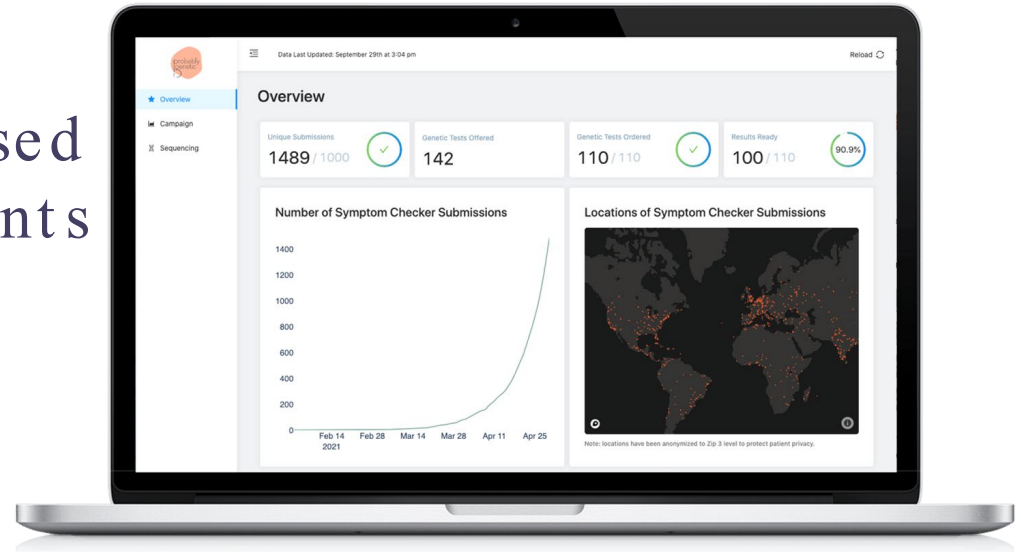


# Finding undiagnosed rare disease patients

Ultragenyx Bootcamp, 2023



Lukas Lange, PhD  
CEO  
[lukas@probablygenetic.com](mailto:lukas@probablygenetic.com)

