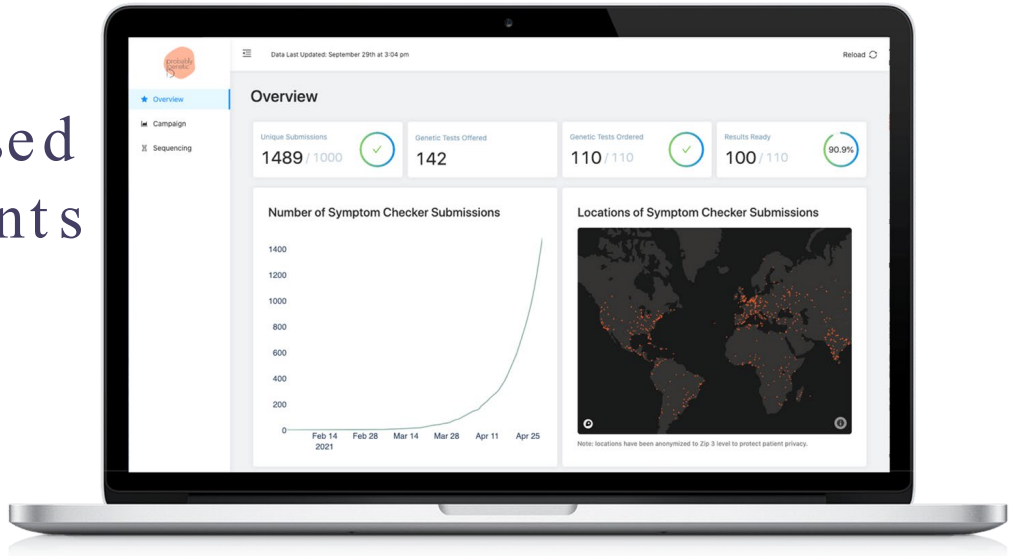
# Finding undiagnosed rare disease patients

Ultragenyx Bootcamp, 2023




Lukas Lange, PhD
CEO
lukas@probablygenetic.com

# Platform to commercialize rare disease treatments

- Patient finding
- Trial recruitment
- Treatment access
- Real-world evidence
- R&D data

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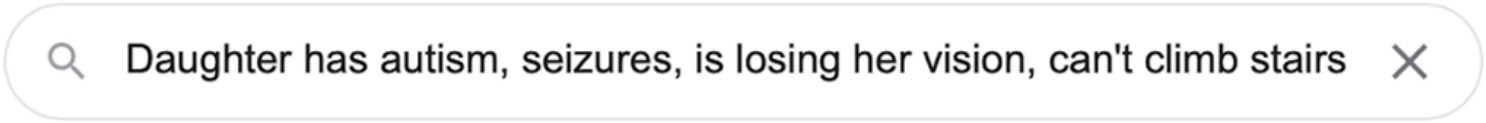
# How we diagnose patients is broken

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




Google

YouTube

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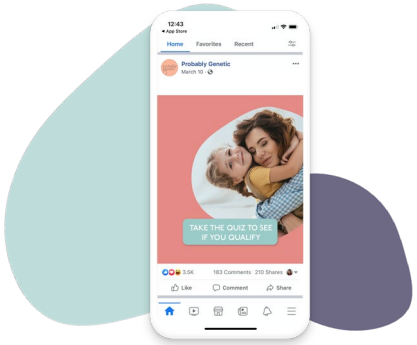
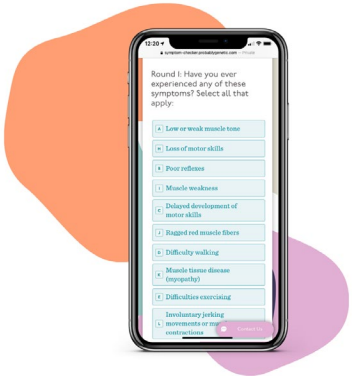
# We find patients online using machine learning and at -home testing

