



Probably Genetic Introduction

Ultragenyx Rare Disease Entrepreneur Bootcamp
May 2024

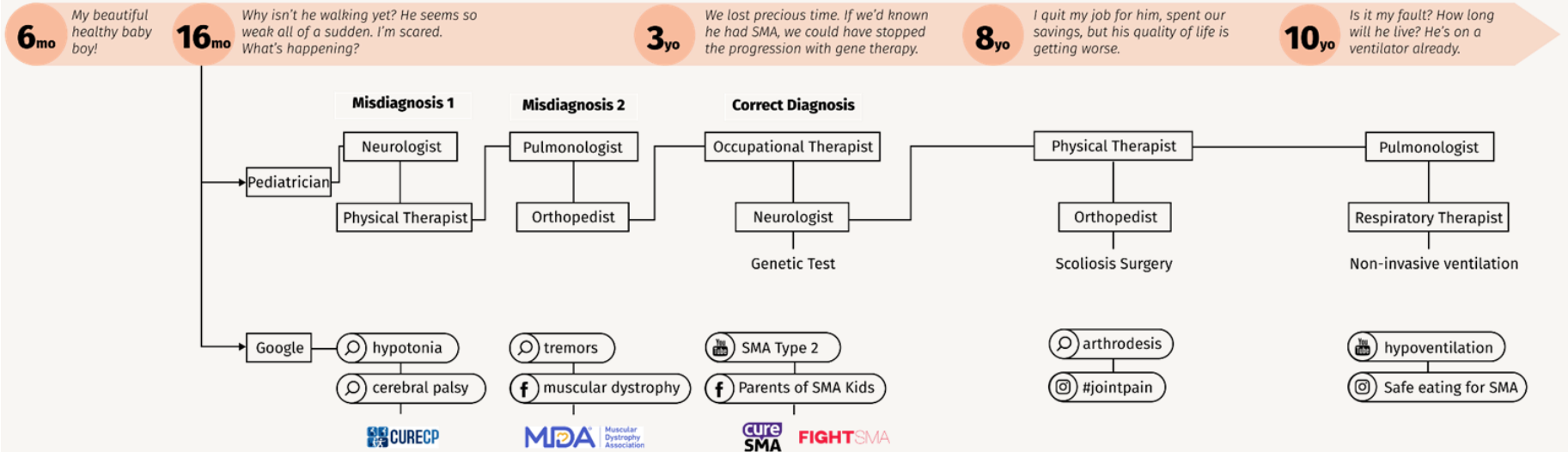
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**30 million people in the US
suffer from a genetic disease**

Patients are sick for years before getting diagnosed

Meet Logan & his mom Danielle.

This is his journey getting diagnosed with Spinal Muscular Atrophy (SMA) Type 2.

