



Citizen Health Community Introduction




**Connecting patients.
Accelerating cures.**

Our mission

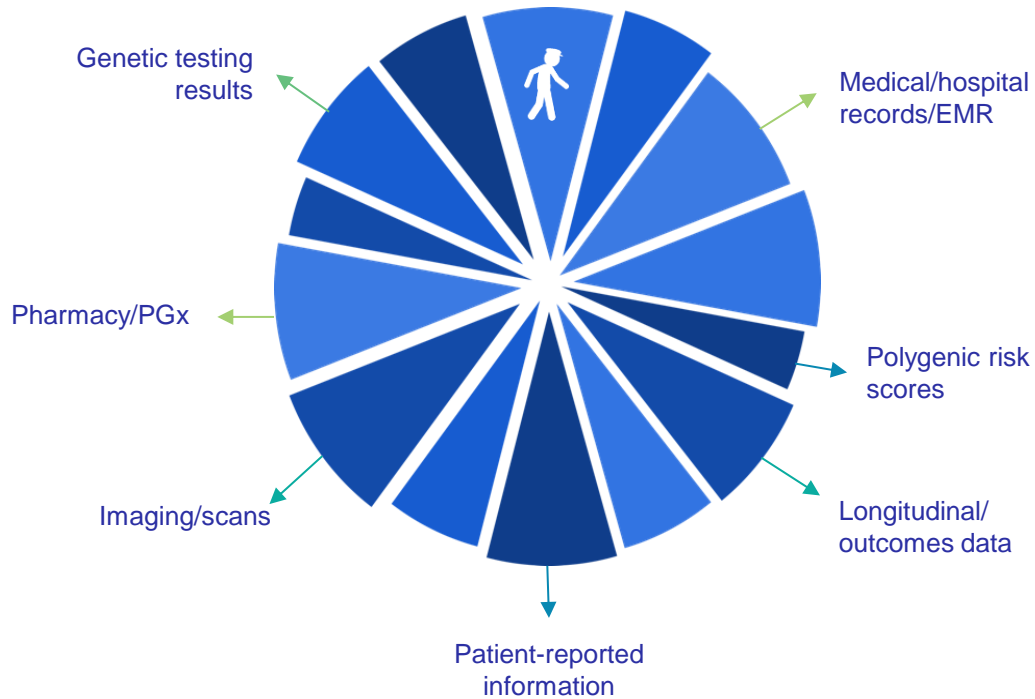


*Use **the power of shared experiences** to **provide relief** to patients and families navigating rare and complex conditions*

To do this, we amplify the rare disease experience using health data to:

- **Empower patient** to better their care and help one another
 - **Accelerate research** for treatments and cures
- 

Valuable patient data is trapped



There is immense value in the medical record, but those are trapped in various institutions



There is no single longitudinal view of the patient's care



Difficult to get access to all of your medical records to help inform your own care

Traditional models are falling short



95%

*Rare Diseases have
no treatment*

\$2.3B

*Average cost of
drug development*

10+ y

*Time to develop
successful therapy*

\$1 Trillion

*Economic cost of 379
rare diseases*

It can **take years** to collect enough data to understand the disease and how it evolves over time

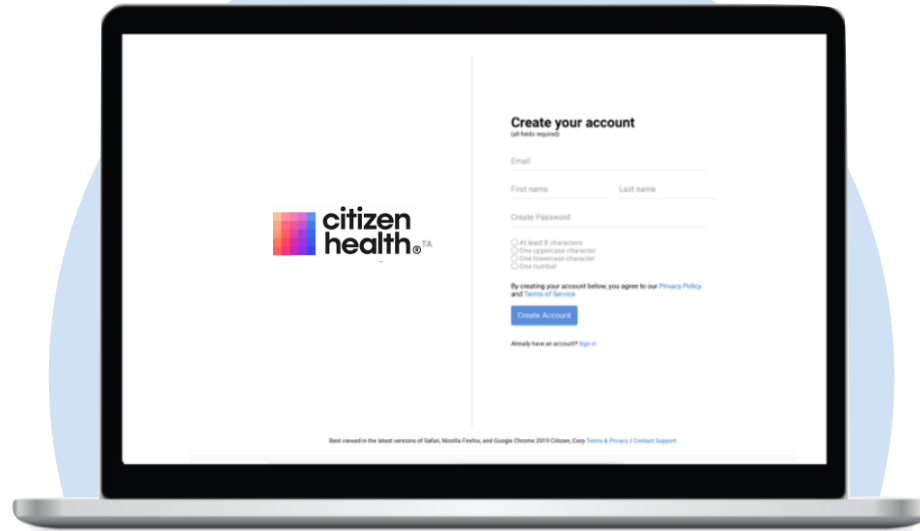
Participants have to **travel** to a clinic for data collection

