




Invitae Real World Data

Imagine knowing more...






Despite major advances in translational research, 95% of all rare disorders do not have an FDA-approved therapy¹

- With novel therapeutic approaches, including gene and cell therapies, there is increasing investment in drug development for rare disorders
- Observational data (like natural history) is required for clinical trial readiness, optimizing clinical trial design

“If you do only one thing, create a natural history study” - Neil Kumar, CEO, BridgeBio

Observational studies are challenging in rare

A photograph of a woman with long, straight dark hair and a young child with curly dark hair. The woman is wearing a yellow sweater and the child is wearing a green top. They are both looking towards the right side of the frame. The background is a plain, light-colored wall.

- Phenotypic variability across distributed populations
- Prohibitively expensive to stand-up, prolonged timelines
- Participation burden for families resulting in attrition
- Fragmentation of care leads to incomplete data
- Traditionally, lack of engagement with the patient community

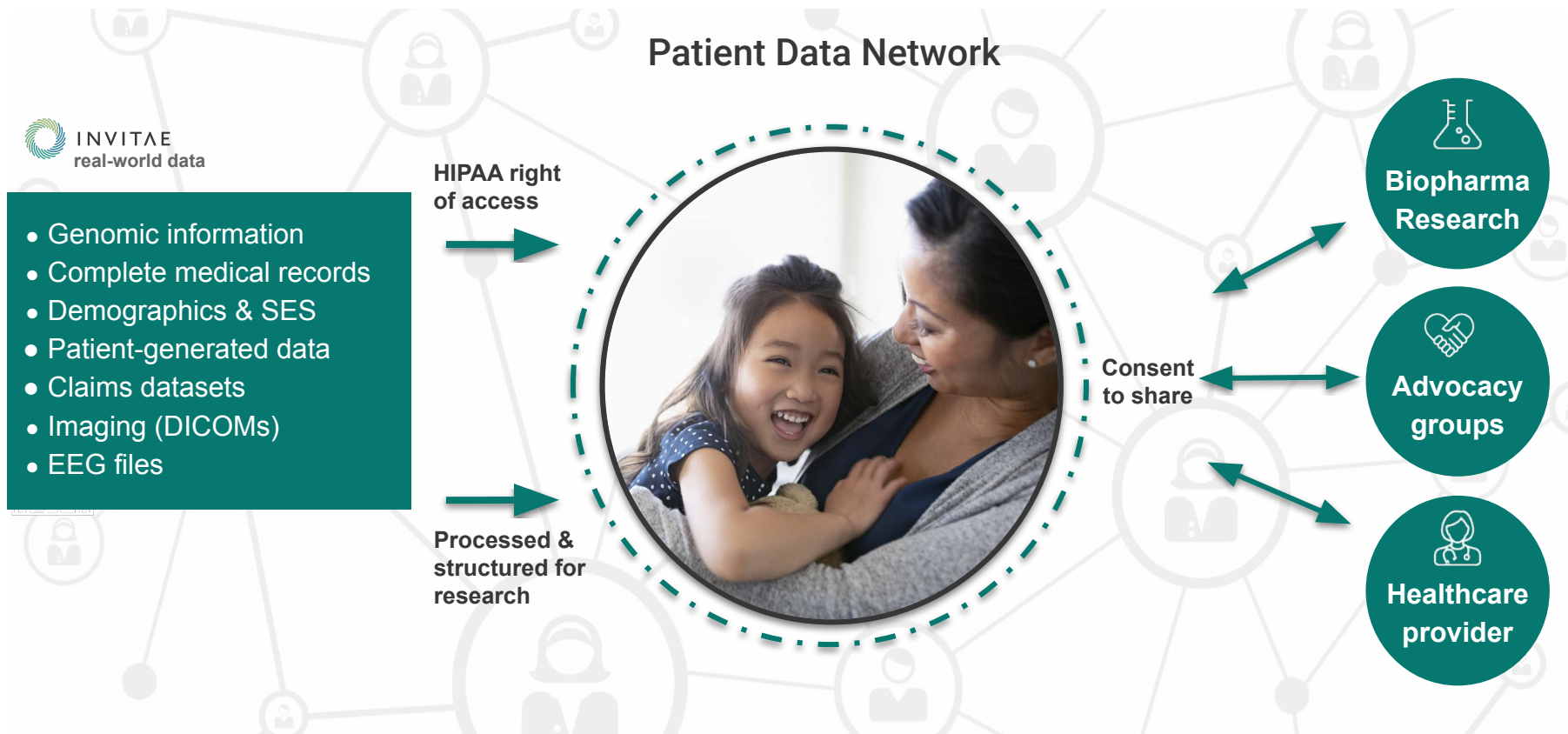
21st Century Cures Act

Signed into law December 2016, intended to:

- Accelerate medical product development
- Incorporate patient perspectives into drug development
- Modernize clinical trial design

The Cures Act positions advocacy groups to drive early investments in clinical data collection

A new approach: patient-centric, data-powered healthcare



Part 1: Longitudinal data acquisition

Ciitizen collects longitudinal medical records and images from every point of care



Caregiver/Patient
onboards to Ciitizen

Caregiver/Patient
consents to share data
with researchers



Regulatory expertise drives
industry leading record retrieval
rate of 99%

Ciitizen uses the patient's HIPAA right of
access to request raw records from all
providers (institutions, home offices, etc)

Ciitizen receives all unstructured medical
records (ex: genetic reports, neurology