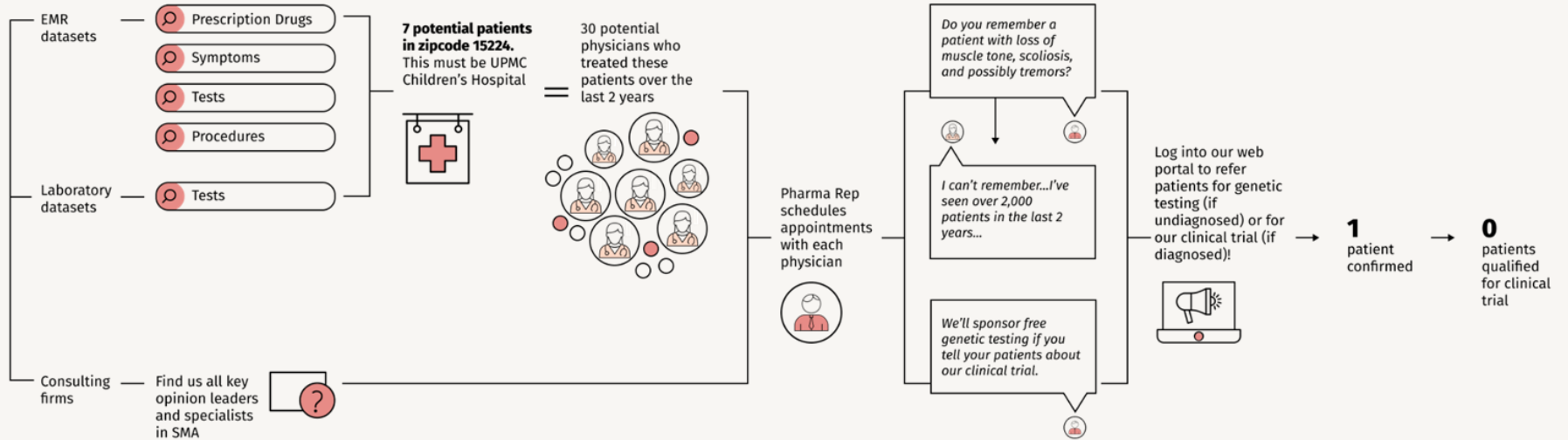


- ▶ **Lack of access to specialists** who would recognize genetic disease symptoms.
- ▶ **Misdiagnoses** lead to wrong treatments and missed treatment opportunities
- ▶ **Staggering emotional and financial strain** on patients and their families.

Finding patients offline is hard

Meet Novartis.

This is their journey to find a Spinal Muscular Atrophy patient for their trial.



▶ Data lives across different EMRs, specialists, and labs, which makes it **extremely hard to identify patients.**

▶ Because of regulatory restrictions, **pharma companies waste time and money trying to find physicians treating target patients.**

▶ **Targeting physicians is an ineffective way to get patients into trials and onto treatments.**

Current solutions only find a subset of patients ...



EHR Records & Claims Searches

Did you hire vendors who analyze medical claims & EMR data, but **don't deliver because patient data is distributed across many different systems and most patients are undiagnosed?**



Awareness Campaigns

Did you partner with specialists, key opinion leaders, and PAGs, but **struggle to convert these awareness efforts into leads?**



Sponsored Testing

Did you sponsor testing, but only **reached a subset of patients who see the right referring specialist?**



Homegrown Solutions

Did you piece together vendors to create your own compliant patient finding funnel, but **struggle with low conversion rates and uptake?**

... We find everyone



Diverse and representative of the entire US vs. just those with access to specialists



Disengaged or underserved but engaging with their symptoms and diagnostic journey online



Consented and can be activated via compliant communications suite

Probably Genetic helps patients and pharma find each other, solving 3 core challenges for pharma and patients.

1. ONLINE PATIENT ACQUISITION

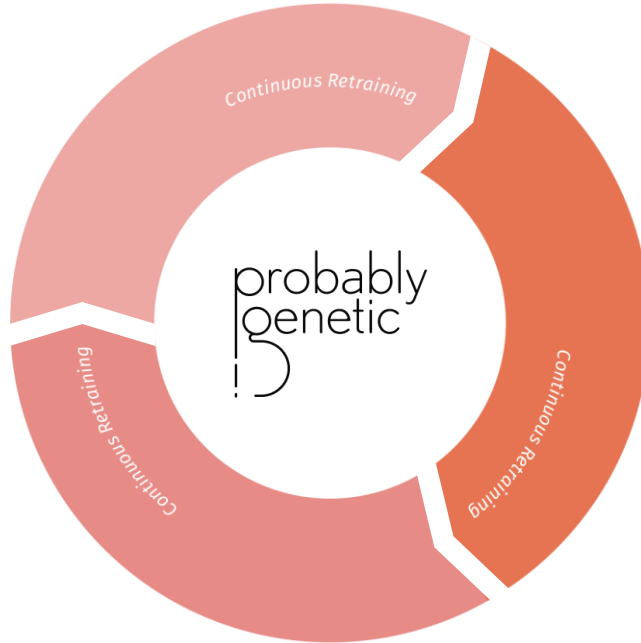
Direct access to hard-to-reach patient populations

Exclusive patient advocacy partnerships and online advertising power our unique direct-to-patient recruitment strategy.

3. PATIENT INTELLIGENCE & ACTIVATION PLATFORM

Seamless recruiting and drug development support

A flexible database enables pharma companies to analyze patient data and activate patients directly.



2. ML-PATIENT SCREENING

Efficient conversion of patients from undiagnosed to diagnosed

Our proprietary machine learning platform identifies undiagnosed patients based on a variety of data types

Text	Photos	Videos*	Audio*
Phenotypic Terms	Face	Seizures	Voice
Family Pedigree	Eyes*	Movement	Hearing
Medical History	Skin*	Behavior	
Meta Data	Hands*		
Lab Reports	Feet*		
EMR Data*			

* Not yet collecting

Pharma companies use our platform to launch treatments



Discover which physicians are treating target patients.



