Riding Therapy	Listening Therapy	Reiki (energy)
Aqua Therapy	Prompt - oral motor	Applied Behavioral Analysis (ABA) Therapy
Terasuit (intensive PT)	Z vibe vibrating device - oral motor	Metha casting (scoliosis)
Dynamic Movement Intervention (DMI)	Class/Book: More Than Words	Chiropractor
Conductive Education	Gemini Software	Acupressure Acupuncture Brushing
		Music and Art Therapy

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WELCOME NEW FAMILIES!

We are so glad you found us. You are not alone. You have a new community here at PPP3CA Hope Foundation to support you every step of the way. Research is currently underway to find a treatment and cure for PPP3CA. In the meantime, when you're ready, here are some ways to get your connected to our community. Please reach out for anything; we are here for you.

GET CONNECTED

1- Contact Us: visit ppp3ca.org and fill out the information to connect with our community.

2- Join our Facebook community: Search PPP3CA Hope Foundation in Facebook

GET INVOLVED

1- Join Simons Searchlight: visit www.simonssearchlight.org

2- Complete our patient registry: visit https://forms.gle/24kN2sBGB3NgXAis6

3- Help fundraising efforts: to fund our current research

GET INFORMED

1- Read: your welcome letter and new family packet

2- Visit our website: ppp3ca.org

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SIMONS SEARCHLIGHT
Driven by science. United by hope.

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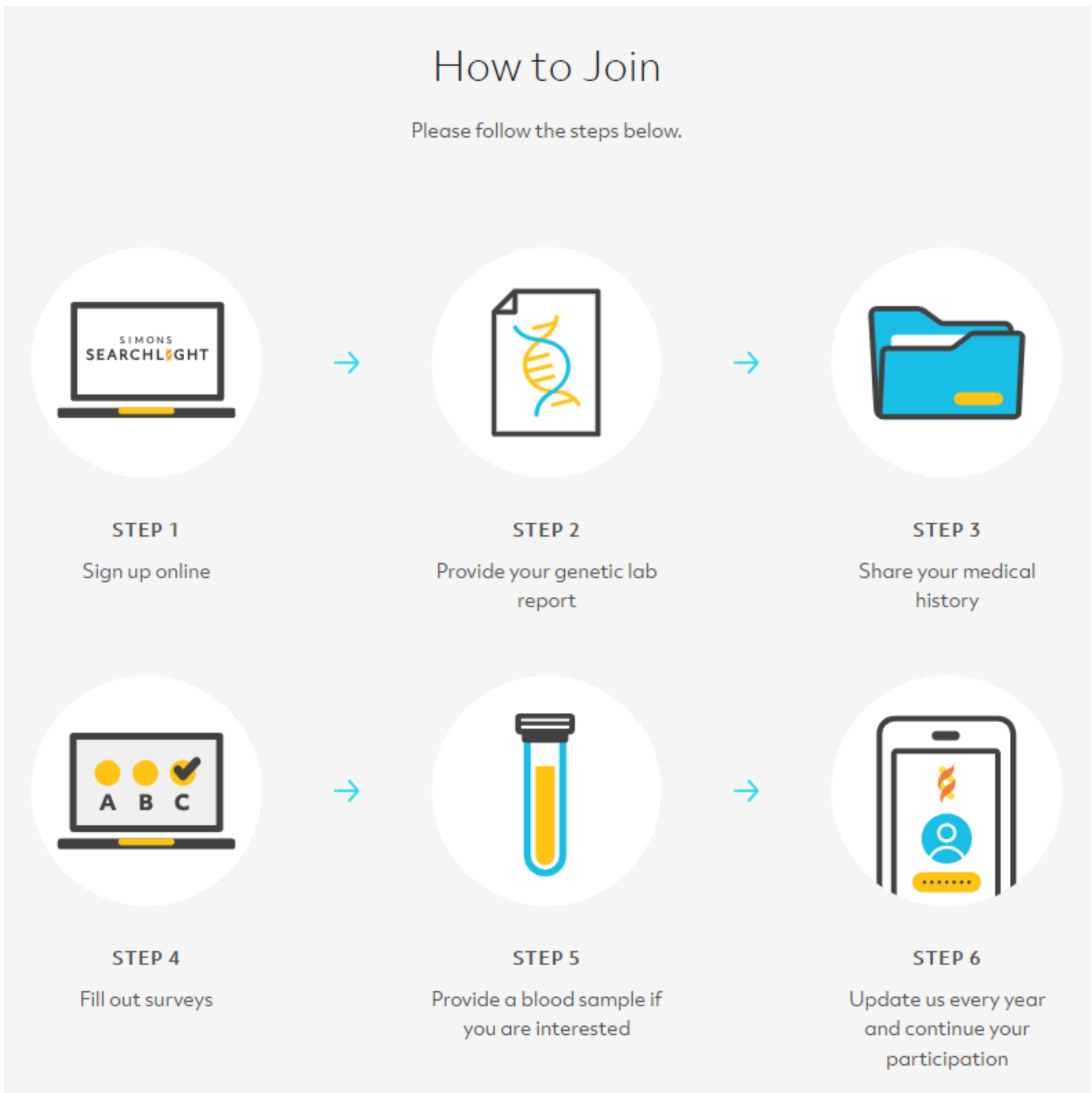
Simons Searchlight is an international research program that is creating an ever-growing database and resource network of rare genetic neurodevelopmental disorders including PPP3CA. To expedite the path to a cure, we need to register as many children with the mutation as possible.