notes, procedure notes) and imaging
studies

HEALTH INSTITUTION	SCORE	REPORTED AS OF
Scott R Anagnoste, 6333 N Federal Hwy 535 E 70th St, New York, NY	☆☆☆☆	1/21/21
Scripps Clinic Torrey Pines 10666 N Torrey Pines Rd, La Jolla, CA	★★★★☆	11/4/20
Scripps Green Hospital 10666 N Torrey Pines Rd, La Jolla, CA	★★★★☆	11/4/20
Scripps Health System – All Locations 10140 Campus Point Dr, San Diego, CA	★★★★☆	11/4/20
Seattle Cancer Care Alliance 1354 Aloha St, Seattle, WA	★★★★★	12/18/20
Seattle Cancer Care Alliance at UWMC – Northwest 1560 N 115th St Level G-16, Seattle, WA	★★★★★	11/24/20
Seattle Children's Hospital 4800 Sand Point Way Nw, Seattle, WA	★★★★☆	1/14/21
Seattle Children's Autism Center 4909 23rd Avenue Northeast, Seattle, WA	★★★★☆	1/14/21
Seattle Integrative Oncology 2859 Eastlake Ave E, Seattle, WA	★★★★☆	6/2/20
Seattle Special Care Dentistry 4915 23rd Avenue Northeast Ste 205, Seattle, WA	★★★★★	9/10/20

Ciitizen created The Patient Record Scorecard to
publicly score hospitals and MD practices on
HIPAA compliance



Part 2: Patients are data owners

Citizen provides shareable raw records to patients



All medical records and images scanned and uploaded to patient's account

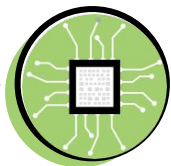
Patient owns their raw records

Patient can easily share records and images with any provider or person

The screenshot displays the Citizen Health Records interface. At the top, the 'Health Records' tab is active, showing a search bar and 'Records' and 'Share' buttons. A sidebar on the left lists categories: 'All' (21), 'Providers' (0), 'Documents' (15), and 'Imaging Studies' (6). The main area shows a grid of document thumbnails, each with a title and upload date (e.g., 'PET-CT Whole Body 05_11_2021', 'Breast Biopsy 4_29_15_1_2021', 'MRI Breast 5_12_15 (1)_1_2021'). A 'Share your Data' dialog box is overlaid, allowing users to share their 'Cancer Summary' or 'Entire Profile' by entering an email or selecting from 'Recently Shared With' contacts like 'sophia+demo...' and 'Miten Mistry'. A 'Create Share Link' button is also present.

