Probably Genetic Introduction

Ultragenyx Rare Disease Entrepreneur Bootcamp
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30 million people in the US suffer from a genetic disease

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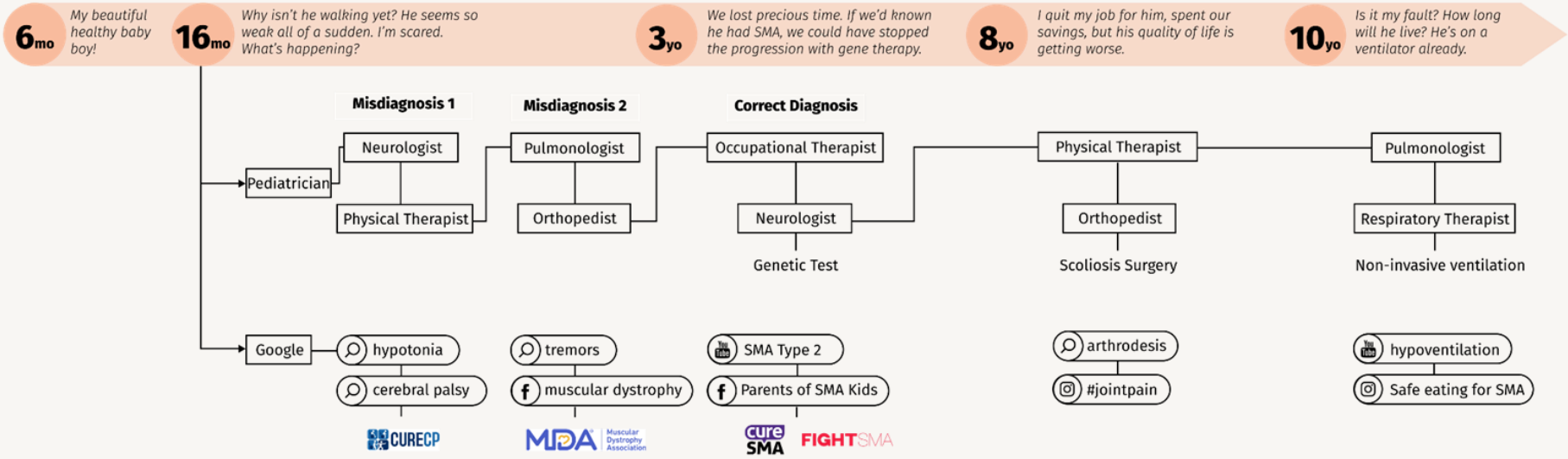
Patients are sick for years before getting diagnosed

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.

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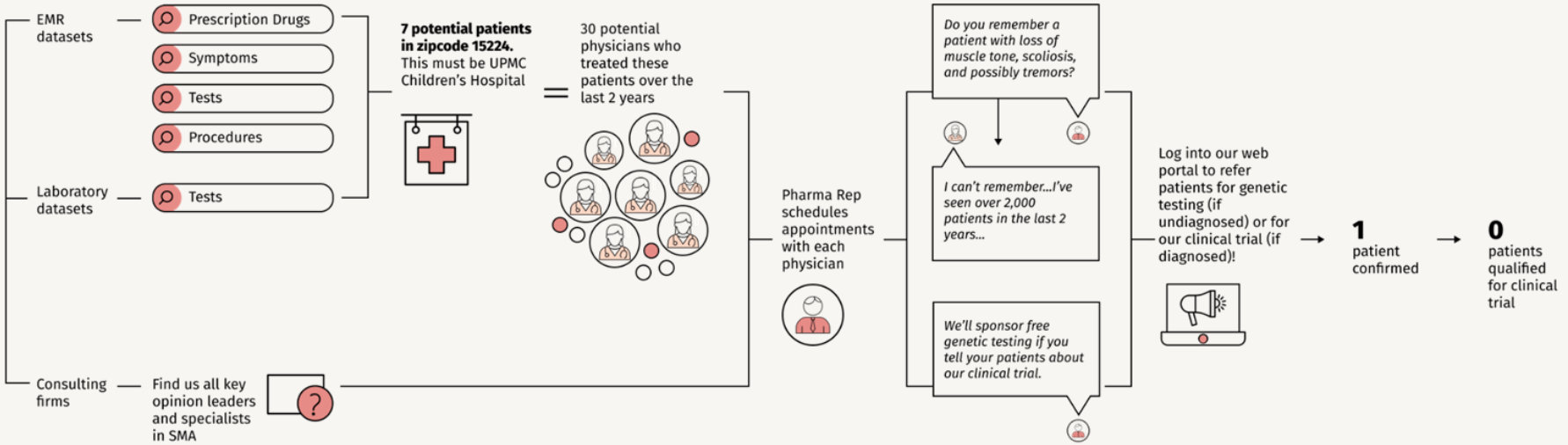
Finding patients offline is hard