Learn which variants target patients carry.



Activate patients to enroll them in clinical trials or get them onto treatments.



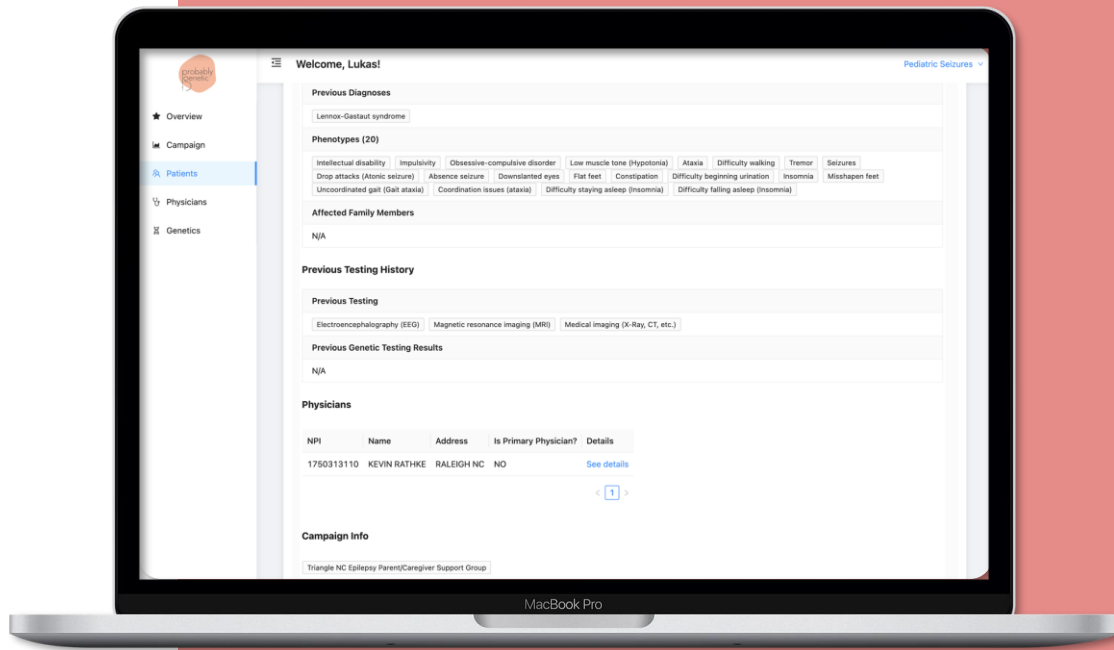
Understand the phenotypic profile of your target patients.



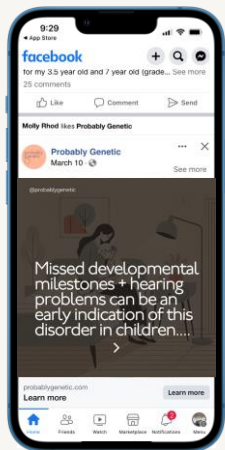
Get information directly to your target patients, not just physicians.



Analyze the diagnostic journey of your target patients.

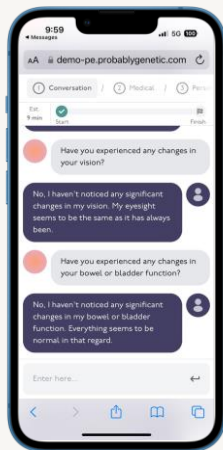


Patients get free testing



Step 1 Discover Outreach

- Find us on social media, Google, our website, a Patient Advocacy website, and other online channels



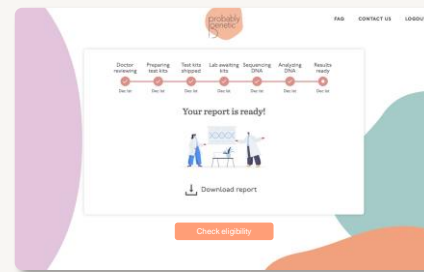
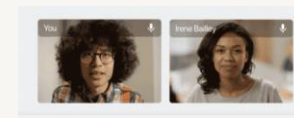
Step 2 Answer a Few Questions