Part 3: Citizen makes data useful

Structured and unstructured machine learning pipeline creates normalized, research ready data



Raw medical records are put through machine learning/NLP-assisted extraction engine



Human assistance and review of pipeline generated data for QA/QC

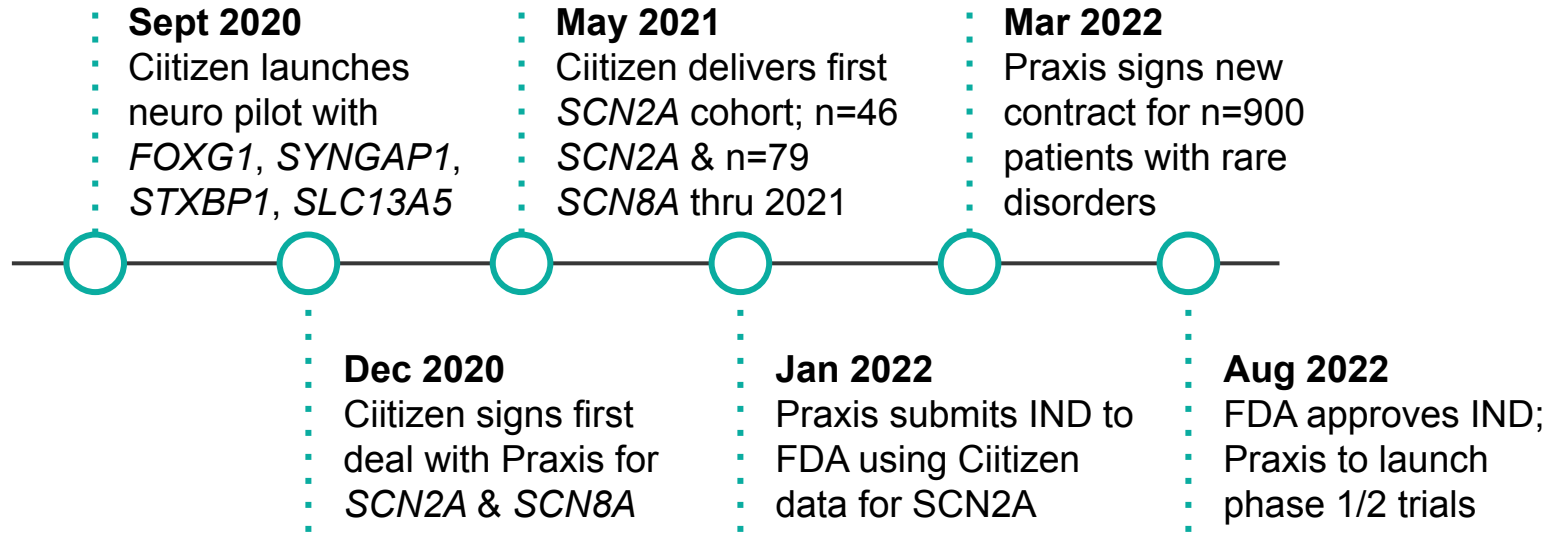


Data output is structured, coded, queryable and portable

Datasets shared at **user's discretion** for longitudinal phenotypic characterization, clinical trial design support, and evaluation of genotype-phenotype correlations, among other uses.




