Probably Genetic Introduction

Ultragenyx Rare Disease Entrepreneur Bootcamp
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Meet our patient focused team!

I'm Caty Reid, VP of Growth & Patient Experience. My passion is driving positive outcomes for every patient we serve.

Kyle, Growth

Bringing patients into our ecosystem and crafting engaging content to help drive education and awareness.

Veronica, Patient Support

Ensuring patients have everything they need throughout their testing journey with us.

Gisele, Programs & Partnerships

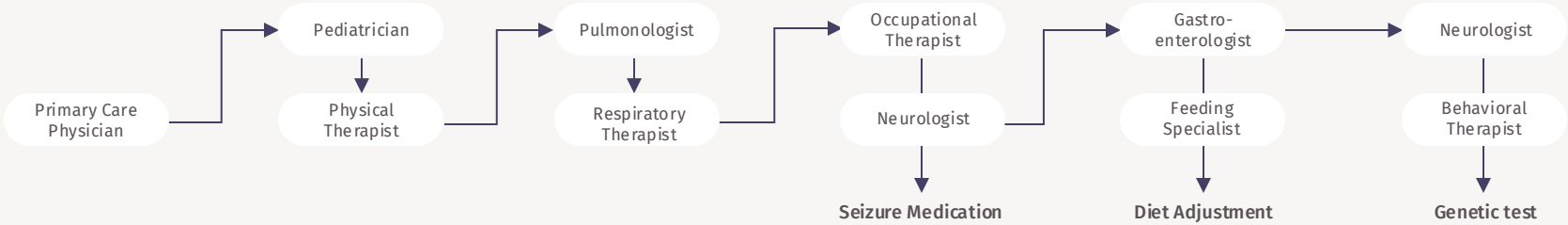
Collaborating with partners to connect the right patients to testing and support organizations following testing.

Only 38% of rare caregivers feel that their care recipient's local hospital or medical practice can handle the underlying condition their patient is challenged by.

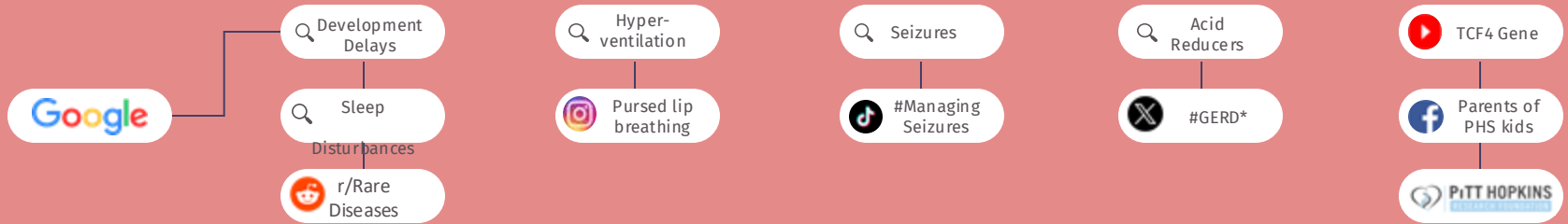
Reference: National caregivers study conducted by the National Alliance for Caregiving, in partnership with Global Genes

Undiagnosed patients look for their symptoms online.

While patients are missed by their doctors ...



... they are desperately searching for answers online



Illustrative Example for Pitt Hopkins (PHS)
*GERD: Gastroesophageal reflux disease

We find those who have been overlooked.

> My insurance didn't cover [testing]. Plus, my doctor said even if I did get tested to confirm the diagnosis, there currently is no therapies or medicine to help me, Also, the testing usually has a higher rate of incorrect results."
>
> Patient 1

> Doctors don't listen to patients ,appointment times are shrinking rapidly and they just don't have the time to really hear us as patients anymore it seems. My General practitioner told me he refused to test for anything that was a syndrome as they were not proven,and therefore not real.
>
> Patient 2

> Our main obstacle to genetic testing was finding a doctor who would submit the orders.
>
> Patient 3

> No one believed me. No doctor [would order genetic testing] and I knew doing it on my own would be too expensive for me.
>
> Patient 4

Traditional clinical solutions are accessible to a subset …

Limited clinician awareness and training
Many physicians lack sufficient training in genetics. This leads to **under recognition in genetic conditions, delayed referrals, or uncertainty of what tests to order.**

Long wait times and referral bottlenecks
Accessing a geneticist or genetic counselor often requires a referral, which can involve months long waiting periods, geographic challenges, and multiple appointments.

