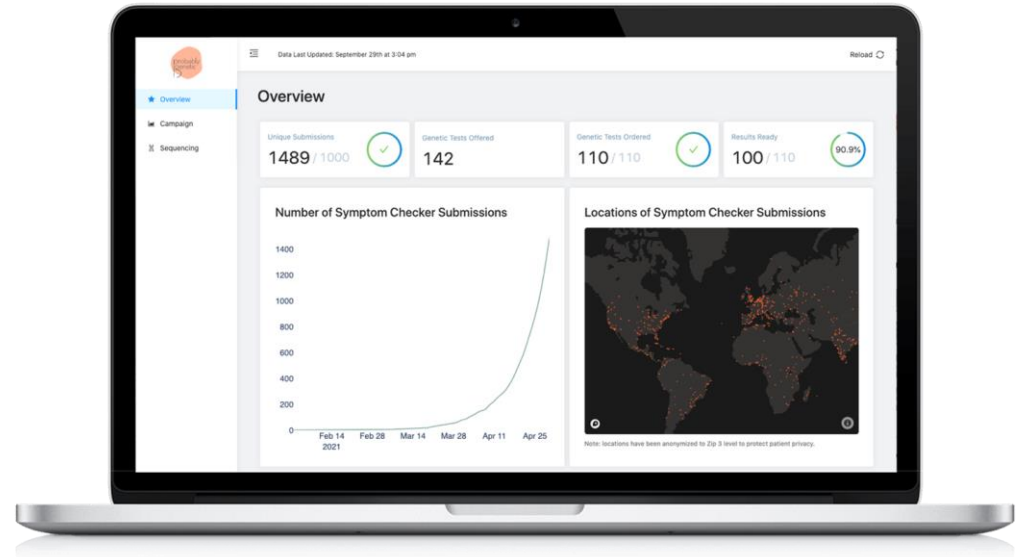


Finding undiagnosed rare disease patients

Ultragenyx Bootcamp, 2023



Lukas Lange, PhD
CEO
lukas@probablygenetic.com

Platform to commercialize rare disease treatments



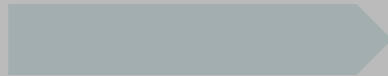
Platform to commercialize rare disease treatments