10 min

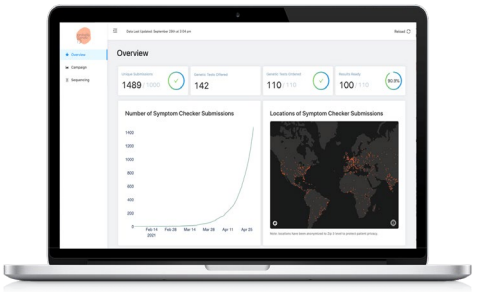
2 days

6 weeks





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## Awareness

Patients find us online

2

## Machine learning

Platform identifies undiagnosed patients

3

## Testing

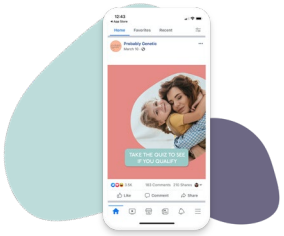
Patients are tested remotely

4

## Results

Patients receive results

Pharma gets data



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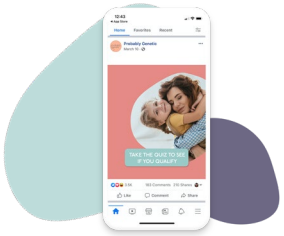
Awareness

# How???



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## Awareness

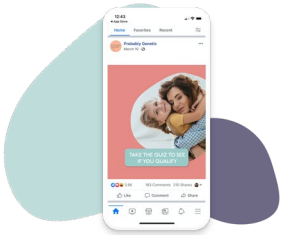
We built an invisible network of patient advocacy groups

- Seizures
- Developmental delay
- Autism

??
?

?

SLC6A1 Connect
SLC6A1 Research & Support



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**SLC6A1**
???

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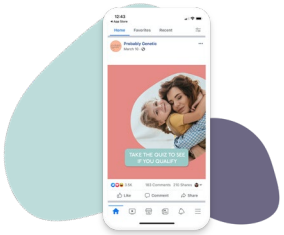
- Phenotyping
- Testing



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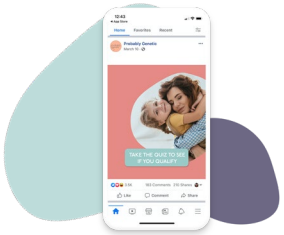

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- Phenotyping
- Testing

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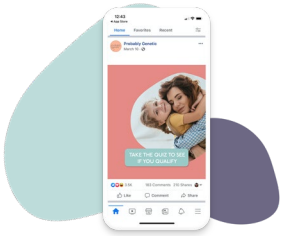
DLG4 Research Fund

DLG4
!!!