Ciitizen and Praxis: a proof of concept



*Through exclusive use of Ciitizen for clinical data, Praxis to launch trials **years earlier.***





Praxis obtained investigational new drug (IND) clearance from FDA for PRAX-222¹

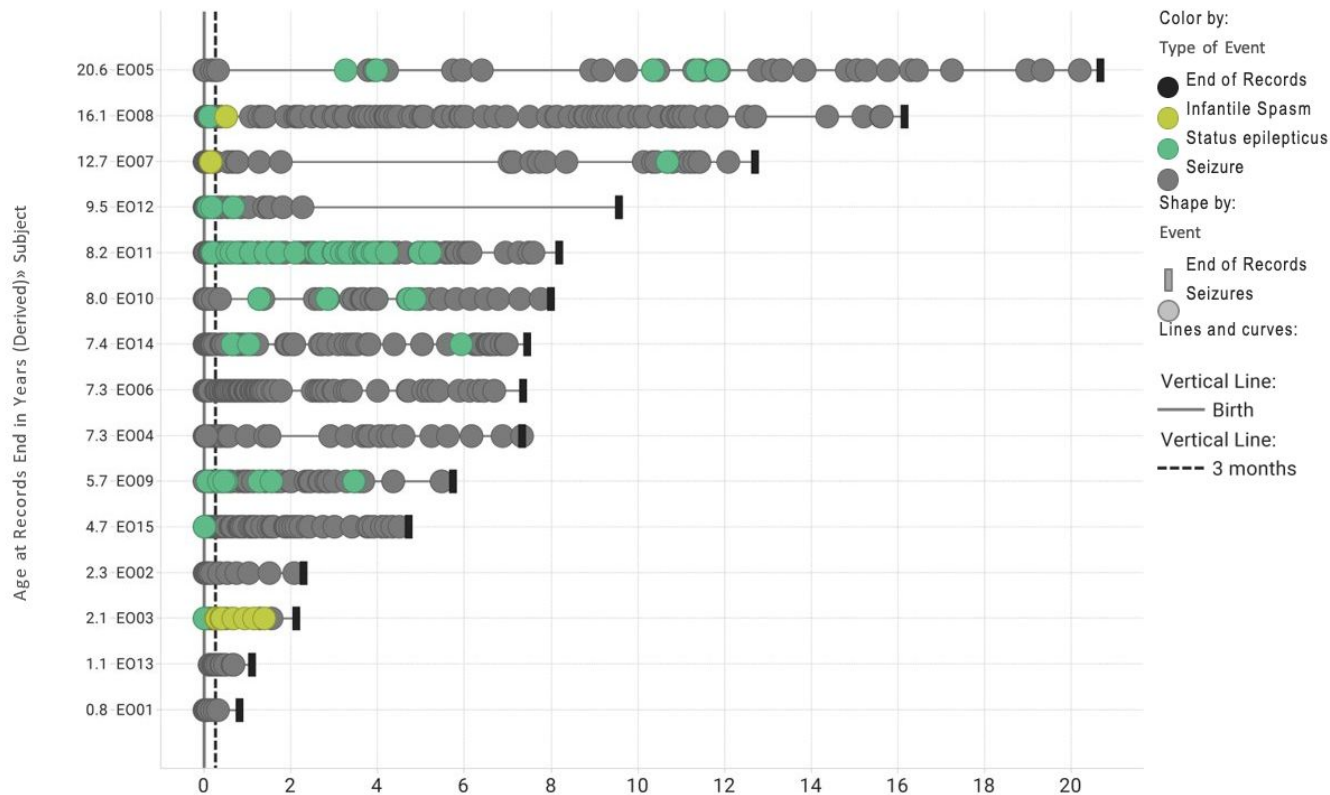
- PRAX-222 - ASO therapy for SCN2A-DEE
- Invitae's Ciitizen RWD used to demonstrate lived experience of SCN2A-DEE patients
- Invitae's Ciitizen data was the only natural history data used in submission package
- Using Ciitizen data advanced program by years

1. Praxis Precision Medicines Press Release. Issued September 7, 2022.
<https://investors.praxismedicines.com/news-releases/news-release-details/praxis-precision-medicines-provides-updates-clinical-stage>