Patient finding



Trial recruitment



Treatment access

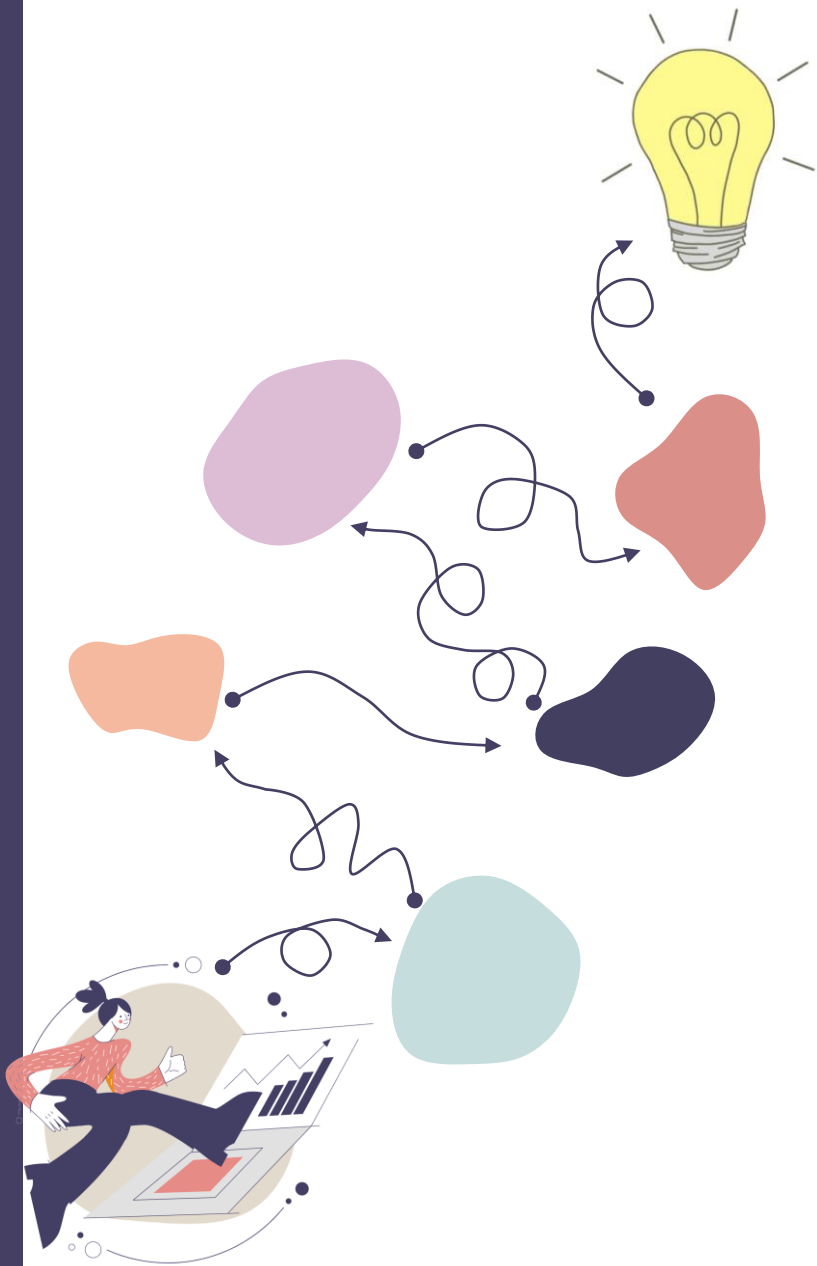


Real-world evidence



R&D data





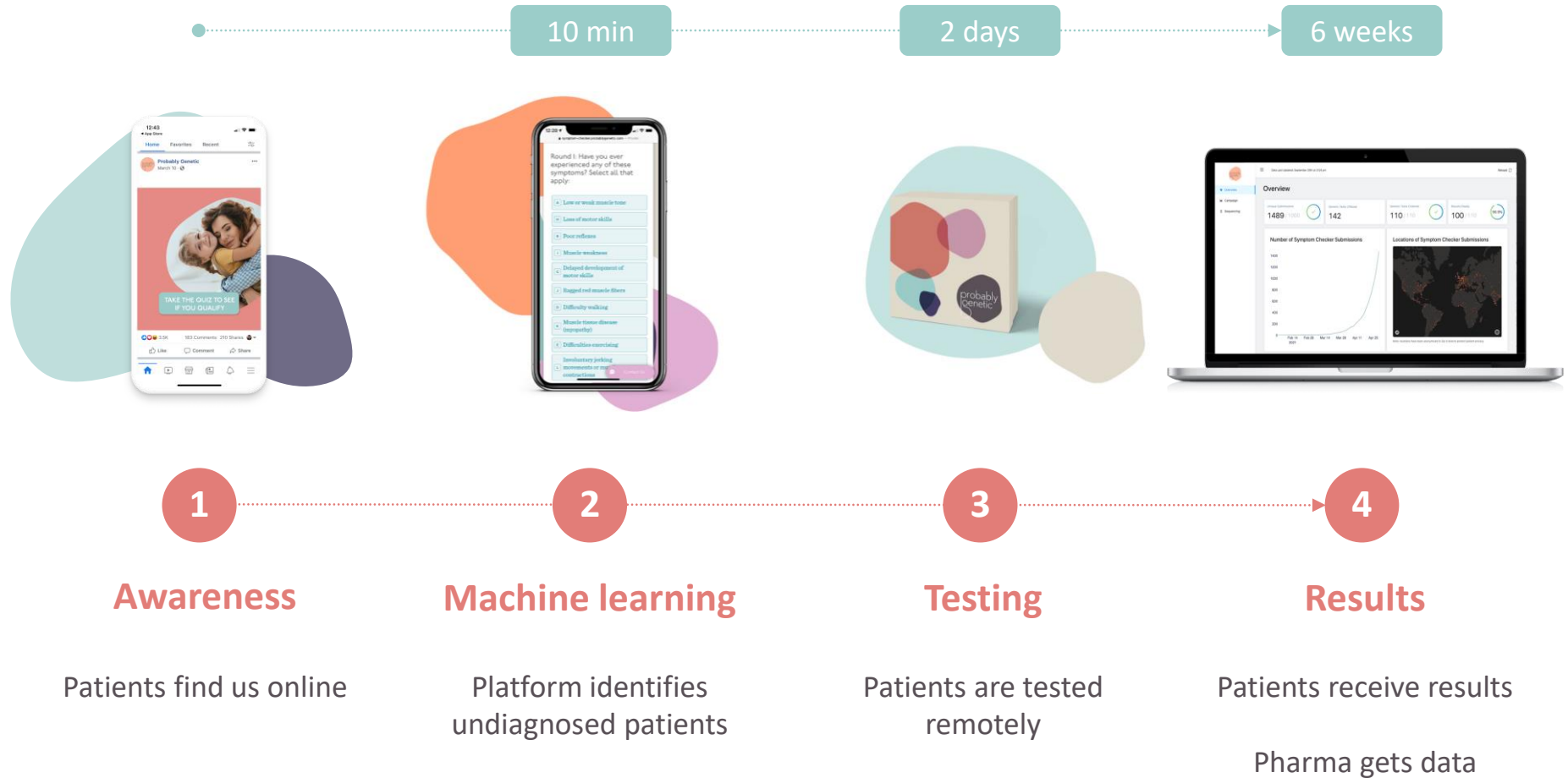
How we diagnose patients is broken

- 8 years
- 3 misdiagnoses
- No trial recruitment & treatment access

🔍 Daughter has autism, seizures, is losing her vision, can't climb stairs ✕



We find patients online using machine learning and at-home testing





1

