Rare Bootcamp™

Citizen Health

citizen health®

Meet Citizen

Unlock the power of health data to accelerate research and improve patient care for rare diseases

Challenging Landscape of Rare Diseases

10,000

Rare diseases

95%

have no treatment

30+ Million

Impacted patients

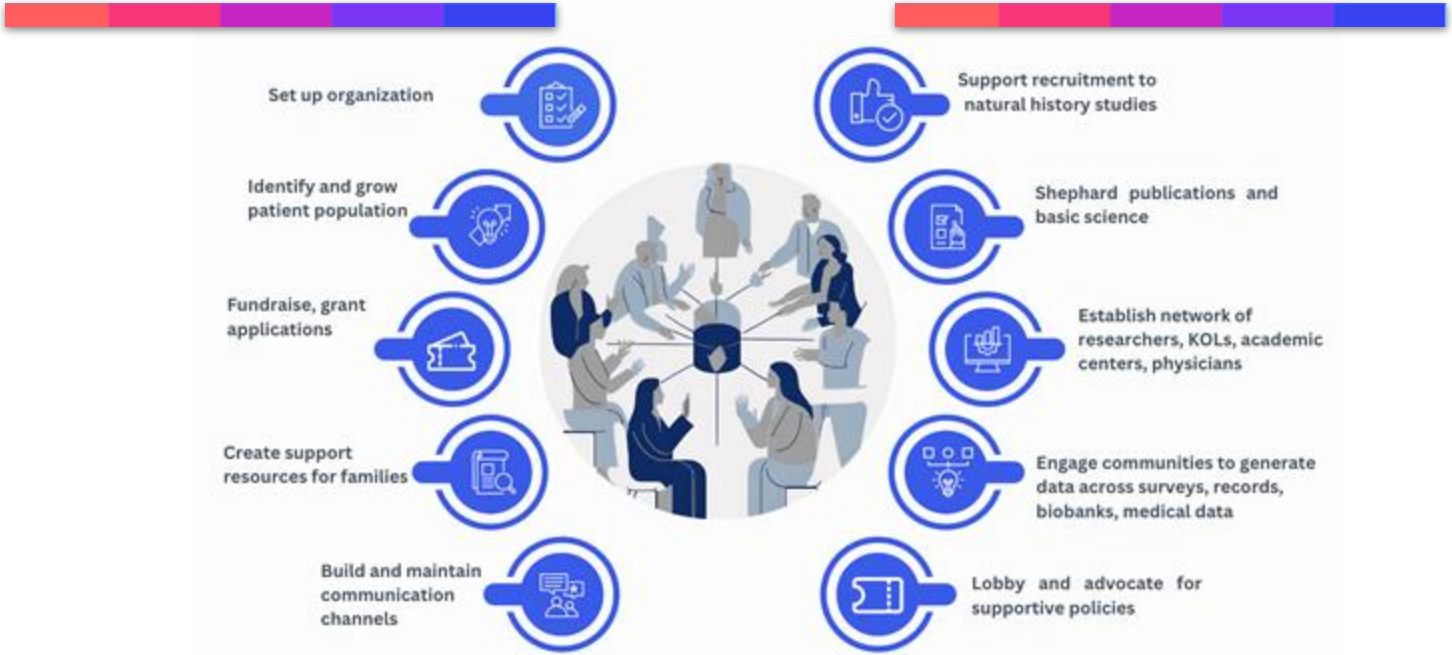
$1 Trillion

Economic cost of 379 rare diseases

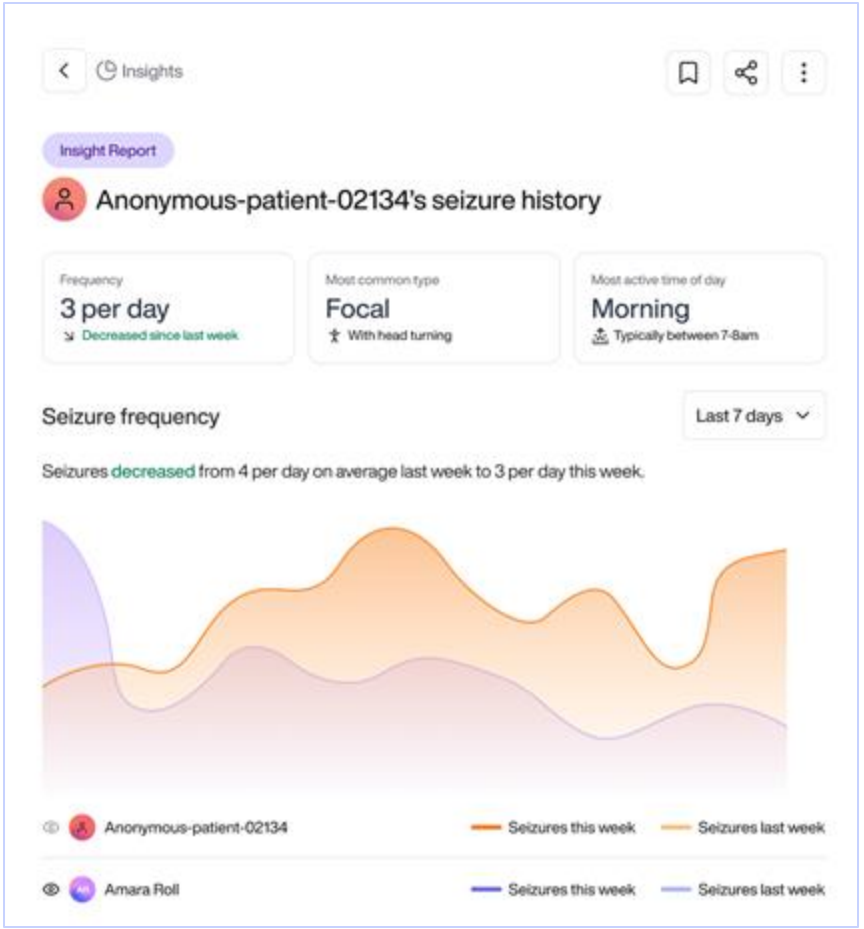
The immense responsibilities of Patient Advocacy Groups