Expensive and difficult to execute in rare disease

We're here to change that

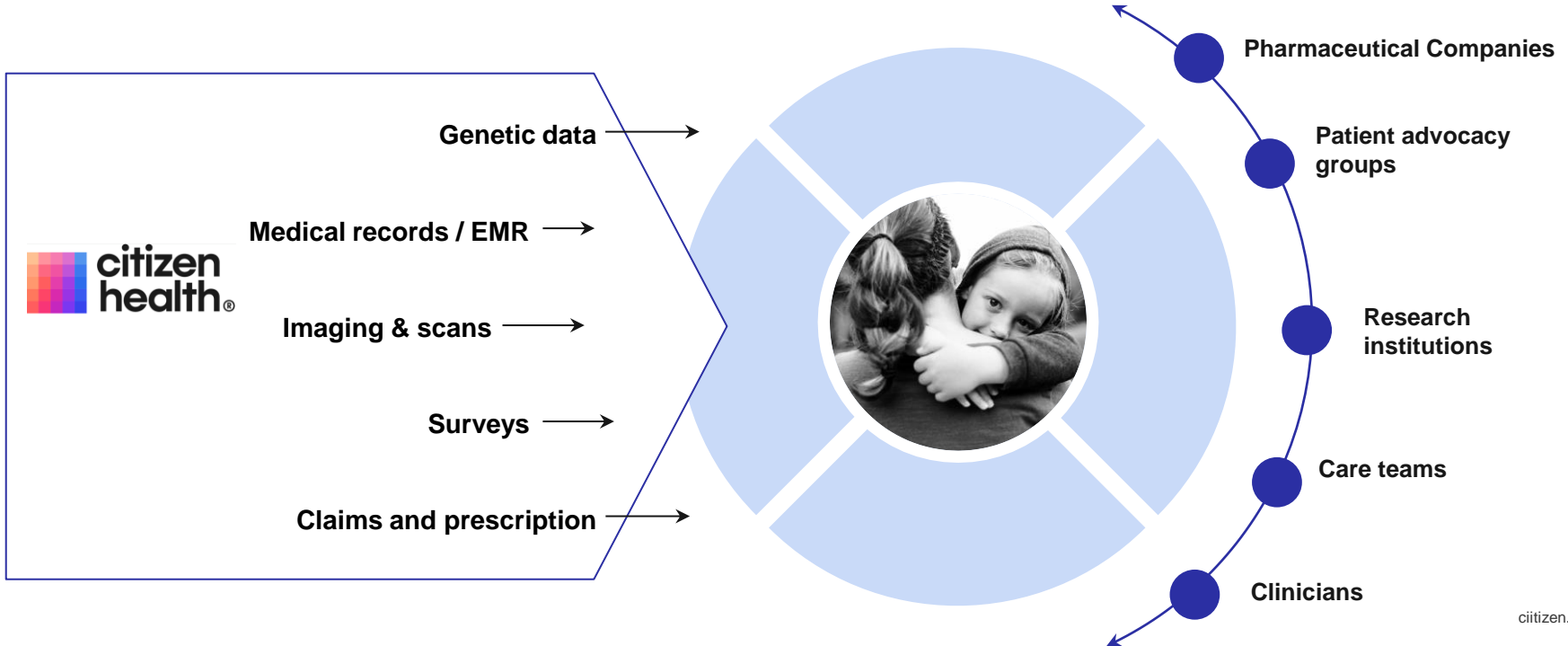


Get access to all your medical data in one place and digitally power research, all with your consent

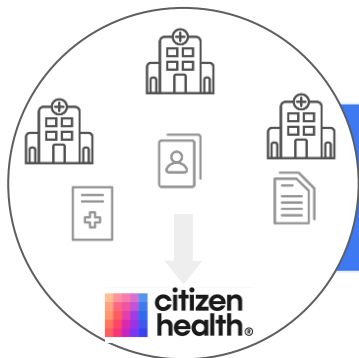


Designed with the needs of families and advocacy groups at heart

Two-sided platform where patients and caregivers have the power to share real world data and receive benefits



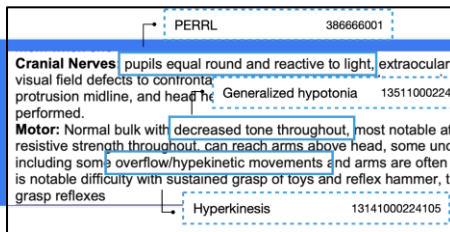
How does it work?