Praxis leverages Citizen data to optimize Ph1/2 design

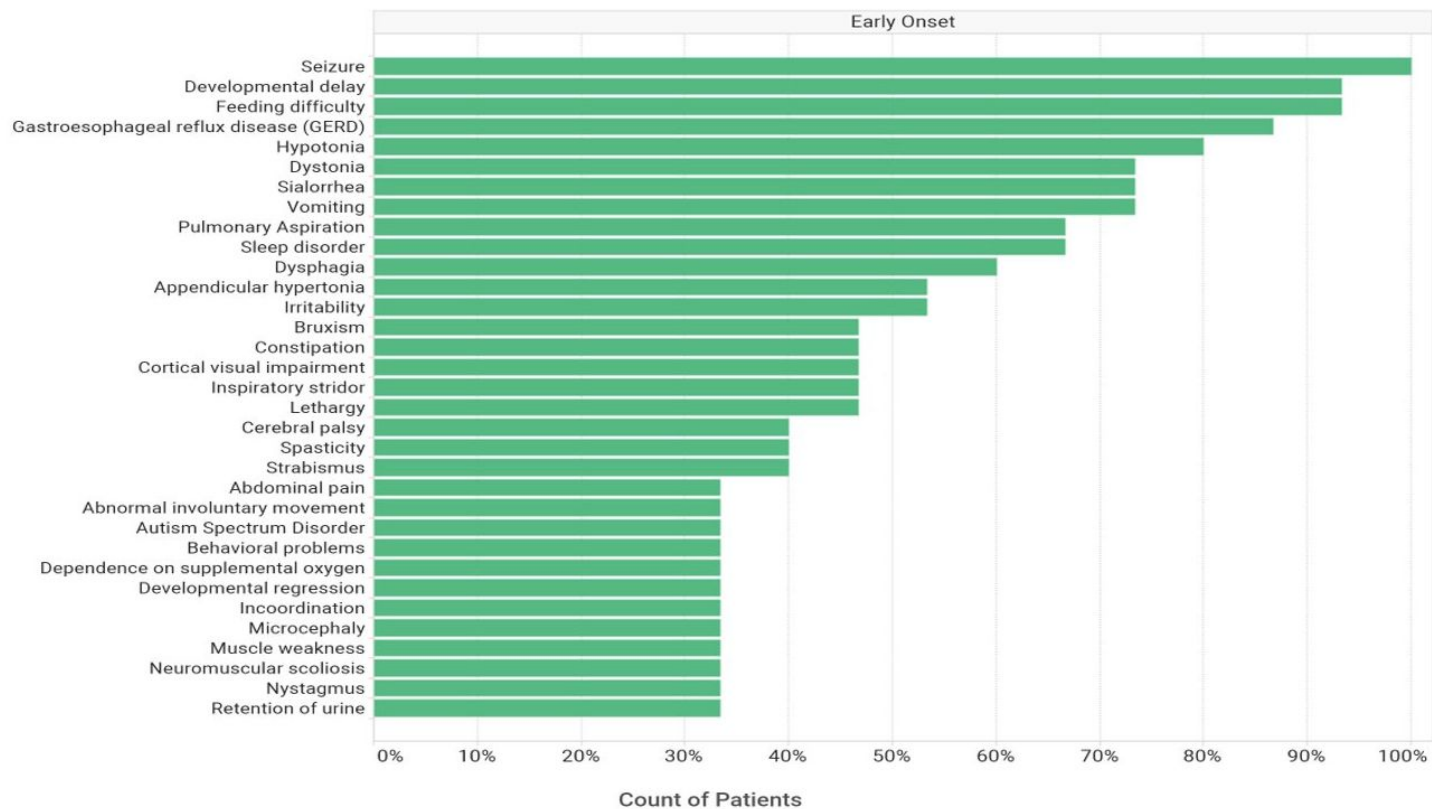
- Inform endpoint selection and characterization
- Refine genotype-phenotype correlations; accurately predict GOF variants
- Define inclusion/exclusion criteria
- Develop and refine an EEG biomarker
- Demonstrate unmet need during key regulatory interactions
- Plan for clinical trial sites and recruitment

Praxis use of Ciitizen data: Seizure frequency as primary endpoint for SCN2A-DEE *throughout life*