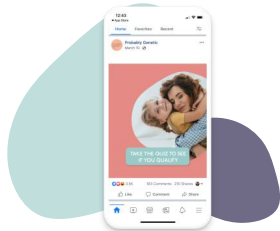
Awareness

How???



- Seizures
- Developmental delay
- Autism

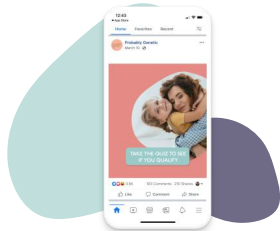
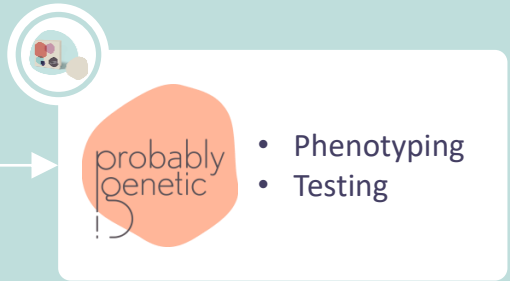
???



1

Awareness

We built an invisible network of patient advocacy groups



1

Awareness

We built an invisible network of patient advocacy groups



- Seizures
- Developmental delay
- Autism

???



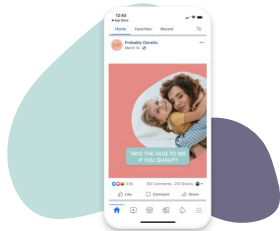
SLC6A1 ???