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.

Current solutions only find a subset of patients …

EHR Records & Claims Searches
Did you hire vendors who analyze medical claims & EMR data, but **don't deliver because patient data is distributed across many different systems and most patients are undiagnosed**?

Awareness Campaigns
Did you partner with specialists, key opinion leaders, and PAGs, but **struggle to convert these awareness efforts into leads**?

Sponsored Testing
Did you sponsor testing, but only **reached a subset of patients who see the right referring specialist**?

Homegrown Solutions
Did you piece together vendors to create your own compliant patient finding funnel, but **struggle with low conversion rates and uptake**?

… We find 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

Consented and can be activated via compliant communications suite

Probably Genetic helps patients and pharma find each other,

solving 3 core challenges for pharma and patients.

1. ONLINE PATIENT ACQUISITION

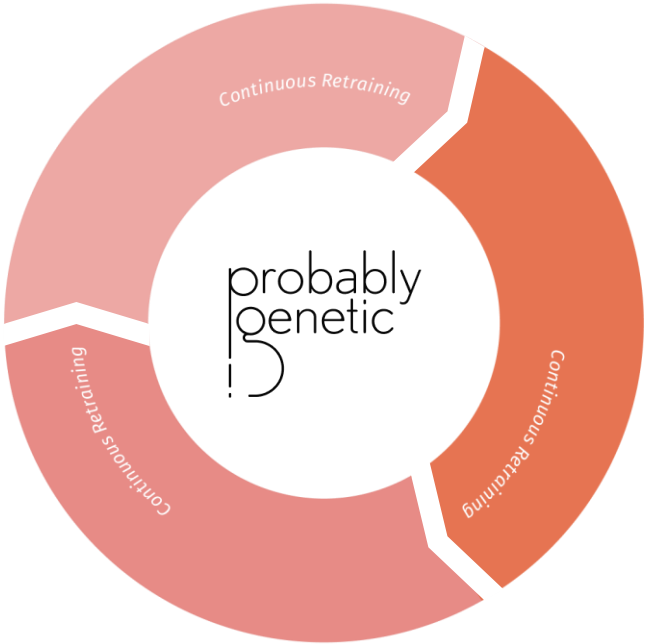
Direct access to hard-to-reach patient populations