- Complete a chat conversation with our LLM - ChatPG



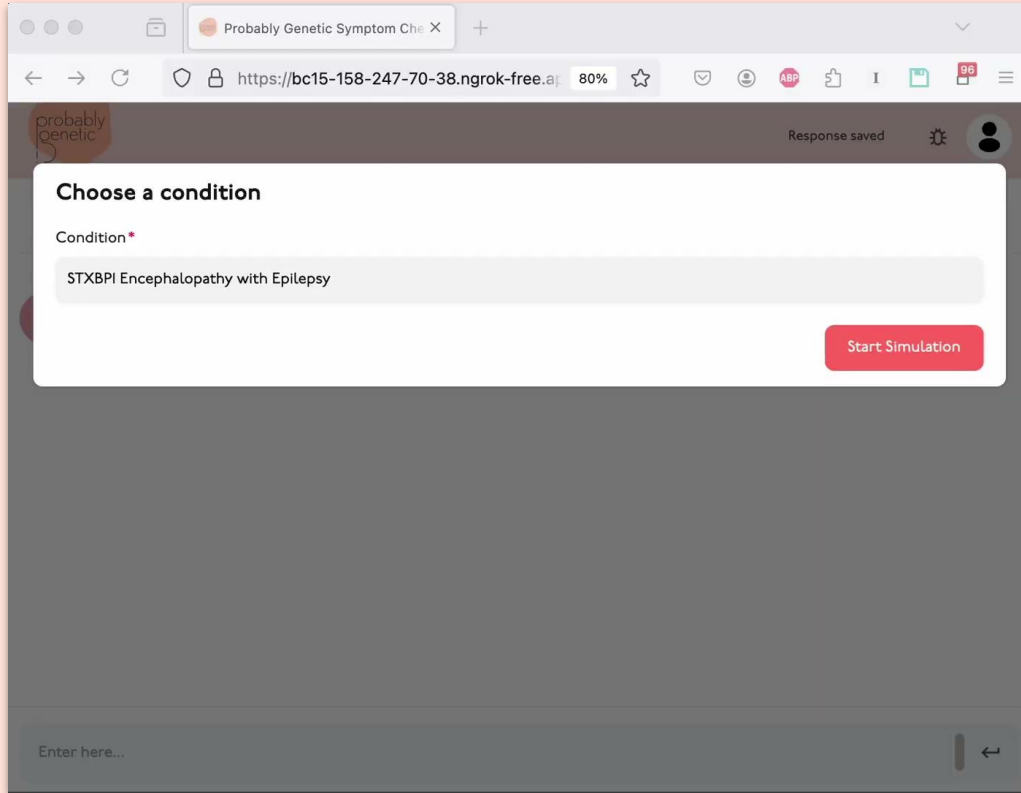
Step 3 Get Tested & Counseled

- Receive an at-home test in days with comprehensive physician and patient support



Step 4 Engage in Next Steps

- Learn about next steps via a physician consultation or direct messaging PG's patient support team