SLC6A1 !!!



probably genetic

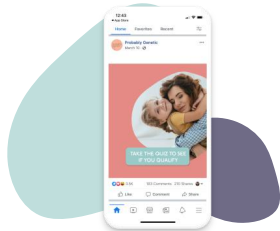
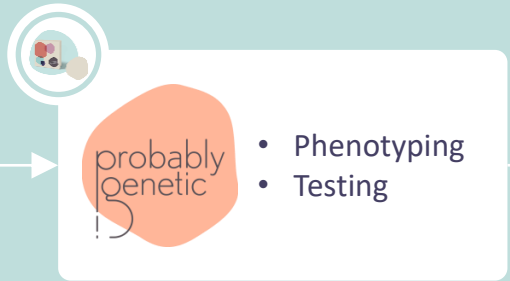
- Phenotyping
- Testing



1

Awareness

We built an invisible network of patient advocacy groups



1

Awareness

We built an invisible network of patient advocacy groups


The Koolen-de Vries Syndrome Foundation has partnered with Probably Genetic to increase access to genetic testing within our community. Probably Genetic's no-cost genetic testing program for pediatric epilepsy disorders is patient-initiated and includes genetic counseling to explain any questions or findings. If you or a loved one are experiencing symptoms associated with pediatric epilepsy or developmental disorders, you can apply to the program by completing the short quiz below. Eligible individuals receive a sample collection kit mailed directly to their home, with a return pickup service included. Results are delivered 6-8 weeks after samples are received at the lab, with genetic counseling conducted virtually or over the phone.

Probably Genetic Symptom Checker

[Sign In](#)



Is our genetic test right for you? Check eligibility.



Take our quiz to see if you are eligible for a **FREE** genetic test. No credit cards, no doctor appointments, and no insurance necessary. All from the comfort of your home.

Note: testing is available in all US states except for NY.

[Learn more](#)

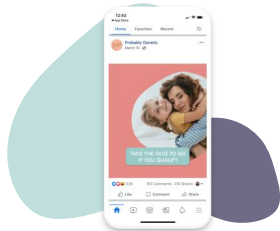
[Para español](#)

Testing available in all U.S. states except NY
This web-tool does not provide medical advice ⓘ