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

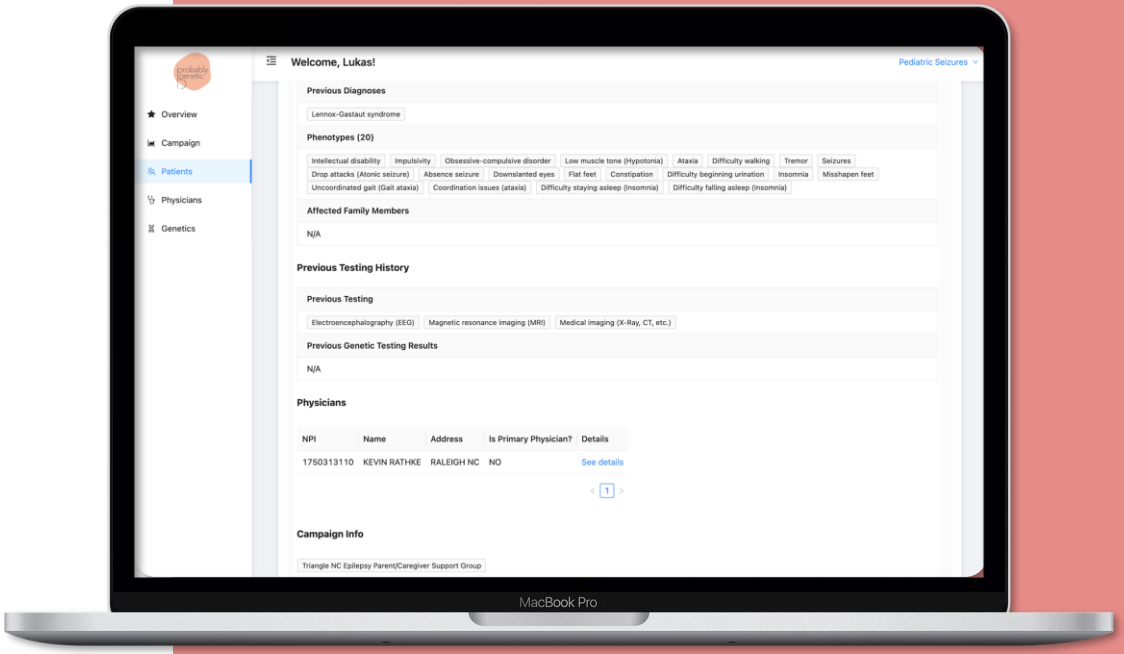
Text	**Photos**	**Videos***	**Audio***
Phenotypic Terms	Face	Seizures	Voice
Family Pedigree	Eyes*	Movement	Hearing
Medical History	Skin*	Behavior	
Meta Data	Hands*		
Lab Reports	Feet*		
EMR Data*			

* *Not yet collecting*

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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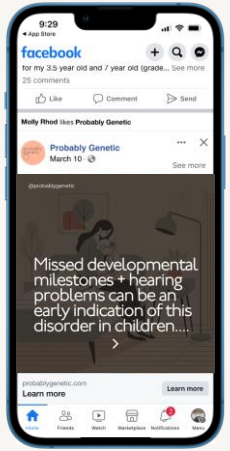
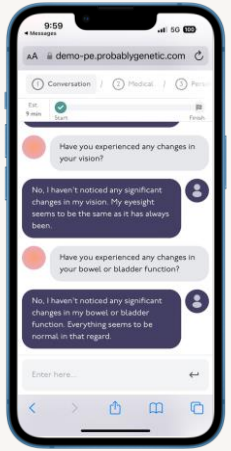
Patients get free testing