Simulated STXBP1 parent

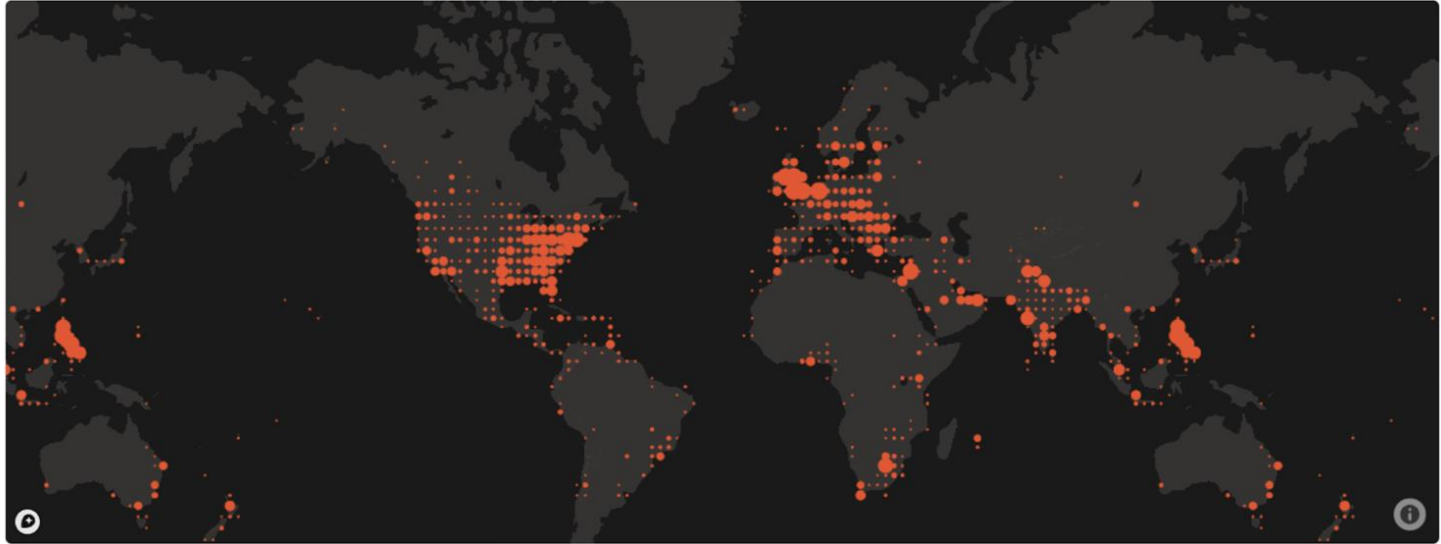
We work on many different disease areas

- ▶ Epilepsies
- ▶ Immunodeficiencies
- ▶ Neurodegenerative disorders
- ▶ Neuromuscular disorders
- ▶ Movement disorders
- ▶ Mitochondrial disorders
- ▶ Bleeding disorders

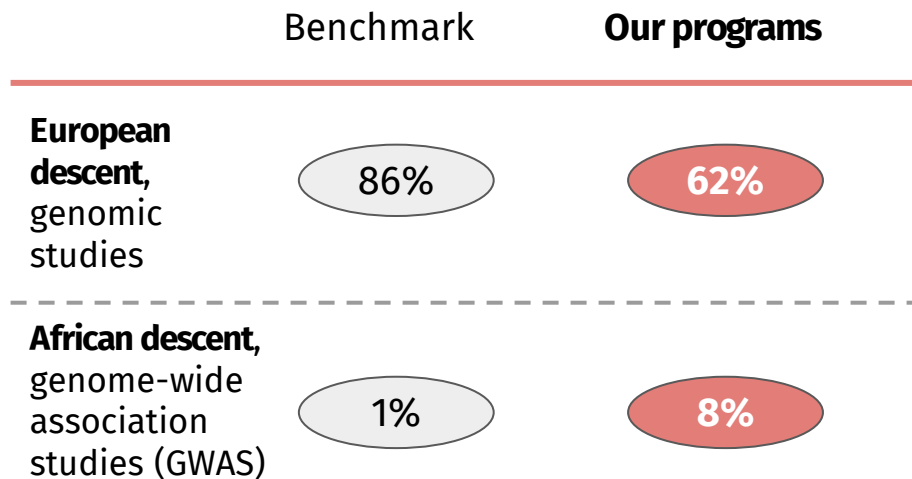
And are adding more every day.

Patients sign up globally

Locations of Symptom Checker Submissions



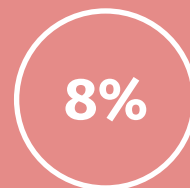
Our program participants are diverse and representative of the US



White / European



Hispanic



African American



Indigenous American



Asian



Ashkenazi Jewish



We reach cohorts with significant health disparities

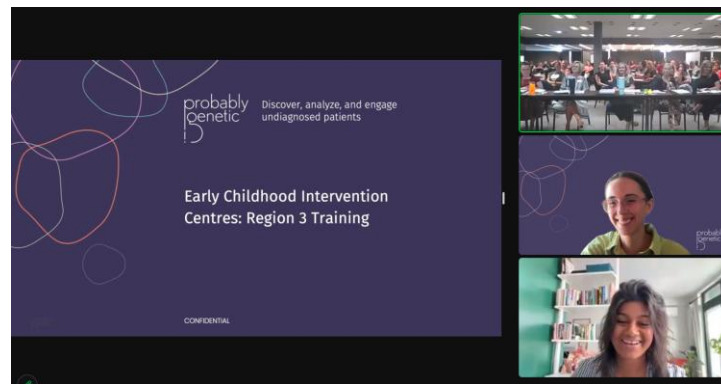
Reach in various languages...