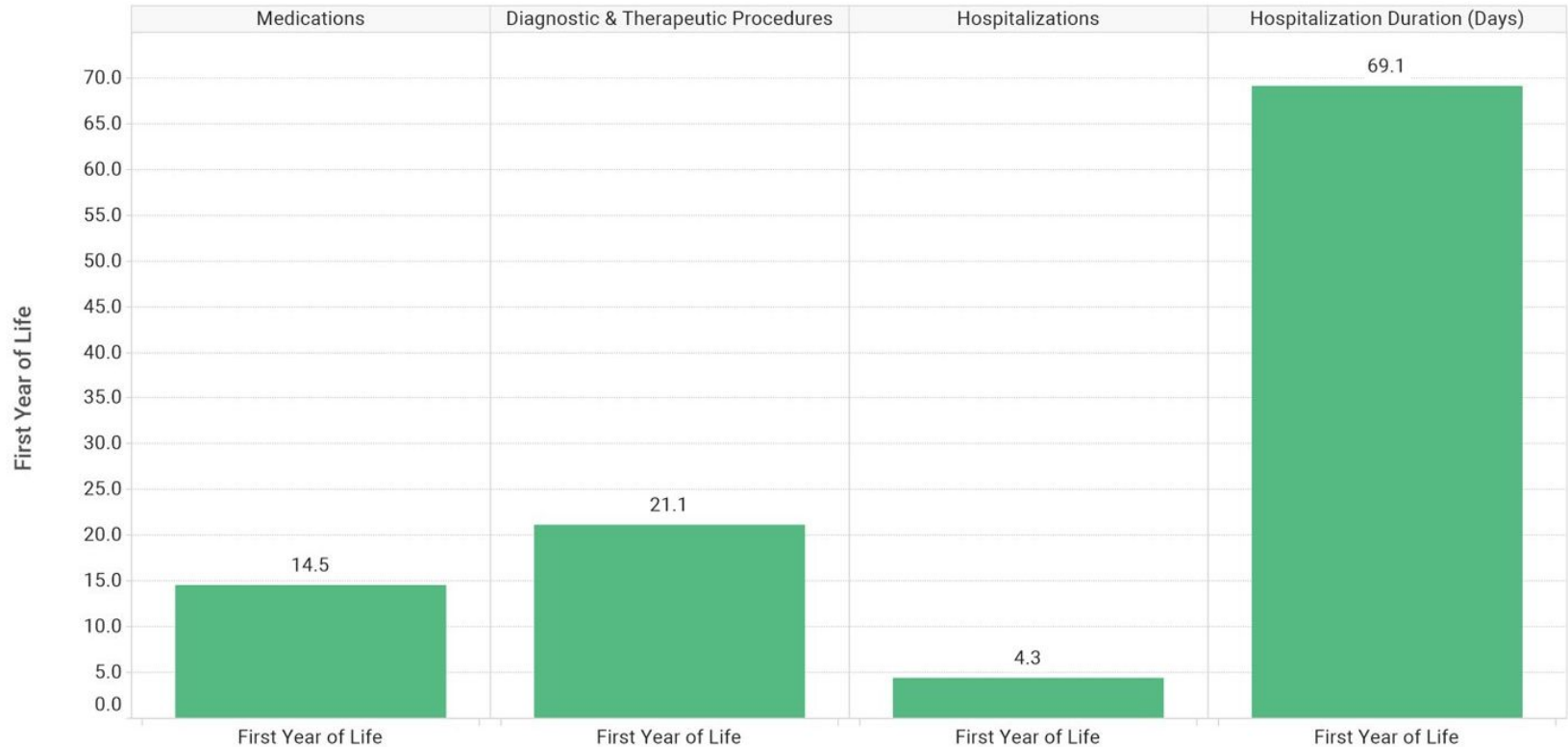


- Literature suggests early onset seizures remit
- Ciitizen found seizures persist
- Data helped confirm primary endpoint and age I/E criteria