We know that going through this isn't easy and thankfully Simon Searchlight makes the registration process very easy. Should you run into any challenges, the support team at Simon Searchlight is happy to assist and even walk you through the process.

To start the process, go to simonssearchlight.org and click "Join Us."

SIMONS SEARCHLIGHT



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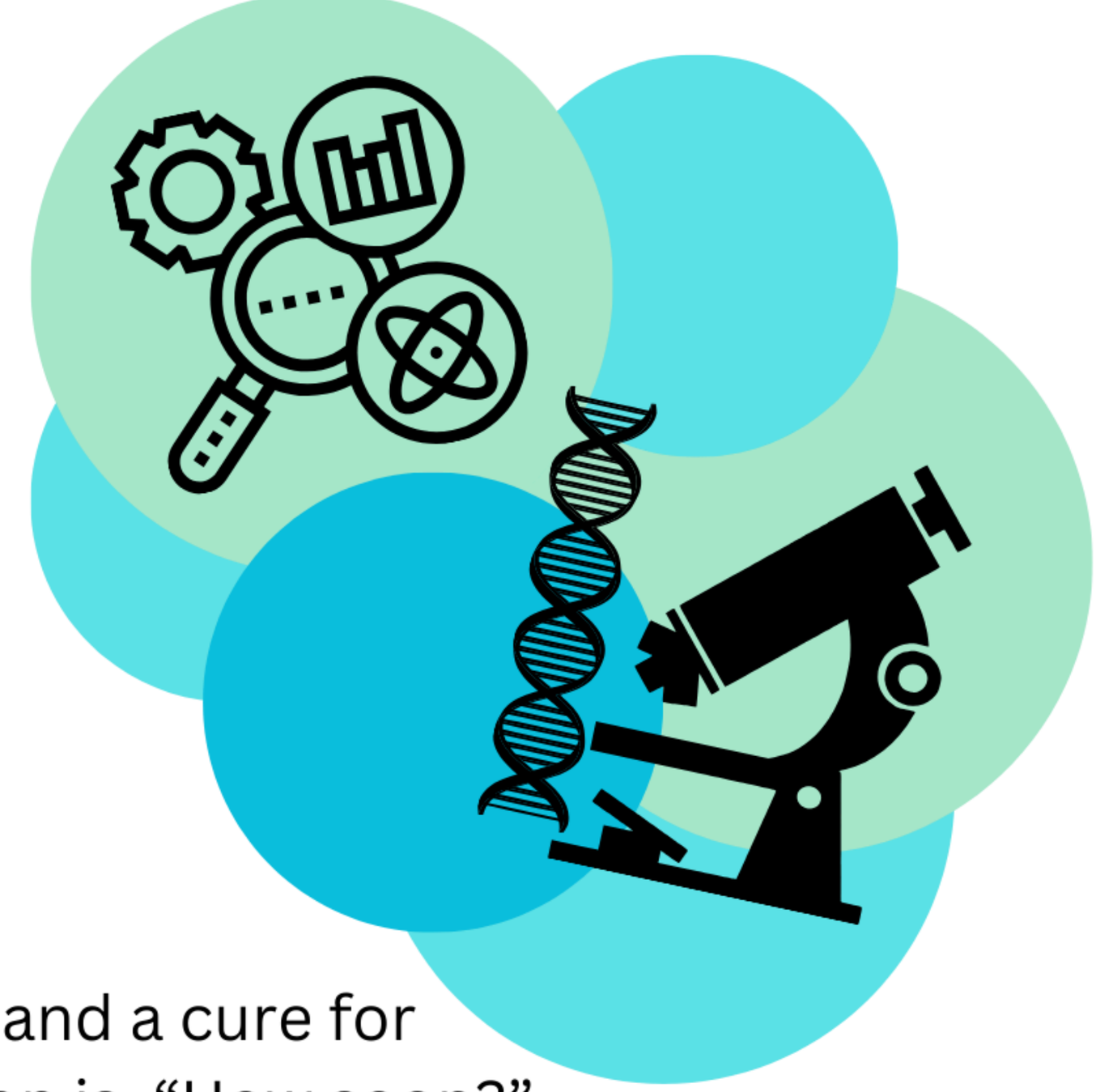
PPP3CA Hope Foundation: JOURNEY TO A CURE



There will be a treatment and a cure for PPP3CA - the only question is, “How soon?”

Below outlines our research plan over the next 5-10 years. Your donation will directly fund these efforts.

1- Build PPP3CA Models

- Identify patients with PPP3CA variants
- Create mice, yeast, zebrafish and stem cells with these PPP3CA variants

2- Study these Models

- Understand the impact of PPP3CA mutations on the brain and body
- Learn what treatments may be effective

3- Test Treatments on Models

- Screen existing drugs to determine if they can be repurposed
- Identify therapies like ASOs, gene therapy, RNA splicing.

4- Safety Studies

- Conduct safety studies on potential therapies in animal models

5- Clinical Trials

- Conduct clinical trials in people with PPP3CA to determine if potential therapies work
- Submit for FDA and other regulatory agency approvals