Step 1

Discover Outreach

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



Step 2

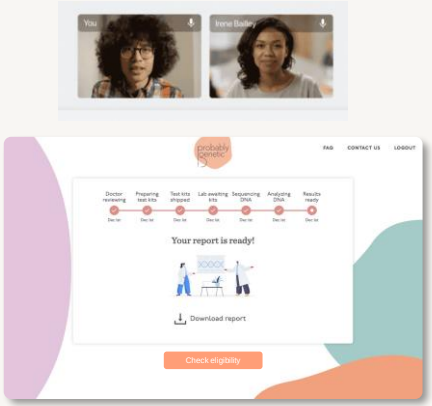
Answer a Few Questions

➔ Complete a chat conversation with our LLM - ChatPG

Step 3

Get Tested & Counseled

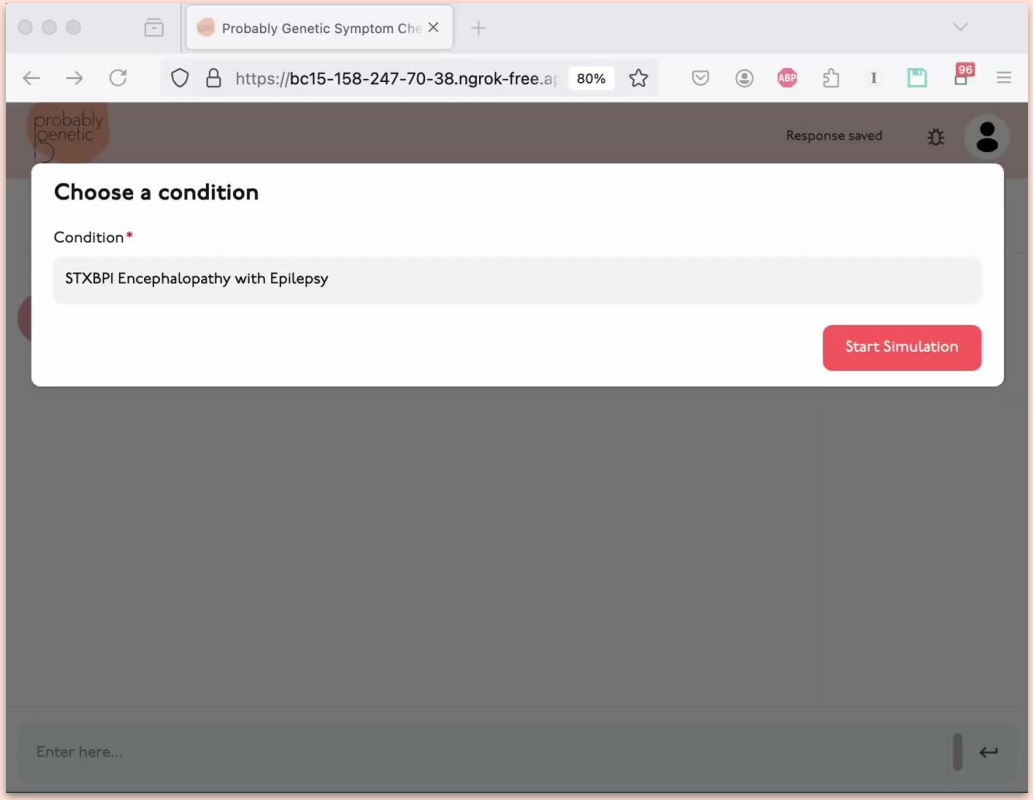
➔ Receive an at-home test in days with comprehensive physician and patient support



Step 4

Engage in Next Steps

➔ Learn about next steps via a physician consultation or direct messaging PG's patient support team



Simulated STXBP1 parent

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.

Patients sign up globally

Our program participants are diverse and representative of the US

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

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62% White / European

11% Hispanic

8% African American

6% Indigenous American

4% Asian

3% Ashkenazi Jewish

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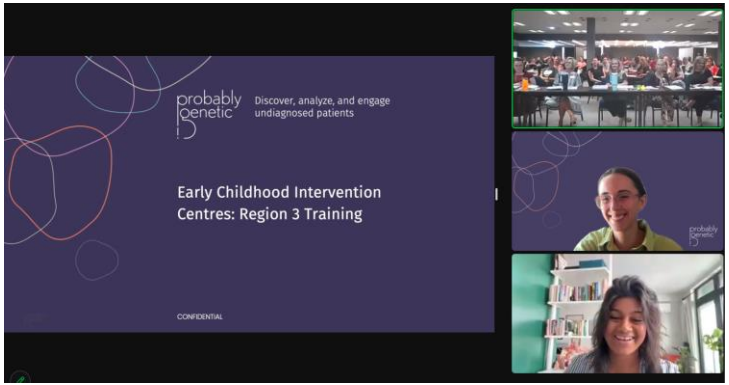
We reach cohorts with significant health disparities

Reach in various languages...

Content viewed 7,300+ times by 4,100+ unique users

... to various communities.

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



100+ Occupational Therapists Engaged

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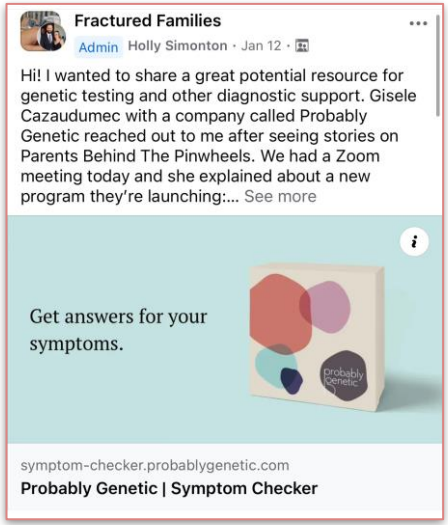
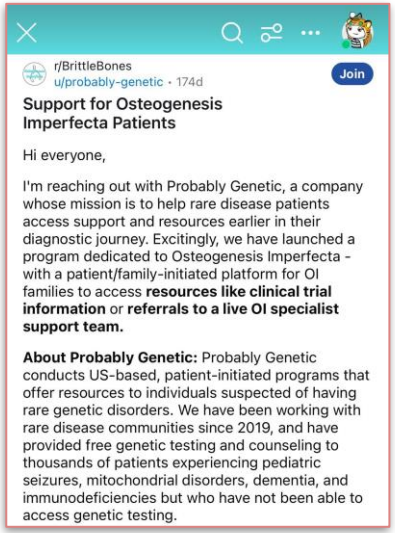
We work hand in hand with patients to expand reach