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

Check Eligibility

press Enter ↵



1

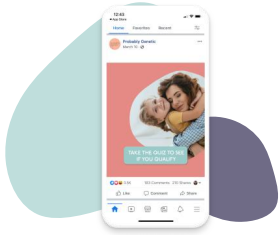
Awareness

We built an invisible network of patient advocacy groups

confidential



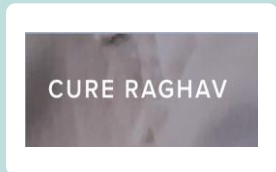
>\$150k monthly ad spend in network



1

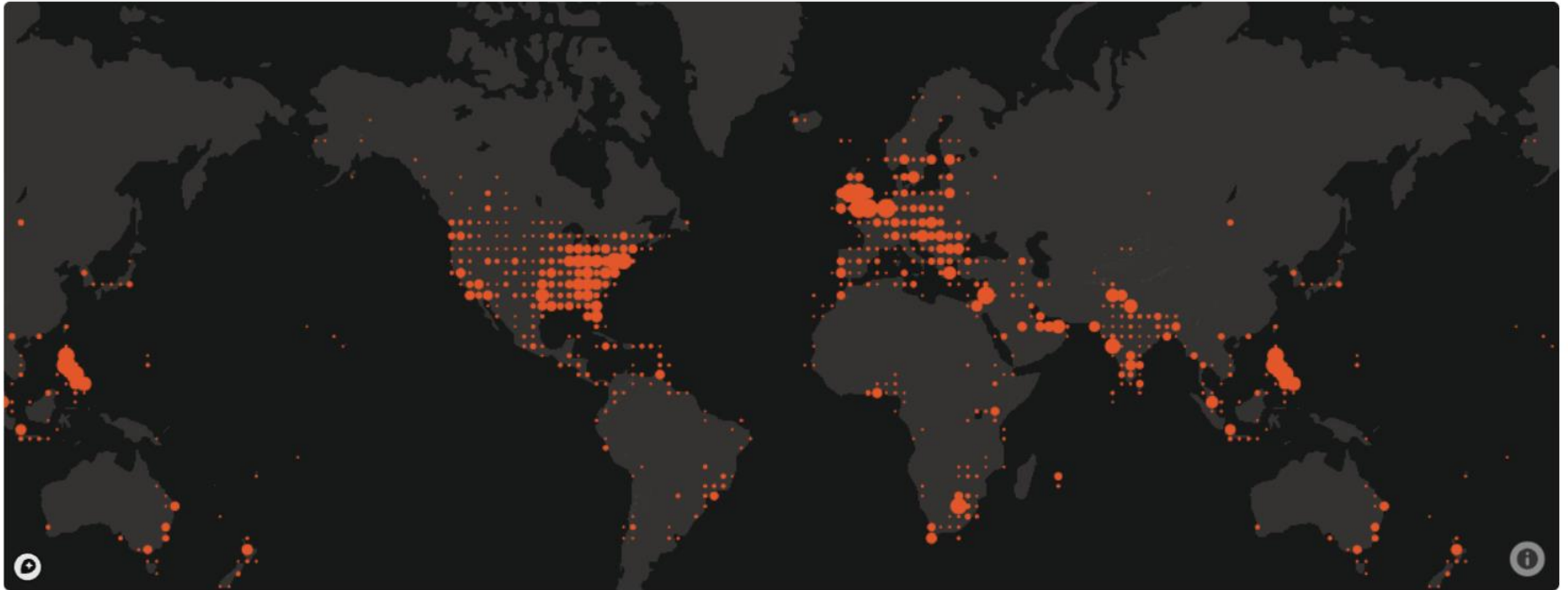
Awareness

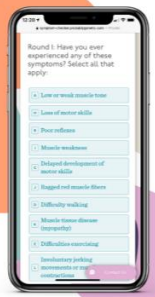
We built an invisible network of patient advocacy groups



Thousands of patients sign up every month, globally

Locations of Symptom Checker Submissions





2

Machine learning

Live learning system identifies undiagnosed patients

Text

- ✓ Phenotypic terms
- ✓ Family pedigree
- ✓ Medical history
- ✓ Meta data
- ✓ Lab reports
- EMR integrations