Cost and insurance barriers
Even when testing is available, high out-of-pocket costs or lack of insurance coverage prevent many patients from proceeding with testing, especially for:

… We are available everyone

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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

Offered free, comprehensive testing through clinical grade, HIPAA compliant free genetic testing programs

We help patients and pharma find each other.

1. ONLINE PATIENT ACQUISITION

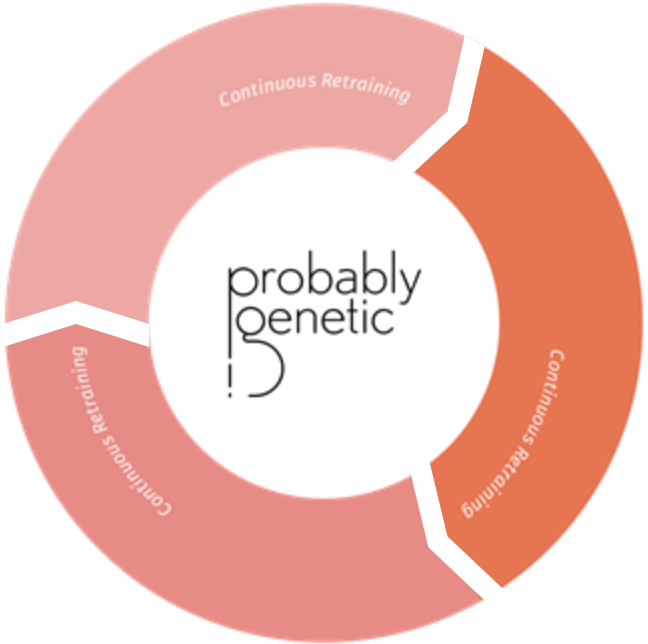
Direct access to hard-to-reach patient populations

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*

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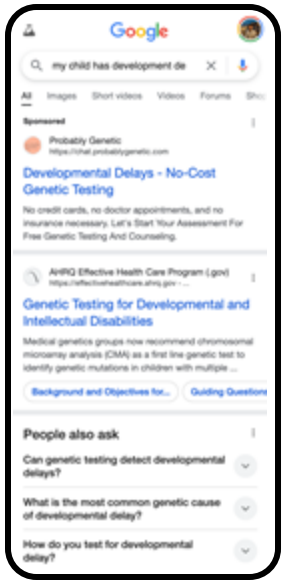
The patient journey

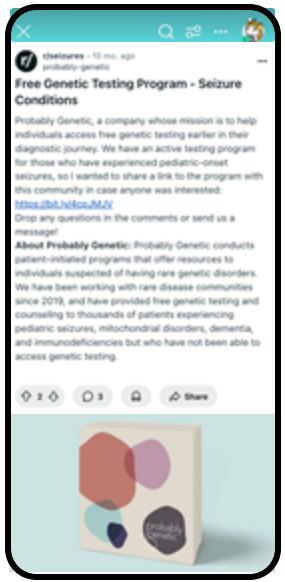
Caregivers or patients find us online.

Search

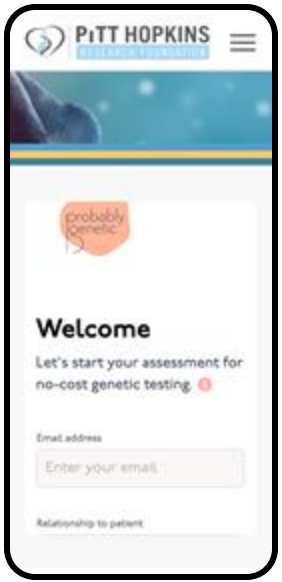


Social including private groups

Public groups

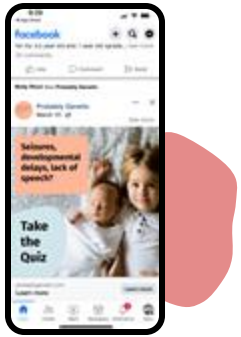


Foundations & influencers



Caregivers or patients get targeted, patients are screened & tested seamlessly.