# Platform to commercialize rare disease treatments



# Platform to commercialize rare disease treatments



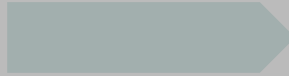
Patient finding



Trial recruitment



Treatment access

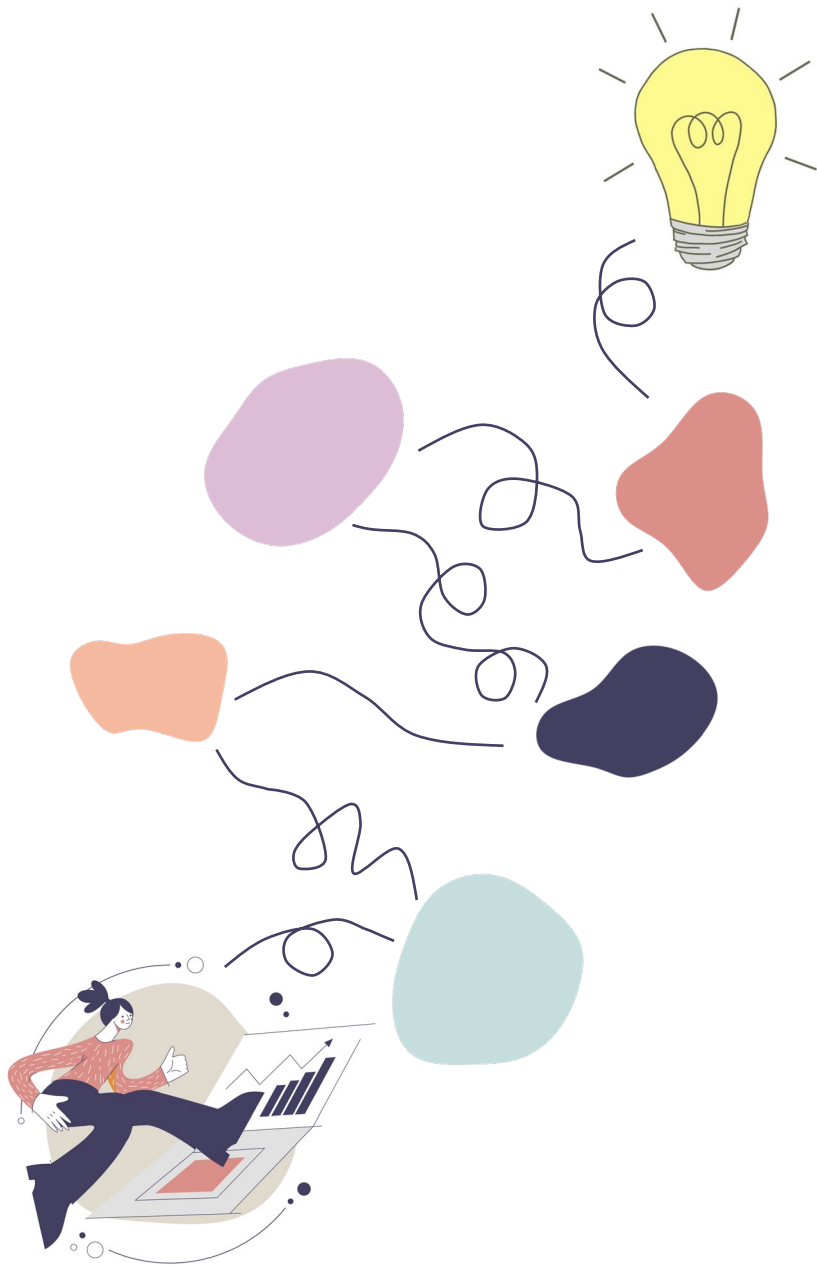


Real-world evidence



R&D data





## How we diagnose patients is broken

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

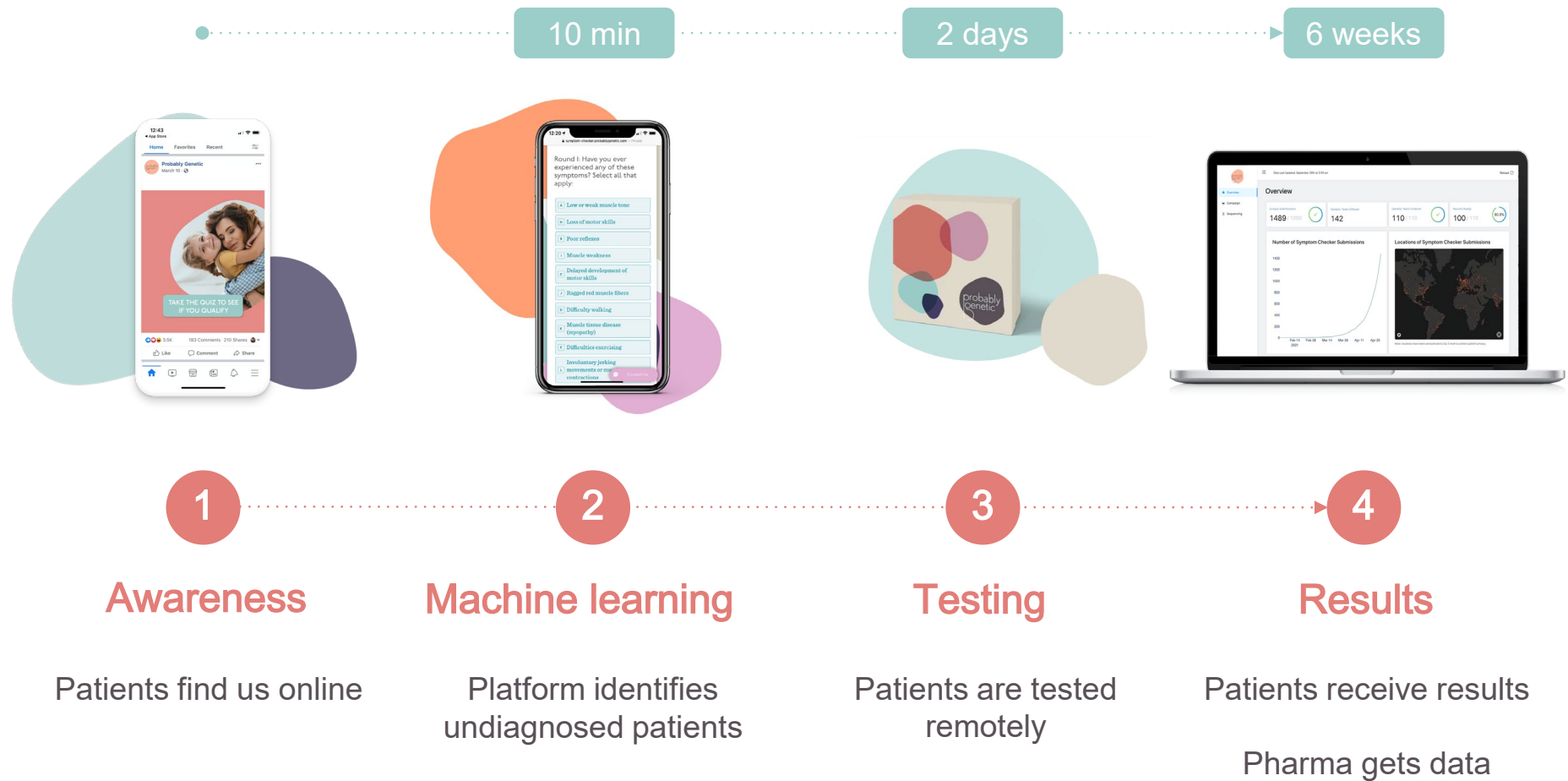
🔍 Daughter has autism, seizures, is losing her vision, can't climb stairs ✕