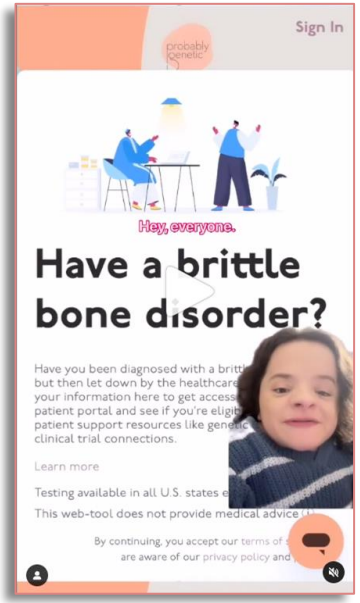
Private Groups



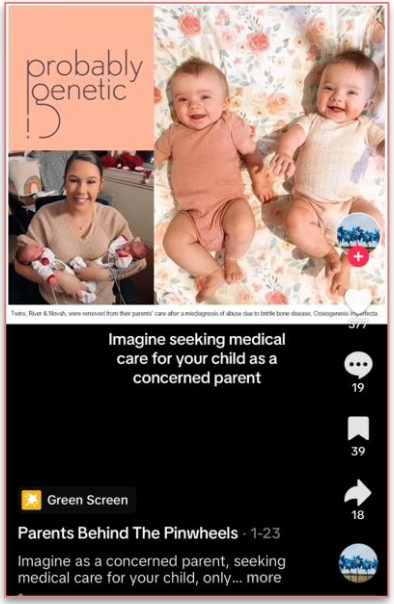
Public Groups



Ambassador & Influencer Program



Patient Foundations



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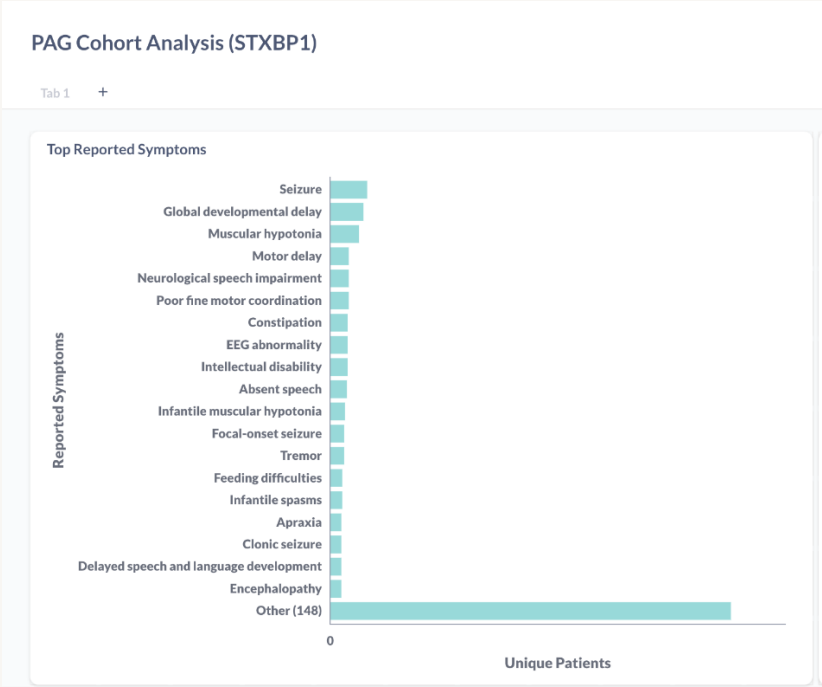
PARTNERSHIPS

We support patient advocacy groups for free in various ways

How we partner:

- **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



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Thank you!

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