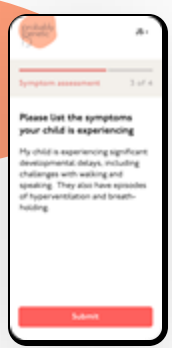
Targeting



1 Click the Link

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

Screening



2 Answer Questions

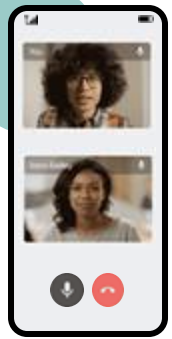
About your diagnostic journey

Testing

3 Free Testing & Counseling

Receive an at-home test and telemedicine counseling

Education



4 Learn More

Progress to the next step in your disease journey

July 11, 2022 • **Patient Story**

A Beautiful Journey with Lucy: Living With Rett Syndrome

At 14 years old, Lucy is like many teens: She loves her mom's veggie stir-fry and ski trips with her family and has had crushes on cute boys. Rett has not stopped her or her family from pursuing new experiences and possibilities. Their journey is one of hope.

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How we partner

200k+

Patients

10+

Therapeutic areas, partnering with small biotech to top-20 pharma

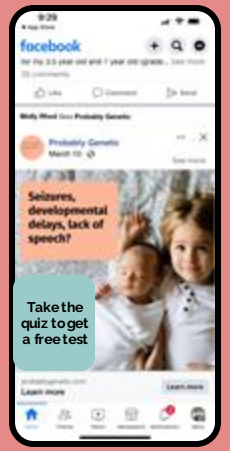
50+

PAG Partners

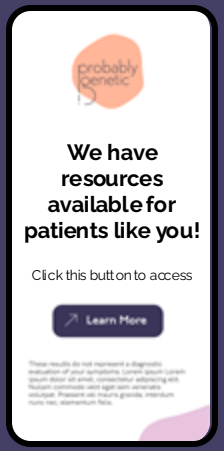
UNITED MITOCHONDRIAL DISEASE FOUNDATION

national neutropenia network

STXBP1disorders



We find patients



And help patients on their journey

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.

We prioritize advocacy partnerships.

Advocacy group partnerships

Screening tests embedded into every partner website, plus co-advertising, testing, and patient funneling.

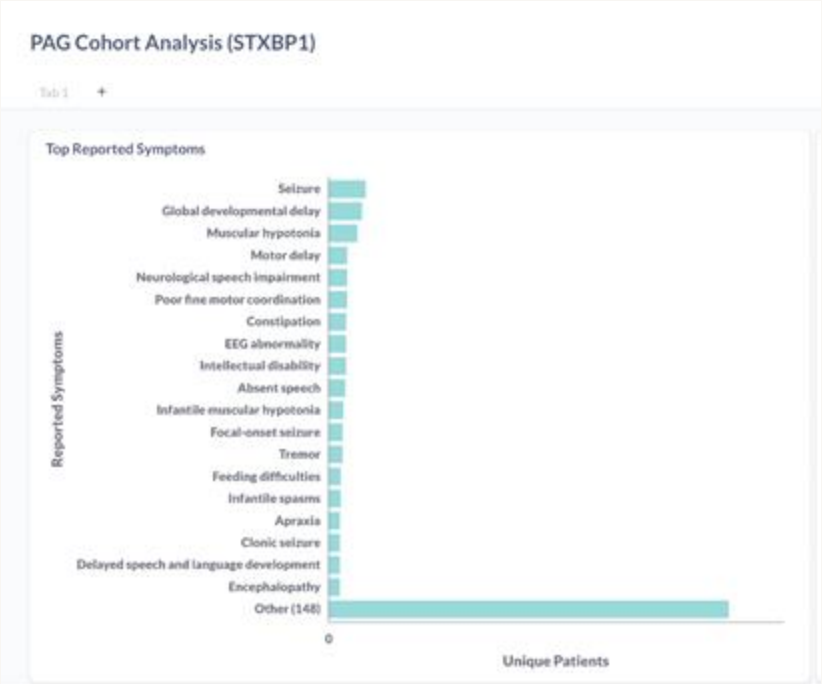
DLG4 Research Fund · CHAMP1 · KDVS · ARRE · WWOX Foundation · IFCR International Foundation for CDKL5 Research · ADNPkids · SLC6A1 Connect · MED13L Foundation · FAM177A1 Research Fund · The Rare Bella Foundation · Cure KCNH1 Foundation · CURE RAGHAV · CTNNB1 · the SHANK2 foundation

+ more

Partner website example with the United Mitochondrial Disease Foundation.