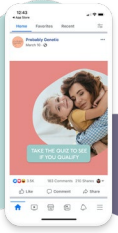
Google



YouTube

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





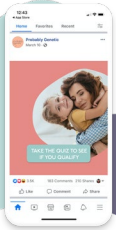
1

Awareness

How???



- Seizures
- Developmental delay
- Autism

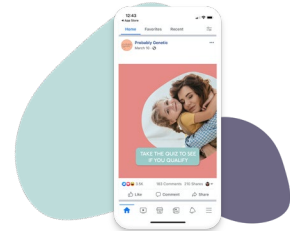
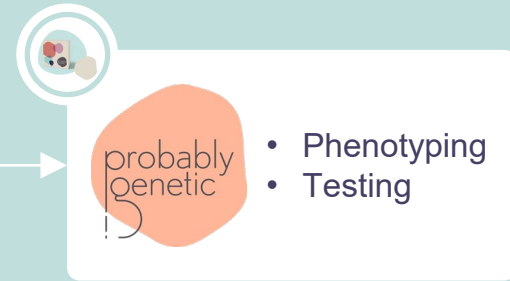


1

## 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

??  
?

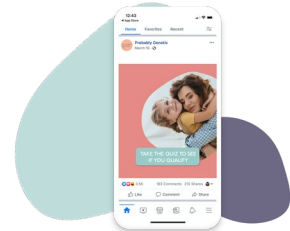


SLC6A1  
???

SLC6A1  
!!!



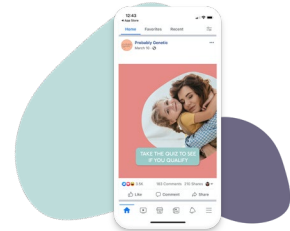
- Phenotyping
- Testing



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](#)

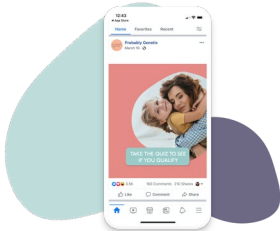
[Para español](#)