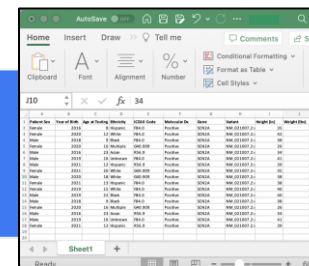
Collect a patient's health data



Upload to the patient's digital account



Codify and structure the data



De-identify and share consented data to drive research

Patients are partners! And we are committed to you



Patients are partners

- Citizen users “own” their data and direct who it is shared with
- All medical records & annotated data are made available to users
- Business model supports revenue sharing with participants & advocacy groups, in compliance with relevant law

Research ethics prioritized

- De-identified Citizen data is only shared with the express consent of participants
- All use of Citizen data must be covered by the participant’s consent
- All use of Citizen data must be reviewed by an IRB

Benefits to patients

- Easiest way to power research
- Access all your medical records in one place
- More control and understanding of your or your loved one's healthcare
- Help build important natural history research for your condition

Why should I join?

citizen.health

- ✓ Get access to all your medical records in one place
- ✓ Easily power and accelerate research efforts without leaving your home
- ✓ Gain more control and understanding of health
- ✓ Support your community in their vital step to build natural history studies
- ✓ **Five minutes, one sign up - we do the rest**



80+ advocacy group partners, and we are just getting started

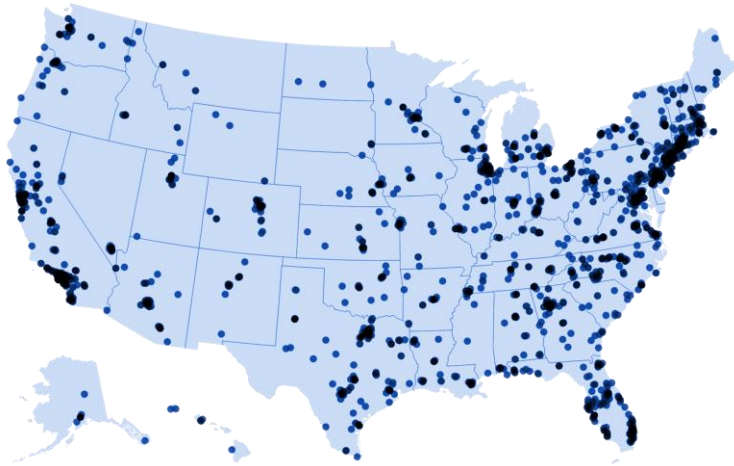
We have partnerships with over 80 patient advocacy groups, 70% of which have come to us organically.



Medical records retrieved from >3,000 unique institutions



Comprehensive and longitudinal medical record collection



- **Established broad relationships with healthcare providers across the US, where we have retrieved medical records from >3,000 unique institutions or offices**
- **Records encompass all data generated and stored within the EHR, and can include images and recordings**
- **Collection follows the patient throughout their journey; on average, medical records are obtained from multiple providers per patient**

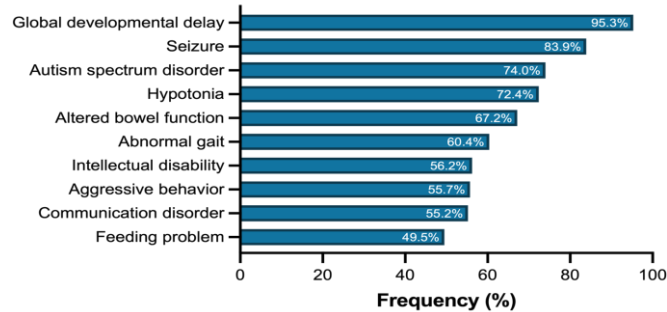
Map generated using Datawrapper¹

¹<https://app.datawrapper.de/>.