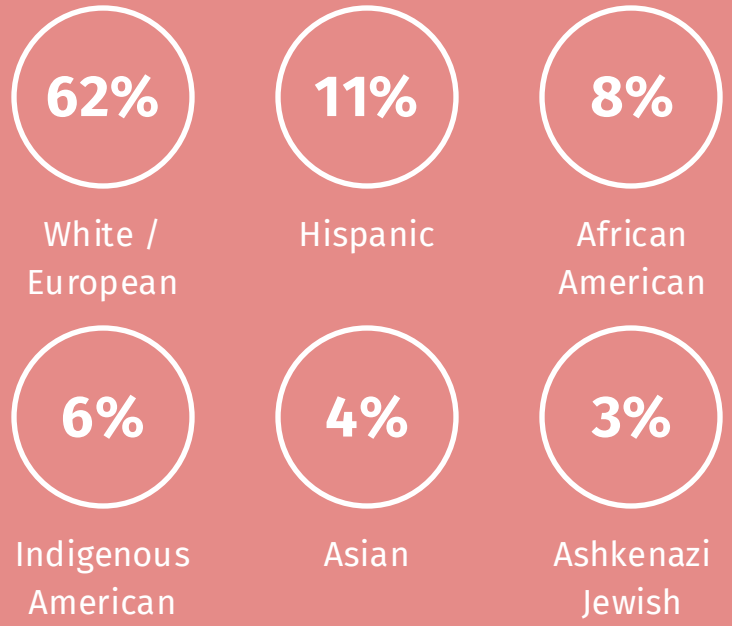
We support our patient advocacy partners.

- **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
- **Create disease education content,** bringing more visibility and traffic to PAG initiatives



Our program participants are diverse.

	Benchmark	**Our programs**
European descent, genomic studies	86%	62%
African descent, genome-wide association studies (GWAS)	1%	8%



Fatumo S, Chikowore T, Choudhury A, Ayub M, Martin AR, Kuchenbaecker K. A roadmap to increase diversity in genomic studies. Nat Med. 2022 Feb;28(2):243-250

We reach those with significant health disparities.

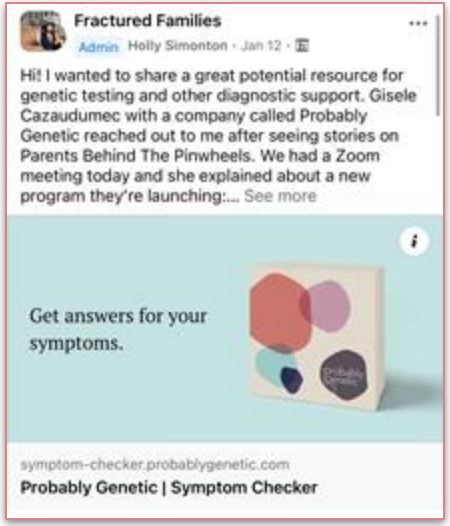
Example: CAN partnership to reach patients being supported by caregivers in US counties with **>20% of the population living at or below poverty level** and with a **<40% white racial demographic.**

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

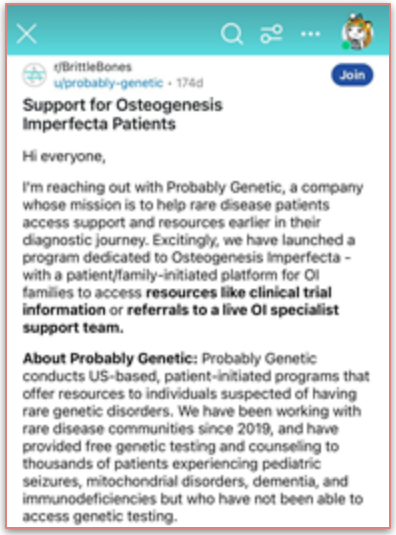
100+ Occupational Therapists Engaged

We develop content with patients directly.