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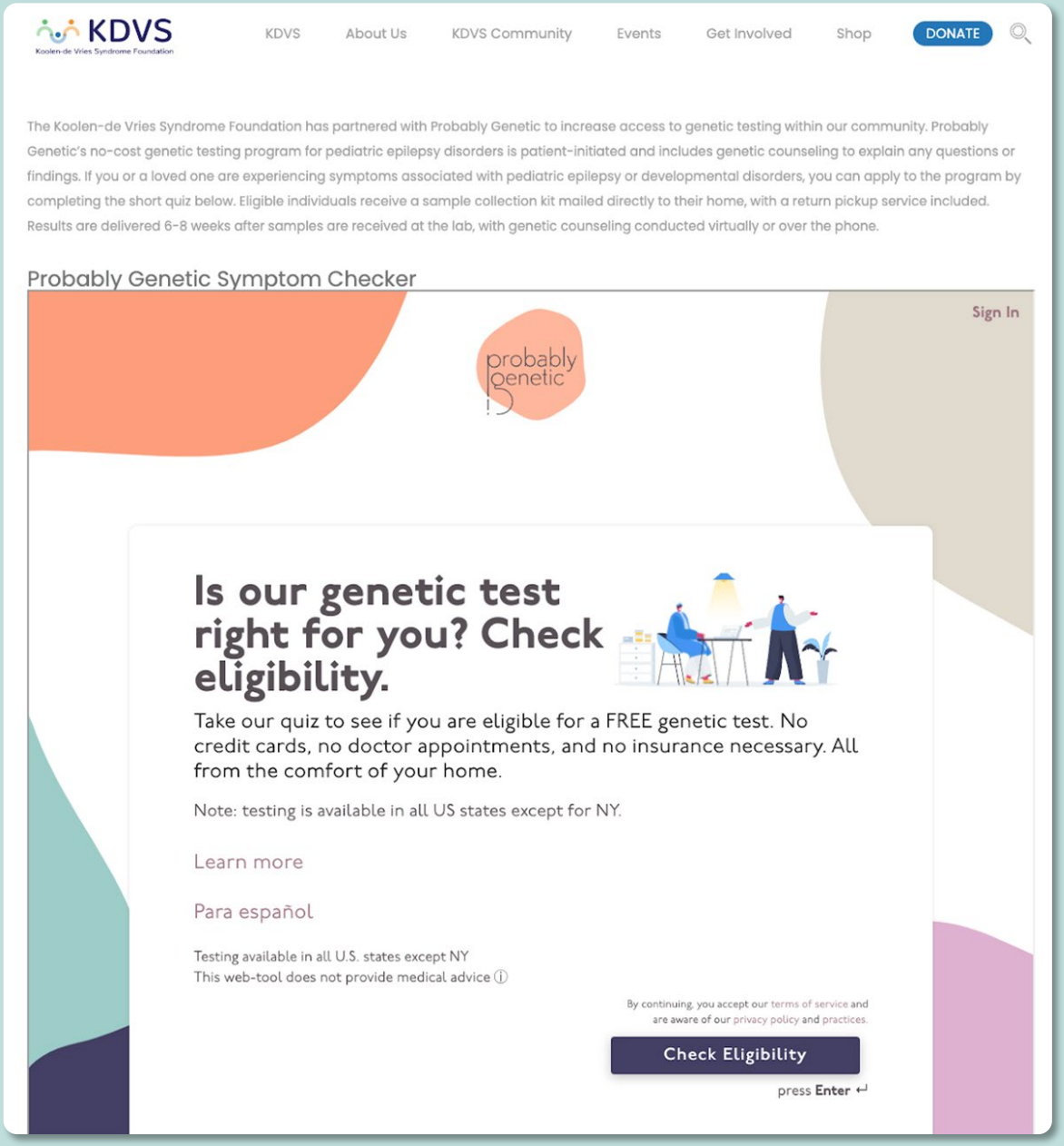

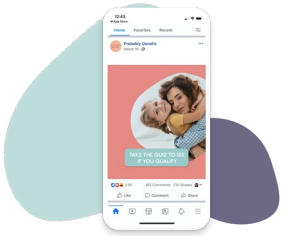

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## Awareness

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**>$150k monthly ad spend in network**

KDVS
Koolen-de Vries Syndrome Foundation

FAM177A1 RESEARCH FUND

CHAMP1 RESEARCH FOUNDATION

STXBP1

ARRE
ASXL RARE RESEARCH ENDOWMENT

IFCR
International Foundation for CDKL5 Research

Cure KCNH1 Foundation

SynGAP Research Fund
Collaboration. Transparency. Urgency.