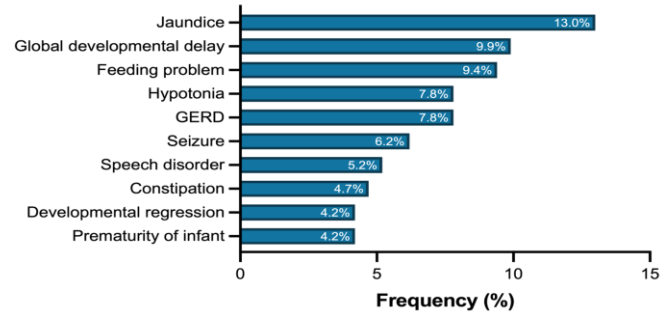
Pre clinical: characterize the condition and design a more effective study

Presenting diagnoses in Rett Syndrome

Most common diagnoses



Most common presenting diagnoses



Help researchers understand diagnosis and symptoms that can help design more effective models and studies

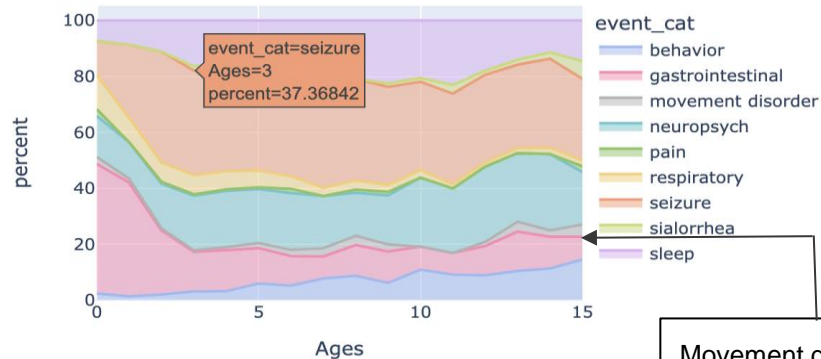
What is my patient population experience and what percent are experiencing them?

Pre clinical: Identify drug development opportunities

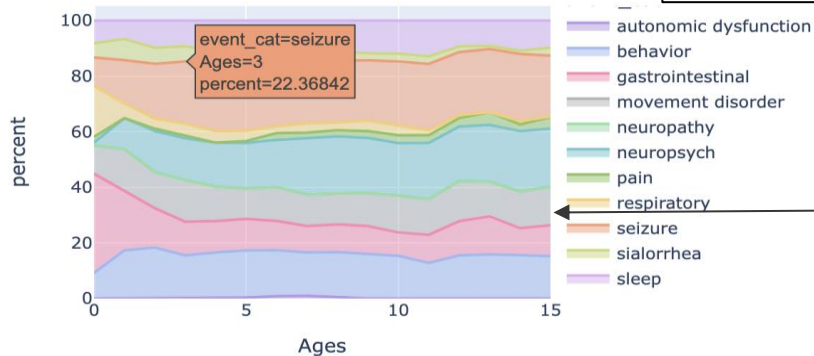
Generate a deeper understanding the treatment landscape and opportunities

What are the most common symptoms that are not being addressed by today's therapies?

SYNGAP1: categorized medication indications

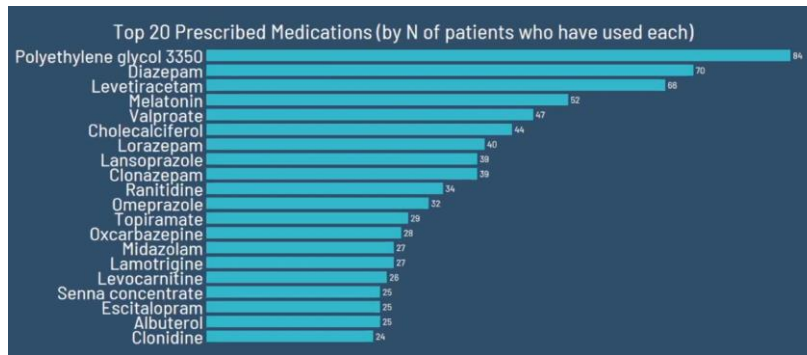
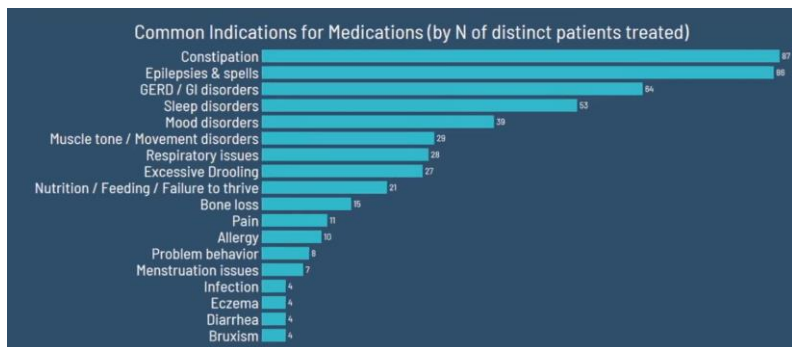