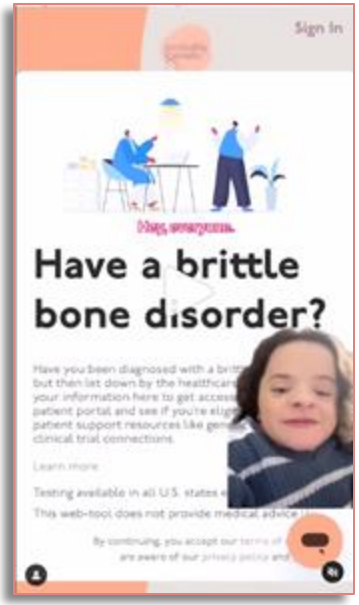
Private groups



Public groups



Ambassador & influencer program



Patient foundations

Thank you!

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Archive

Lukas Lange, PhD | CEO
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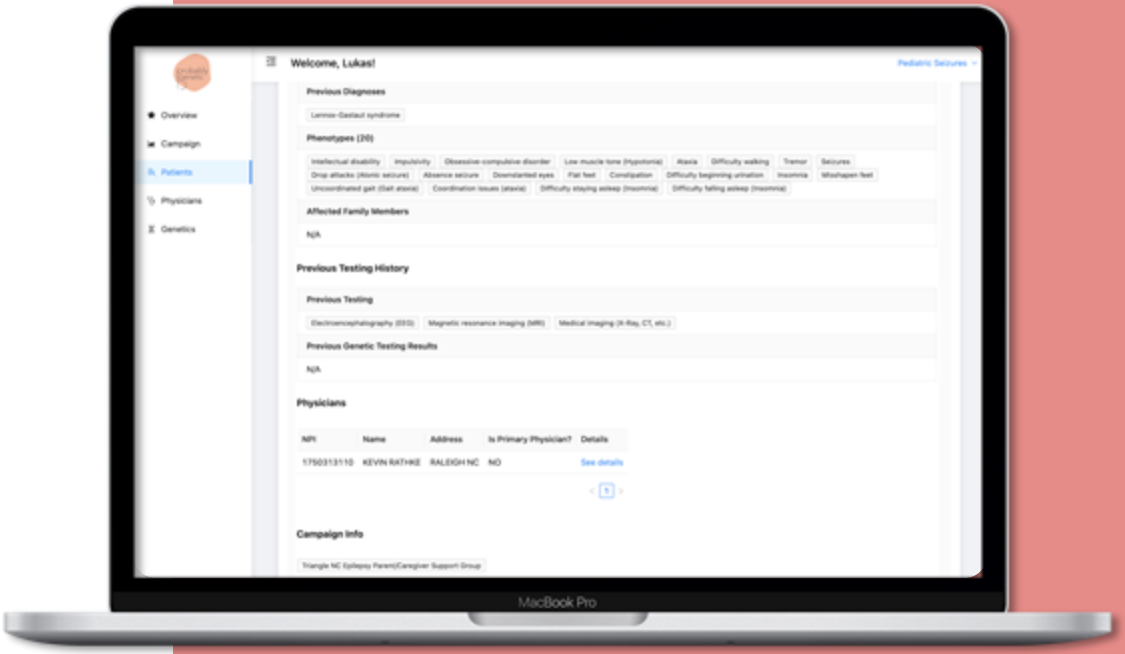
PRODUCT

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.