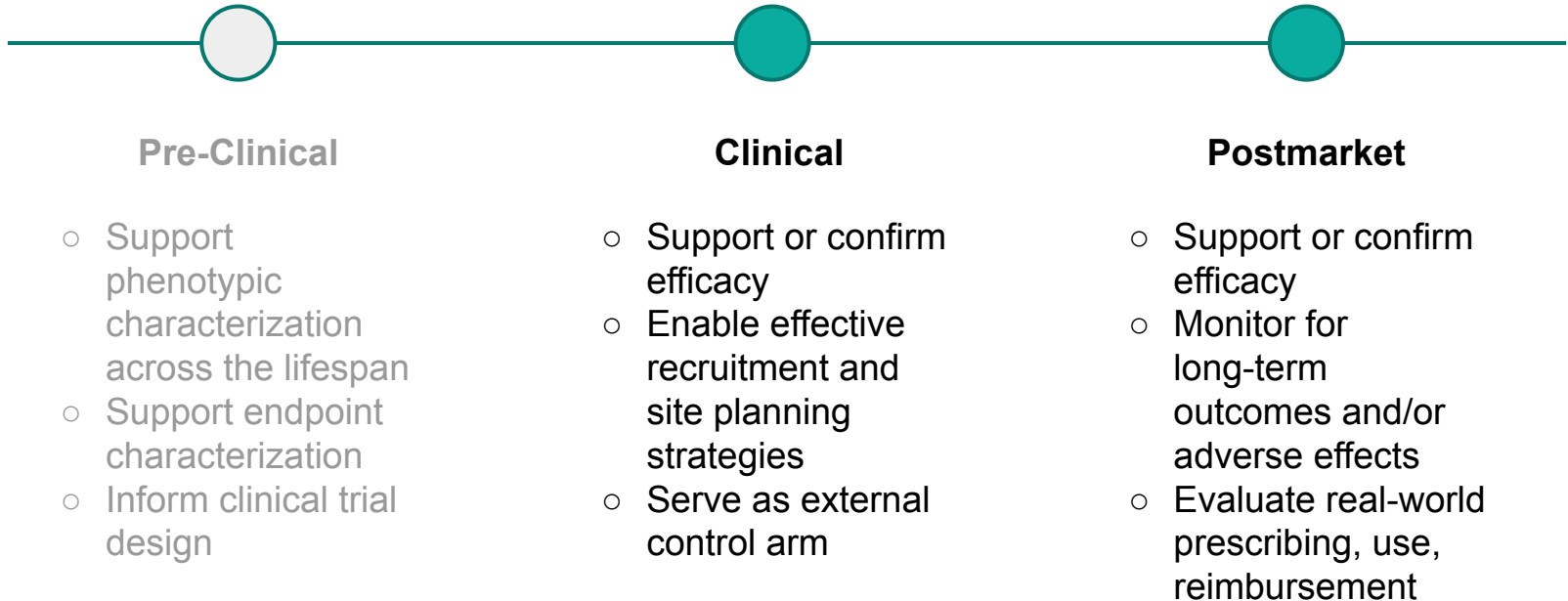
Praxis use of Ciitizen data: Exploratory endpoints and clinical trial design



Praxis use of Ciitizen data: Burden of Disease



What comes next?



Today, we are expanding our platform



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Join our Rare Patient Network

Now supporting childhood onset of epilepsy and/or developmental delay.

[Get started](#)



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Patient families: Who can join?

Any patient who meets these criteria is invited to join at no-cost:

- ✓ ≥ 1 seizure with onset before the age of 18 years old
- ✓ Developmental delay in any domain
- ✓ Receiving medical care in the United States

Learn more at ciitizen.com/rarenetwork



Patient benefits

Medical records made easy, research made possible



Say goodbye to the binder

View your existing medical records in one secure account.*



Build a faster way forward

Share summarized medical history with researchers, if you choose.



