probably genetic

Finding patients

Ultragenyx Rare Disease Entrepreneur Bootcamp
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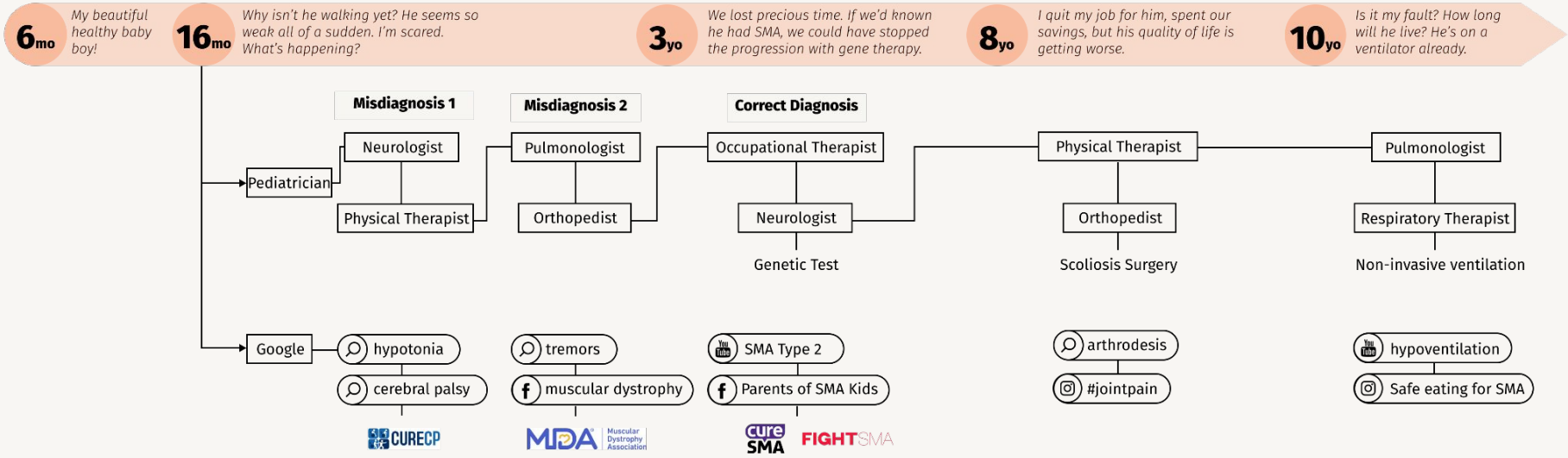
Genetic disease patients are sick for years before getting diagnosed

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.

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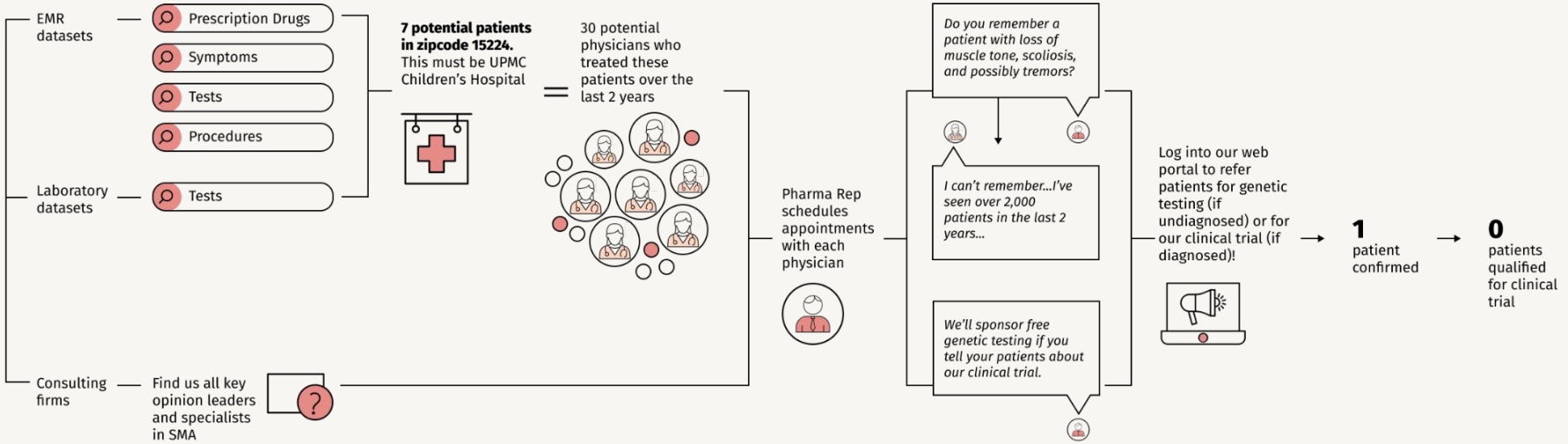
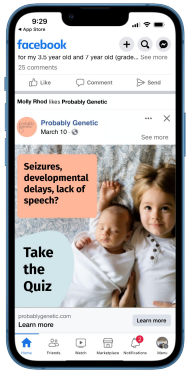
And pharma companies can't find them