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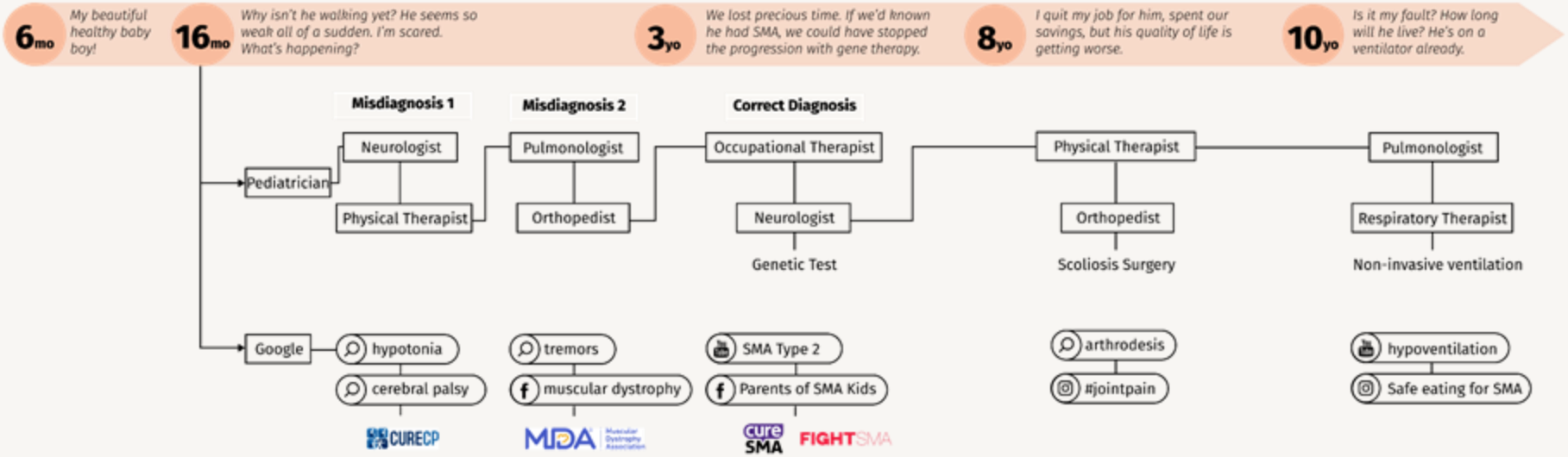
Patients are sick for years before getting diagnosed

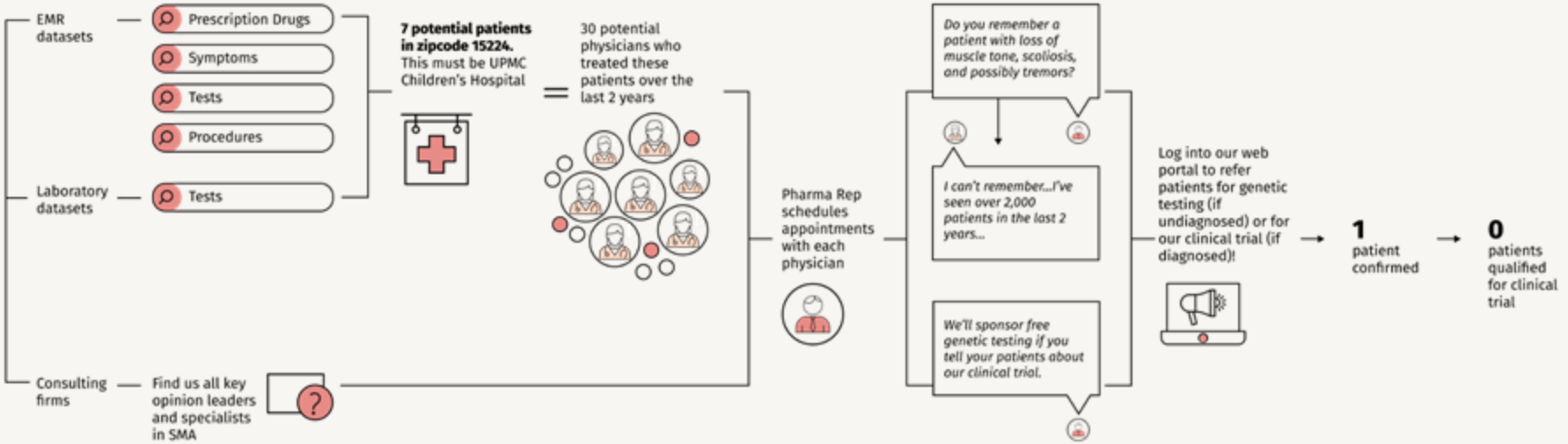
Meet Logan & his mom Danielle.
This is his journey getting diagnosed with Spinal Muscular Atrophy (SMA) Type 2.



- **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.