Content viewed 7,300+ times by 4,100+ unique users

... to various communities.

Example: PG-lead early **childhood intervention program in Texas** for **Hispanic population on Medicaid**



100+ Occupational Therapists Engaged

We work hand in hand with patients to expand reach



Private Groups

Fractured Families
Admin Holly Simonton · Jan 12 · 📷

Hi! I wanted to share a great potential resource for genetic testing and other diagnostic support. Gisele Cazaudumec with a company called Probably Genetic reached out to me after seeing stories on Parents Behind The Pinwheels. We had a Zoom meeting today and she explained about a new program they're launching:... See more

Get answers for your symptoms.

symptom-checker.probablygenetic.com
Probably Genetic | Symptom Checker



Public Groups

r/BrittleBones
u/probably-genetic · 174d Join

Support for Osteogenesis Imperfecta Patients

Hi everyone,

I'm reaching out with Probably Genetic, a company whose mission is to help rare disease patients access support and resources earlier in their diagnostic journey. Excitingly, we have launched a program dedicated to Osteogenesis Imperfecta - with a patient/family-initiated platform for OI families to access **resources like clinical trial information or referrals to a live OI specialist support team.**

About Probably Genetic: Probably Genetic conducts US-based, patient-initiated programs that offer resources to individuals suspected of having rare genetic disorders. We have been working with rare disease communities since 2019, and have provided free genetic testing and counseling to thousands of patients experiencing pediatric seizures, mitochondrial disorders, dementia, and immunodeficiencies but who have not been able to access genetic testing.



Ambassador & Influencer Program

Sign In

Hey, everyone.

Have a brittle bone disorder?

Have you been diagnosed with a brittle bone disorder but then let down by the healthcare system? Get your information here to get access to our patient portal and see if you're eligible for our patient support resources like genetic testing and clinical trial connections.

Learn more

Testing available in all U.S. states except Alaska and Hawaii.

This web-tool does not provide medical advice.

By continuing, you accept our terms of service and you are aware of our privacy policy and consent.



Patient Foundations

probably genetic

Imagine seeking medical care for your child as a concerned parent

19

39

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Green Screen

Parents Behind The Pinwheels · 1-23

Imagine as a concerned parent, seeking medical care for your child, only... more

We support patient advocacy groups for free in various ways

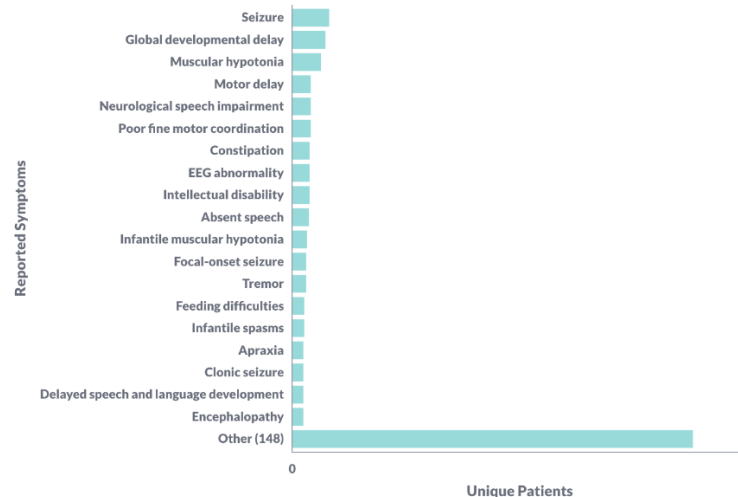
How we partner:

- ▶ **Collect information from already diagnosed patients** to evaluate AI models and share data with PAGs
- ▶ **Refer newly diagnosed patients to PAGs**
- ▶ **Help PAGs setup and manage Google Ad Grants**, bringing more traffic to PAG websites

PAG Cohort Analysis (STXBP1)

Tab 1 +


Top Reported Symptoms





probably
genetic
!

Thank you!



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