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



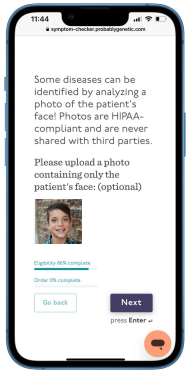
- Data lives across different EMRs, specialists, and labs, which makes it **extremely hard to identify patients**.
- Because of regulatory restrictions, **pharma companies waste time and money trying to find physicians treating target patients.**
- **Targeting physicians is an ineffective** way to get patients into trials and onto treatments.

We find patients online using AI and at-home testing



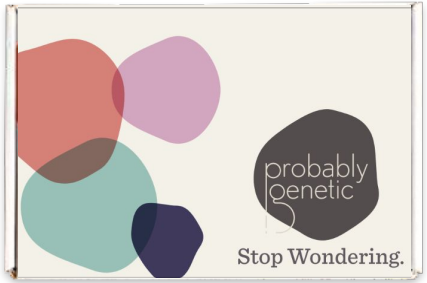
Awareness

Patients find us online



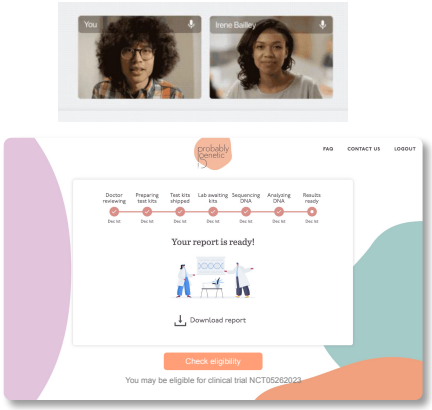
AI

Platform identifies undiagnosed patients



Free testing

Patients are tested remotely



Results

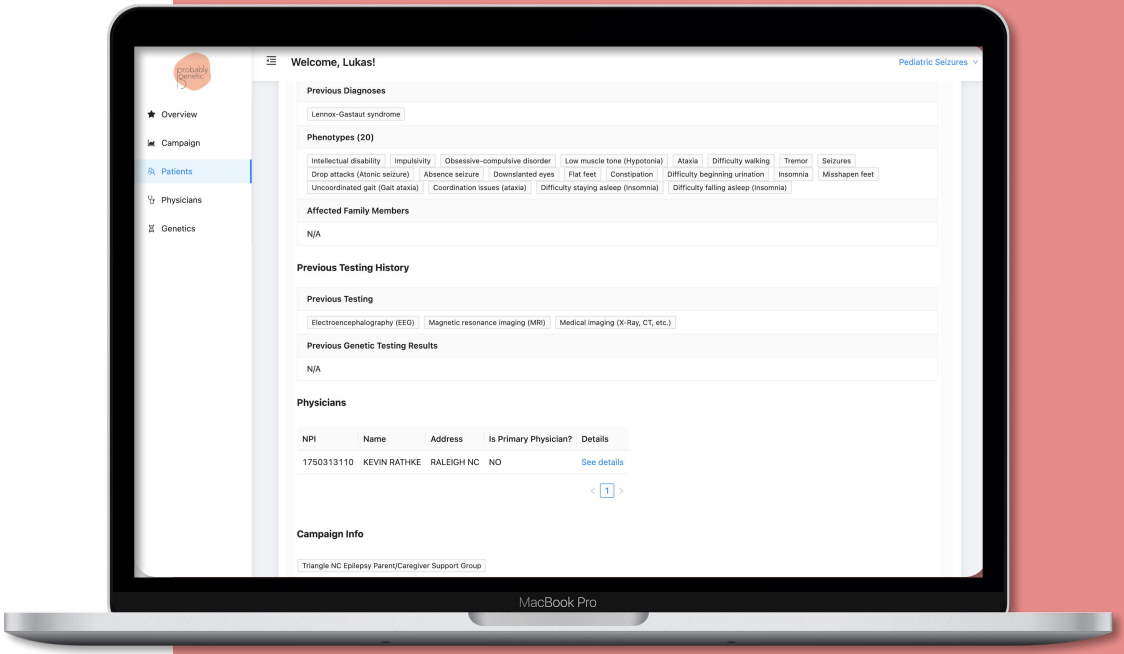
Patients receive results

Pharma gets data

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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We work on many different disease areas