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

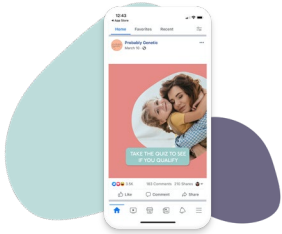
## Awareness

We built an invisible network of patient advocacy groups

confidential



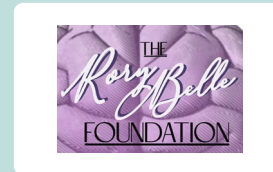
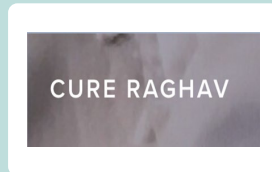
>\$150k monthly  
ad spend  
in network



1

## Awareness

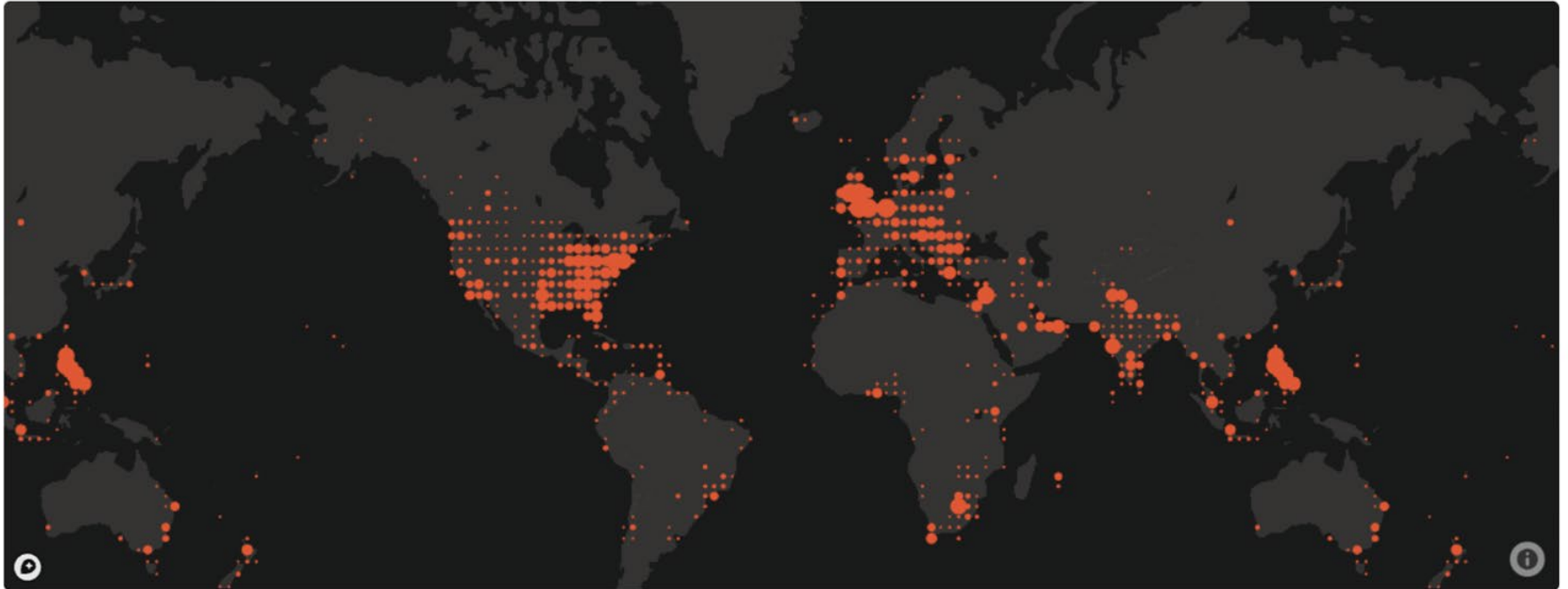
We built an invisible  
network of patient  
advocacy groups

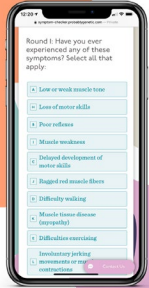


# Thousands of patients sign up every month, globally

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## Locations of Symptom Checker Submissions