Photos

- ✓ Face
- Skin
- Hands
- Feet

Videos

- Seizures
- Movement
- Behavior

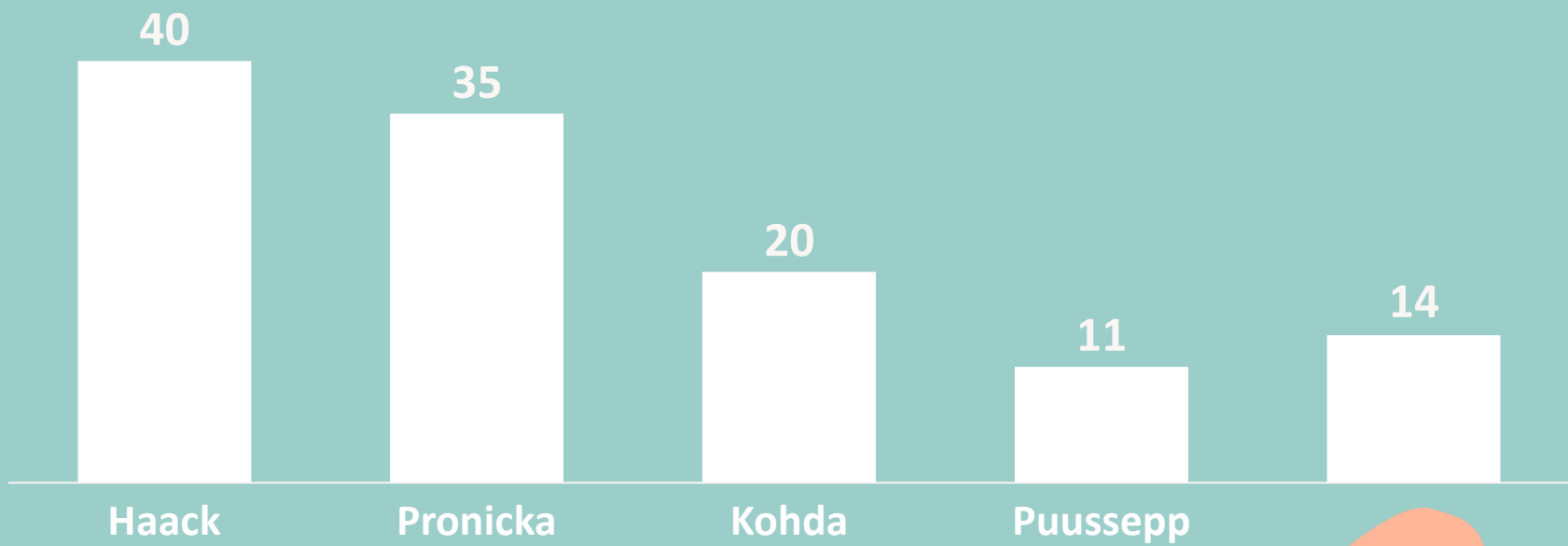
Audio

- Voice
- Hearing



Mito program competitive with published studies

% of patients tested confirmed to have a mitochondrial disorder



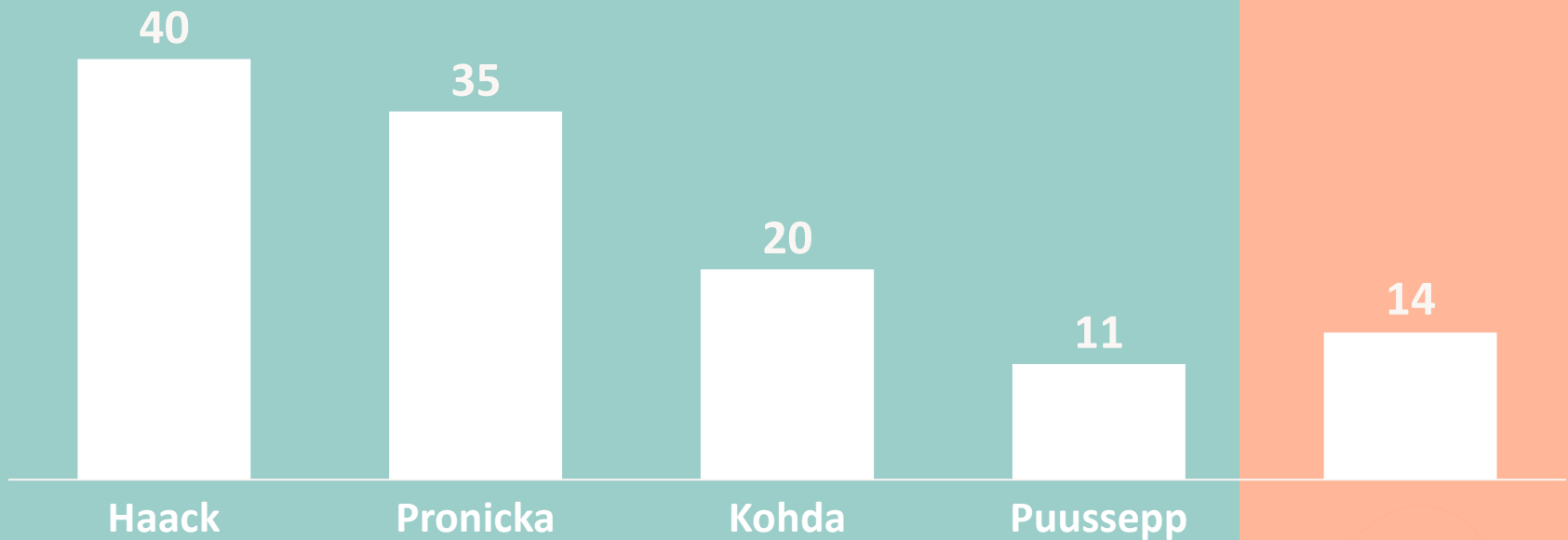
probably genetic



Clinically diagnosed with mitochondrial disease prior to testing by specialists

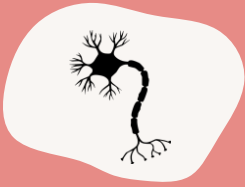
Selected by machine learning algorithm

% of patients tested confirmed to have a mitochondrial disorder





Immuno-
deficiencies



Seizure
disorders



Movement
disorders



Mito



Dementias



To be
announced



Trying to find patients? Sign up for free

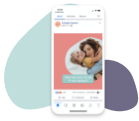


Lukas Lange, CEO
lukas@probablygenetic.com



Patient Experience

1



Patient acquisition: patients find us online via **paid** and **organic** traffic



free mito testing

About 93,200,000 results (0.50 seconds)

Ad · <https://www.maybemito.com/>

MayBeMito: No-Cost Testing - Mitochondrial Disease Info
 Learn More About **Mitochondrial** Disease, Including Epidemiology, Prevalence, And Symptoms. Discover A Path Forward With No-Cost Genetic **Testing For Mito** In Your Patients. Sign Up For Updates. Symptom Information. No-Cost Genetic **Tests**.

Discover No-Cost Testing
 Review No-Cost Genetic Tests. Know What Is Best For Patients.

Mitochondrial Disease
 Learn About Types And Prevalence. View Symptoms And Misdiagnoses.

Uncover HCP Resources
 How To Connect With Other Experts. Explore Additional Organizations.

Explore Info On TK2d
 Know Thymidine Kinase 2 Deficiency. View Early And Late Onset Symptoms.

<https://www.genomeweb.com> > ... > Business News

Free Mitochondrial Disease Genetic Testing Program ...