Be in control

Send a link to clinicians, caregivers and loved ones—or don't. It's up to you.



Advocacy Leaders: New Partnership Opportunities



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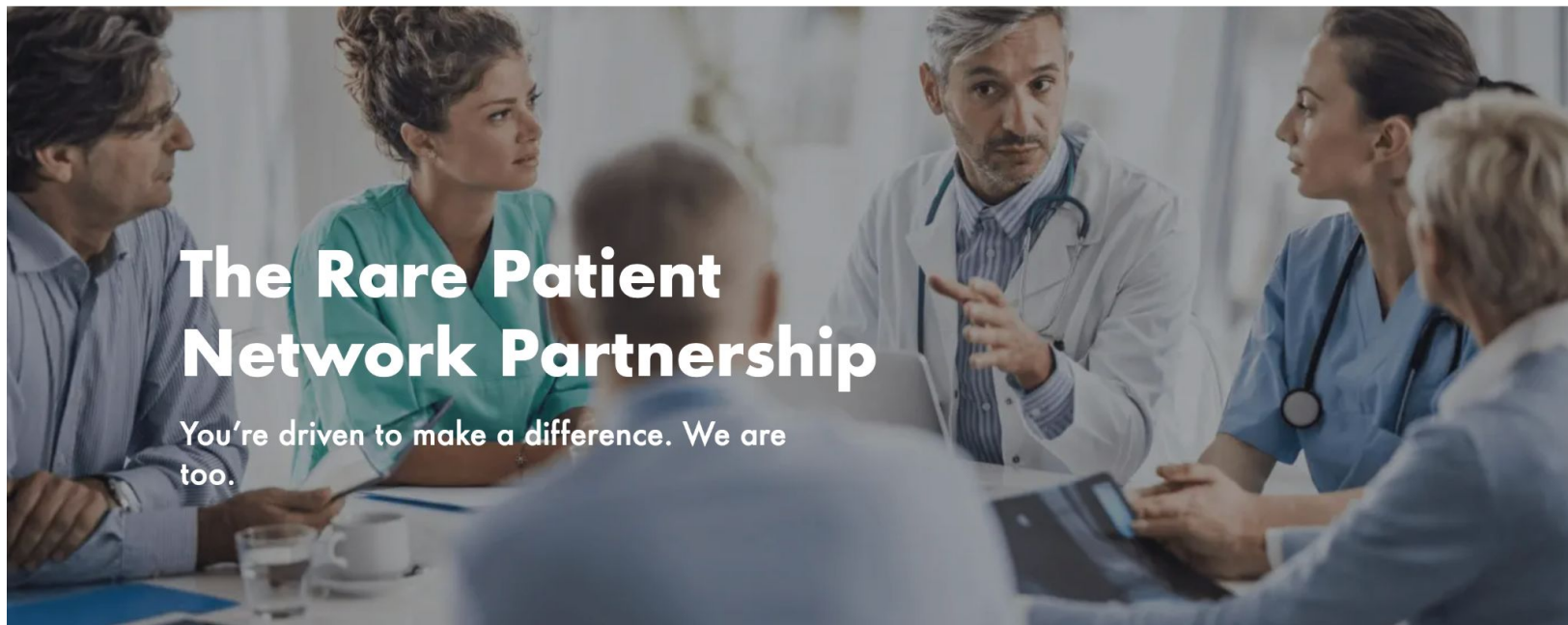
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The Rare Patient Network Partnership

You're driven to make a difference. We are too.

Learn more at citizen.com/rare-partners



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Advocacy Group benefits

Benefits

Potential benefits to your community include



Provide caregivers and patients with one resource to access, manage and share medical records



Obtain best in class, research-ready datasets that can be shared with researchers and biopharma to accelerate therapeutic opportunities and understanding of conditions



Overcome gaps in understanding of biomarkers, clinical outcomes or critical needs for clinical trial readiness for your condition



De-risk your condition to biopharma and position your community as a strong partner