2

## Machine learning

Live learning system identifies undiagnosed patients

Text

Photos

Videos

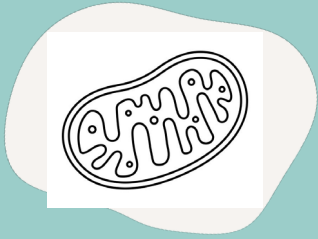
Audio

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

- ✓ Face
- Skin
- Hands
- Feet

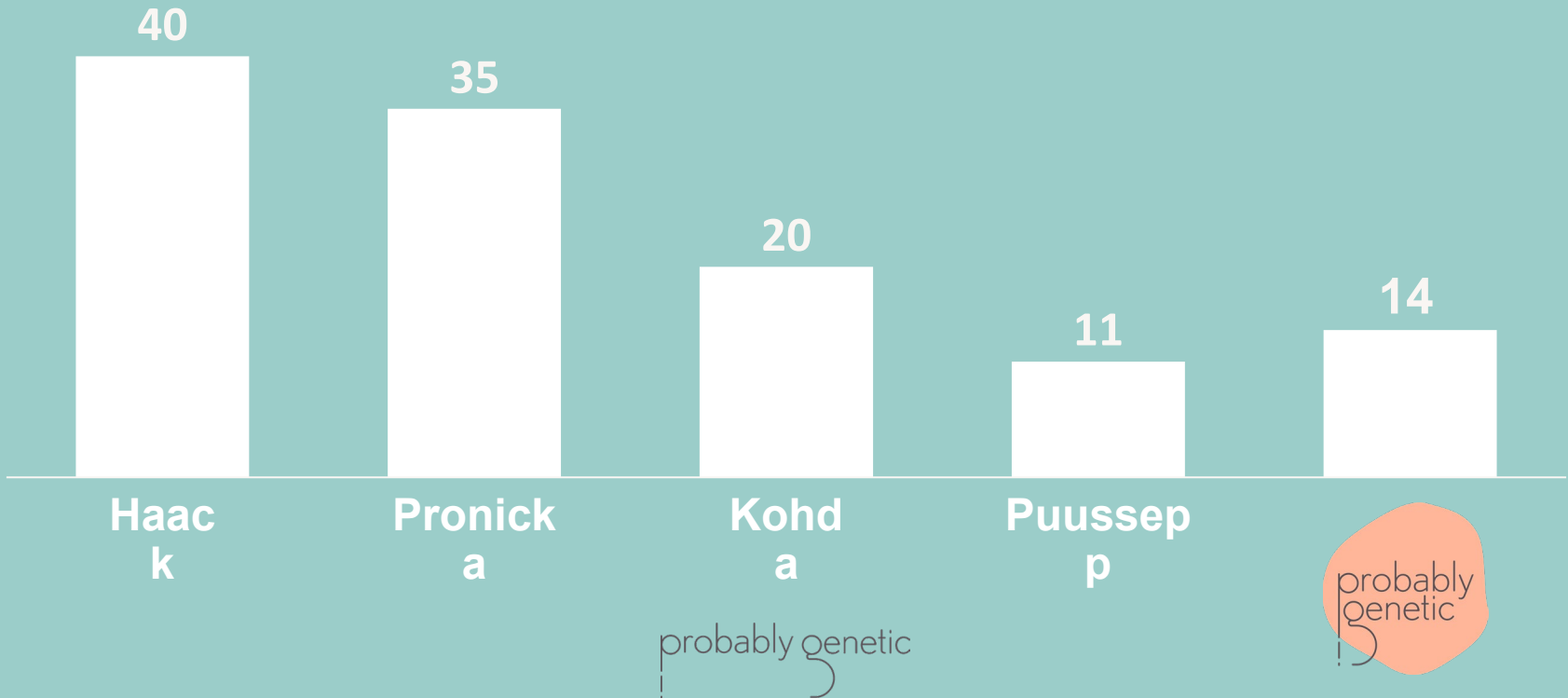
- Seizures
- Movement
- Behavior

- Voice
- Hearing



## Mito program competitive with published studies

% of patients tested confirmed to have a mitochondrial disorder

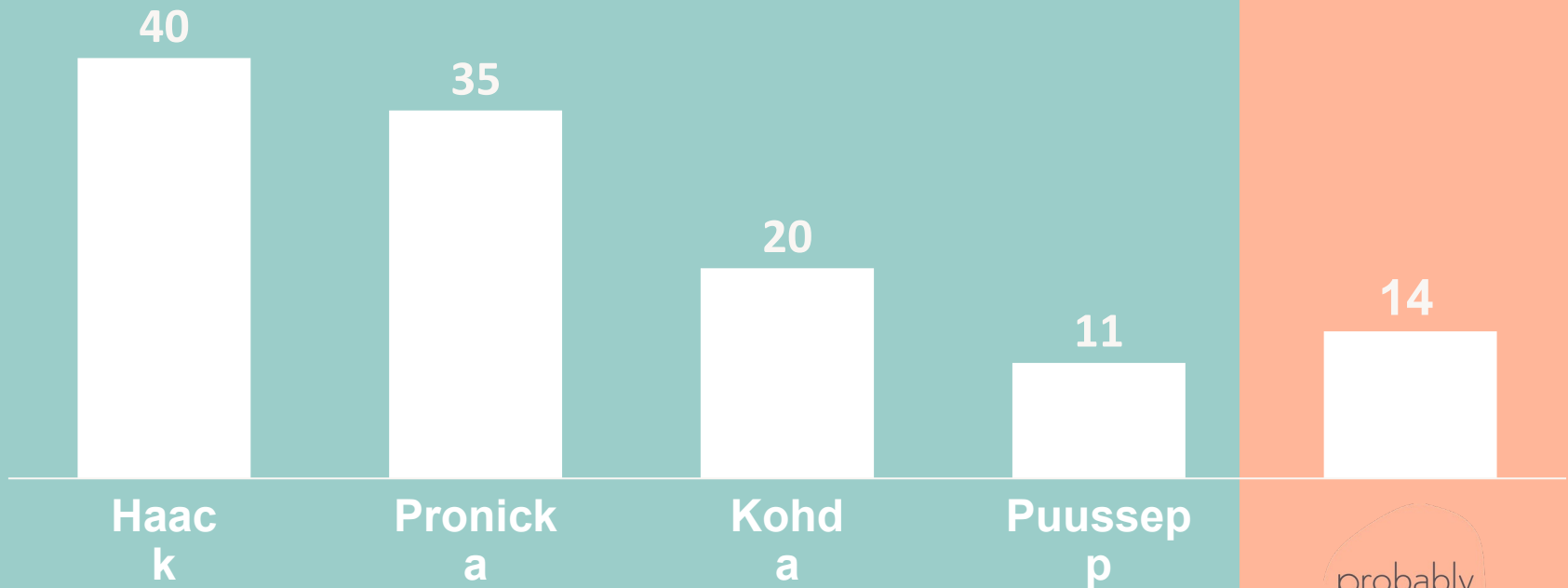




# Clinically diagnosed with mitochondrial disease prior to testing by specialists

Selected by machine learning algorithm

% of patients tested confirmed to have a mitochondrial disorder



confidential



Immuno-  
deficiencies



Seizure  
disorders



Movement  
disorders



Mito



Metabolic  
disorders



Dementias



Encephalitis

...

To be  
announced

probably genetic



**Trying to find patients?  
Send us an email**

Lukas Lange, CEO  
[lukas@probablygenetic.com](mailto:lukas@probablygenetic.com)

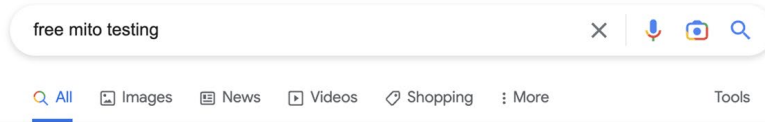


# Patient Experience

# Patient acquisition: patients find us online via

paid and organic

1



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**.

Paid ad

**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 ...

Organic article

<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**.

PG website

<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 ...

Advocacy group

# Patient acquisition: patients find us online via

paid and organic

1



## 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.provablygen...  
**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.