SYNGAP1: categorized diagnoses



Movement disorders a common symptom, but few correlating therapies

Pre clinical: Identify drug development opportunities



Generate a deeper understanding the treatment landscape and opportunities

What are the most common symptoms that are not being addressed by today's therapies? What may be a good endpoint?

Citizen enables immense research acceleration

Example Research Posters

- [Comparing phenotypes across five developmental and epileptic encephalopathies \(DEEs\) through evaluation of 2490 patient data years](#) (2022)
- [SYNGAP1-related disorder genotype-phenotype analysis through a digital registry](#) (2023)
- [A novel patient-centric longitudinal data registry platform generates insights into real-world cholangiocarcinoma \(CCA\) clinical practice](#) (2023)
- [A Novel Relationship between Interictal Epileptiform Discharge Burden and Gross Motor Developmental Delay in SCN2A Developmental and Epileptic Encephalopathy](#) (2023)
- [Expanding the phenotype of CHD2-related disorders through 97,010 phenotypic annotations](#) (2023)
- [Patient characteristics, treatment patterns, and outcomes in patients with cholangiocarcinoma](#) (2024)

Example Manuscripts

- [Characterizing a rare neurogenetic disease, SLC13A5 citrate transporter disorder, utilizing clinical data in a cloud-based medical record collection system](#) (2023)
- [Delineating clinical and developmental outcomes in STXBP1-related disorders](#) (2023)
- [Increasing Clinical Trial Participation of Black Women Diagnosed with Breast Cancer](#) (2024)
- [Comprehensive phenotypes of patients with SYNGAP1-related disorder reveals high rates of epilepsy and autism](#) (2024)
- [Autism gene variants disrupt enteric neuron migration and cause gastrointestinal dysmotility](#) (2024)
- [Validation and clinical discovery demonstration of breast cancer data from a real-world data extraction platform](#) (2024)



Hear from our patients and families



It took less than two minutes

My son's case manager nurse asked for a list of all of his diagnoses. I just had to pull his Citizen's neuro summary and share with her. It took me less than two minutes.

SYNGAP1 syndrome mom

Organized and searchable

A couple weeks ago my 13-year-old son was invited to be seen at a specialty clinic at Children's Hospital Colorado for the first time. They asked me to have his pediatrician fax over a referral and multiple documents. I said to the nurse, I can do better than a fax. I have my child's entire medical history available in Citizen. I shared his Citizen link with her and she then had access to everything organized and searchable through that link.

Rare disease mom

Such a relief

It is such a relief to know my extensive records are being collected in a central place and that they can be accessed when needed. I was diagnosed with bladder cancer this year and my records continue to grow. If I can help with any research that will find cures for these cancers, I want to participate. Knowing that my records can be accessed through Citizen makes it possible to help in such research studies.

Bladder cancer patient

I can stop repeating myself

We are constantly in and out of emergency rooms with our daughter due to seizures. It is emotionally draining to have to explain her past over and over. Now, I can log-in to her Citizen account, share her history with the doctors and nurses, and stop repeating myself.

SLC13A5 deficiency mom

I don't have to leave my home

A few years ago, I was asked to participate in a natural history study for our disorder. I had to take time off work, drive two hours, and leave my other kids at home. Honestly, it was too much. I like that with Citizen I can still contribute to research but don't have to leave my home.

FOXG1 syndrome dad

Central storage for reports and images

Just today I used images from Citizen to discuss my issues with a new provider. As you know, having many institutions trying to access my reports and images can be difficult... Having a central storage for these reports and images is critical when trying to keep at least 7 doctors up-to-date.

Cholangiocarcinoma patient