WWOX Foundation

CURE RAGHAV

APBDRF
ADULT POLYGLUCOSAN BODY DISEASE RESEARCH FOUNDATION

MED13L FOUNDATION

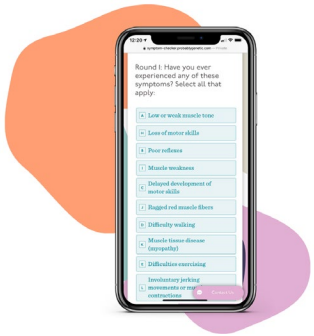
CTNNB1
CONNECT CURE

the SHANK2 foundation

THE Rory Belle FOUNDATION

SLC6A1 Connect
SLC6A1 Research & Support

# Thousands of patients sign up every month, globally

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## Machine learning

Live learning system identifies undiagnosed patients

**Text**

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

**Photos**

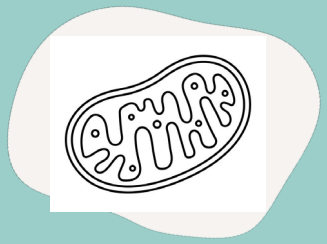
- ✓ Face
- • Skin
- • Hands
- • Feet

**Videos**

- • Seizures
- • Movement
- • Behavior

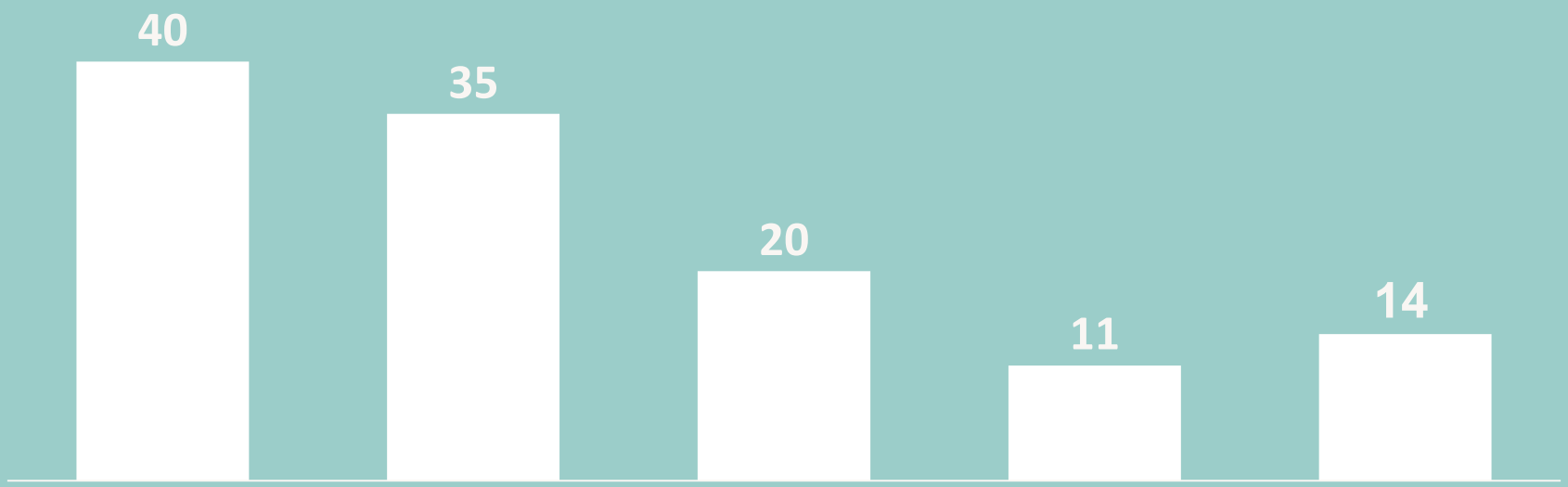
**Audio**

- • Voice
- • Hearing



# Mito program competitive with published studies

**% of patients tested confirmed to have a mitochondrial disorder**

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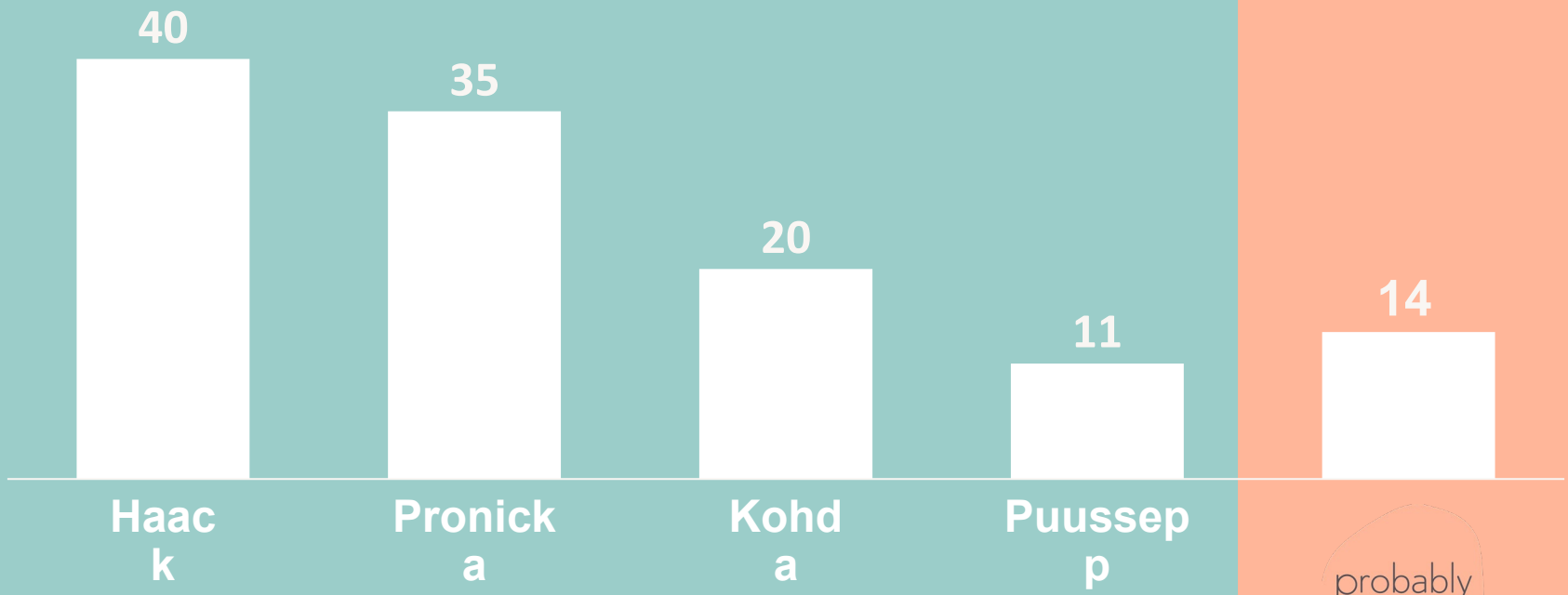
**Clinically diagnosed with mitochondrial disease prior to testing by specialists**

**Selected by machine learning algorithm**

**% of patients tested confirmed to have a mitochondrial disorder**

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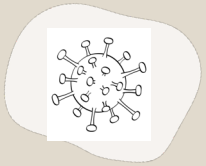
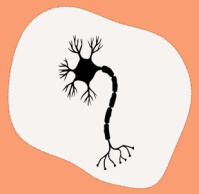
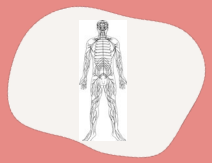