- Epilepsies
- Immunodeficiencies
- Neurodegenerative disorders
- Neuromuscular disorders
- Movement disorders
- Mitochondrial disorders
- Brittle bone disorders

And are adding more every day.

Case Study #1: Ultra Rare Monogenic Disease

Expanded known patient population of Ultra-Rare Disease by 10%

Scenario

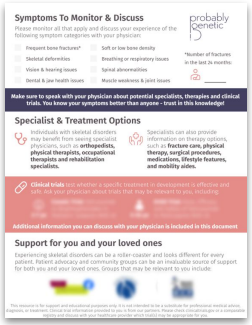
CHALLENGE	Ultra-rare monogenic disease (1 in 5 million incidence rate)
STAGE	Pre-Commercial Stage
TIMELINE	Data generation for 12+ months

Outcomes

PATIENTS	Increased known patient population by 10%
HCP DATA	Found 200+ physicians treating phenotypically similar patients

Case Study #2: Bone disorder

Exceeded 3-month goal for clinical trial patient finding **by 3x within 2 months**



Scenario

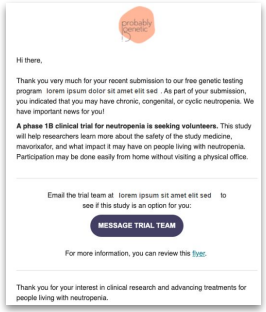
CHALLENGE	Targeted towards patients diagnosed with rare genetic bone disorder
STAGE	Recruitment for Phase 2 clinical trial
TIMELINE	3-months

Outcomes

PATIENT IMPACT	Reached 3-month goal (10 patients consented) within 3 weeks of program launch
	Exceeded 3-month goal by 3x within 2 months
	64% of target patients came from advocate awareness posts

Case Study #3: Immunodeficiency

40+ patients directly engaged with clinical trial recruitment process



Hi there,

Thank you very much for your recent submission to our free genetic testing program lorem ipsum dolor sit amet elit sed . As part of your submission, you indicated that you may have chronic, congenital, or cyclic neutropenia. We have important news for you!

A phase 1B clinical trial for neutropenia is seeking volunteers. This study will help researchers learn more about the safety of the study medicine, mavorixafor, and what impact it may have on people living with neutropenia. Participation may be done easily from home without visiting a physical office.

Email the trial team at lorem ipsum sit amet elit sed to see if this study is an option for you:

MESSAGE TRIAL TEAM

For more information, you can review this flyer.

Thank you for your interest in clinical research and advancing treatments for people living with neutropenia.

Scenario

CHALLENGE Targeted towards patients diagnosed with specific immunodeficiency

STAGE Recruitment for Phase 1/2 clinical trial

Outcomes

PATIENT IMPACT

44% open rate for recruitment outreach

40+ patients directly engaged with clinical trial recruitment process

We reach patients via many channels

Advocacy Group Partnerships

Our technology is embedded into every partner website. We build network effects by co-advertising, testing, and funneling patients into appropriate advocacy groups.

CTNNB1 · the SHANK2 foundation · KDVS · Cure KCNH1 · Ruzzo-Lo Foundation · SynGAP Research Fund · ARRE · STXBP1disorders · SLC6A1 Connect · MED13L Foundation · WWOX Foundation · CURE MITO Foundation · PITT HOPKINS · CHAMP1 · FAM177A1 Research Fund · PBD project

+ more

Product-led Referrals

Built-in referral features drive organic growth among networks of undiagnosed patients and their families.

Ambassador Program

We partner with active community members to educate and refer other patients.

Influencer Partnerships

We partner with patient Instagram influencers, Youtube vloggers, and Tiktok stars.

Paid Advertising

We developed a phenotype-based targeting platform to reach undiagnosed individuals through search and social.

Unlocking New Resources for Osteogenesis Imperfecta

Patients sign up globally

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Thank you!

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