 Feb 15, 2022 — The UMDF Pilot Genetic **Testing** Project is intended to help patients get a confirmed genetic diagnosis of **mitochondrial** disease and is available ...

<https://mito.probablygenetic.com>

Probably Genetic | Free DNA Testing for Mitochondrial Diseases
Free genetic **testing** for **mitochondrial** disease. Answer our short quiz to see if you qualify for our **free**, at-home, whole exome sequencing **test**.

<https://www.umdff.org> > research > mitochondrial-genet...

Mitochondrial Genetic Testing - UMDF
 "We know the diagnostic journey for a **mitochondrial** disease patient is often long and difficult. By offering a no-cost genetic **testing** program, UMDF aims to ...

Paid ad

Organic article

PG website

Advocacy group

1



Patient acquisition: patients find us online via **paid** and **organic** traffic



Paid

Organic

Probably Genetic
Sponsored · 🌐

Probably Genetic offers free genetic testing and counseling for those that qualify. Take a short quiz to see if you are eligible.

Seizures, autism, and breathing abnormalities are symptoms of Pitt-Hopkins syndrome.

Take the quiz to see if your child qualifies for free genetic testing.

symptom-checker.probablygen...
Take the quiz today
Free genetic testing for pe... **Apply now**

probablygenetic
Sponsored

The Probably Genetic test analyzes genes known to cause frontotemporal dementia (FTD).

TAKE THE QUIZ TO SEE IF YOU QUALIFY FOR FREE TESTING

Frontotemporal dementia can cause changes in behavior and problems with language. Probably Genetic offers free genetic testing and counseling for those that qualify. Take a short quiz to see if you are eligible.

Apply now

jayceeandmore

OUR FAMILY TRIO TESTS FROM @PROBABLYGENETIC ARE HERE!!

probably genetic
Stop Wondering.

These are not sponsored stories, I'm just updating on my health journey.