Immuno-deficiencies



Seizure disorders



Movement disorders

Mito

Metabolic disorders

Dementias

Encephalitis

To be announced

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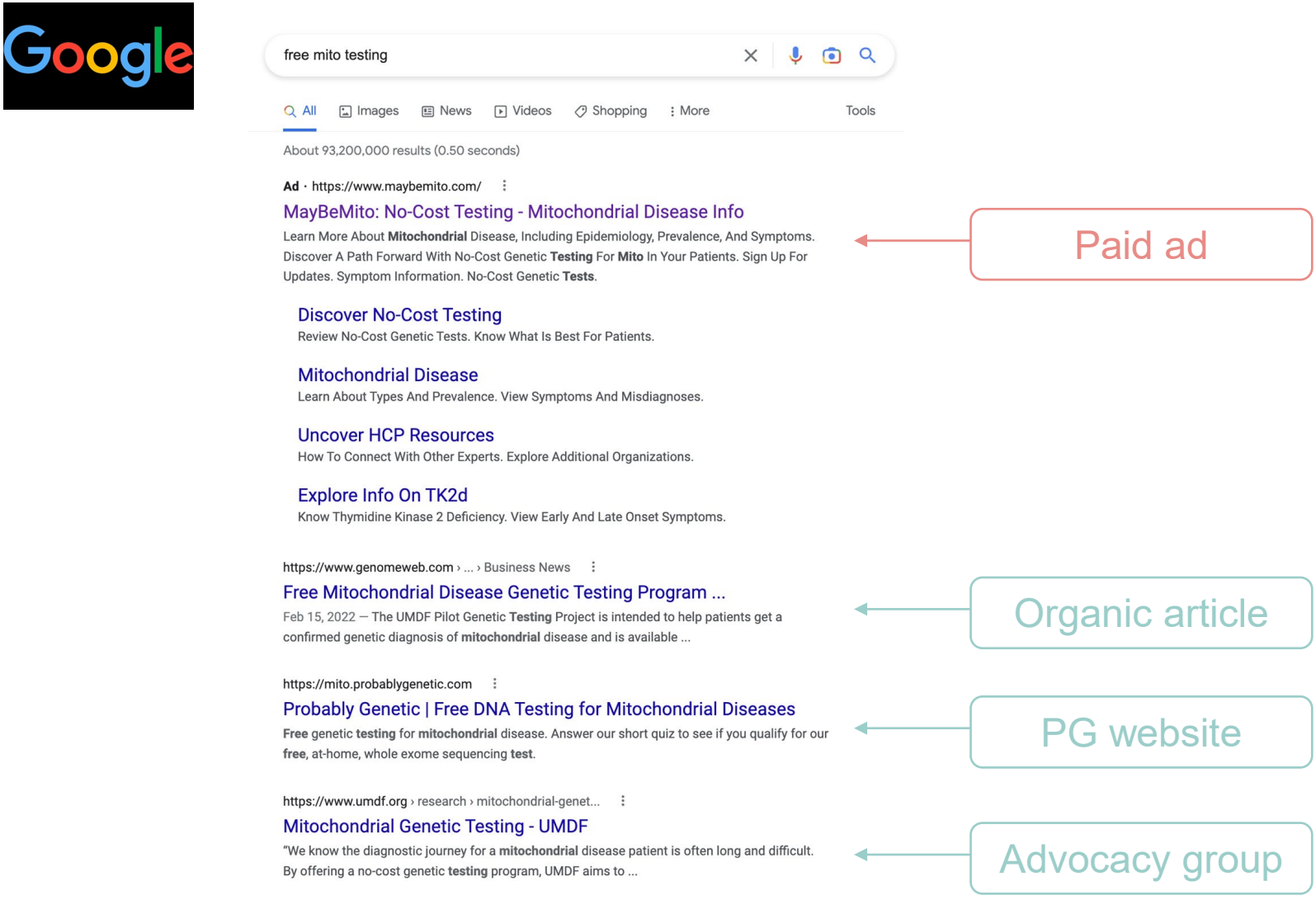
# Trying to find patients?
# Send us an email

Lukas Lange, CEO
lukas@probablygenetic.com


Patient Experience

# 1 Patient acquisition: patients find us online via paid and organic

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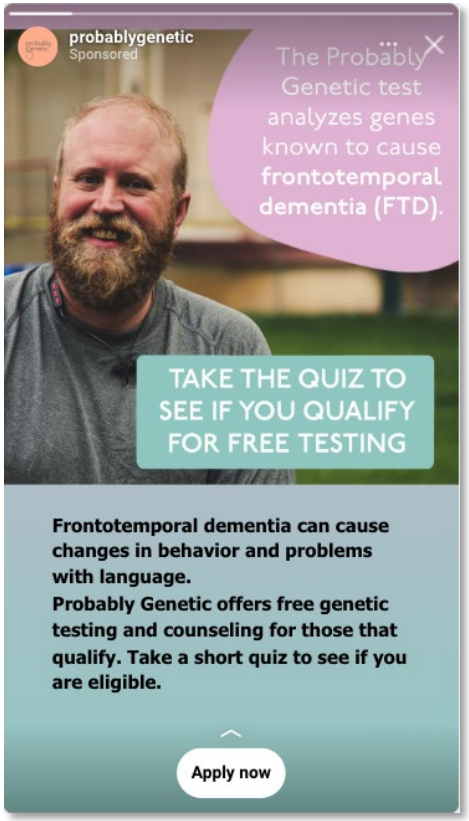
# 1 Patient acquisition: patients find us online via paid and organic

## Paid


## Organic