Support patient communities through complex care journeys

Advance research with limited resources in a challenging landscape



Robust patient data is essential to support communities and drive research



Natural History Studies are a cornerstone of rare disease research

- Characterizing the full spectrum of disease manifestation
- Identifying biomarkers and outcome measures
- Supporting regulatory submissions
- Guiding patient care decisions
- Establishing baseline for measuring treatment effects

Natural History Studies are a cornerstone of rare disease research

$ Millions to conduct in person NHS studies

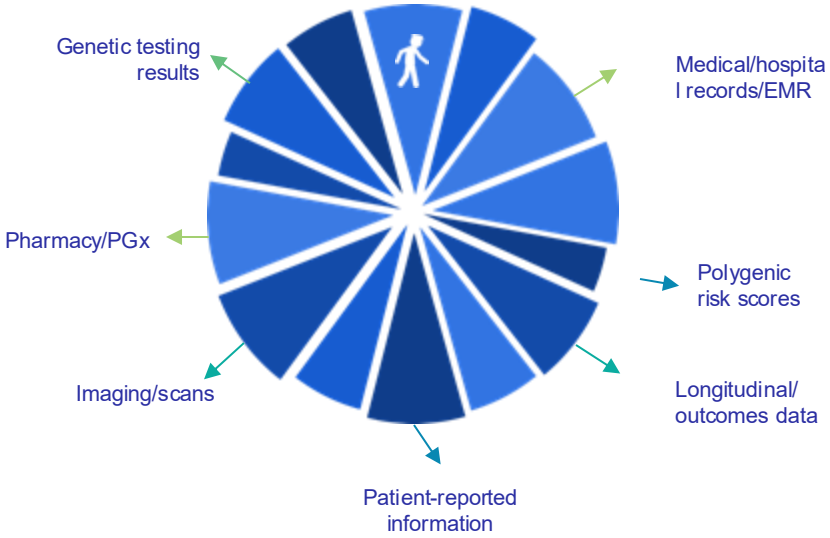
Millions of dollars just to support typical rare natural history study

High patient burden collection

Travel for in-person visits, repeat tests, long surveys,

- Characterizing the full spectrum of disease manifestation
- Identifying biomarkers and outcome measures
- Supporting regulatory submissions
- Guiding patient care decisions
- Establishing baseline for measuring treatment effects

Valuable patient data is trapped



Data is siloed and trapped

Across all the institutions a patient has received care

No single longitudinal view

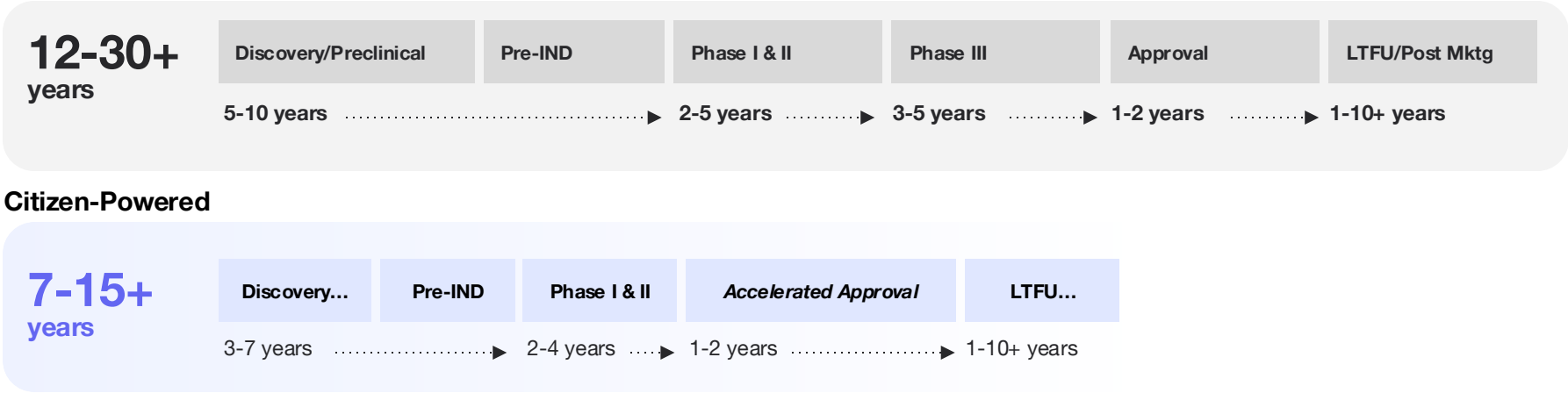
Of a patient's medical history to accelerate in person studies and support care

Slow and expensive drug development model

Empower advocacy groups and patients to **drive care and accelerate treatments** by **unlocking the power of their health data.**

- Make data accessible: Transform fragmented medical data into research-grade datasets and community driven insights
- Make data actionable: Enable **low-burden, digital studies** to compress drug development timelines
- Provide valuable tools for advocacy groups and their communities to support care journeys and research acceleration

Slow and expensive drug development model disrupted with Citizen

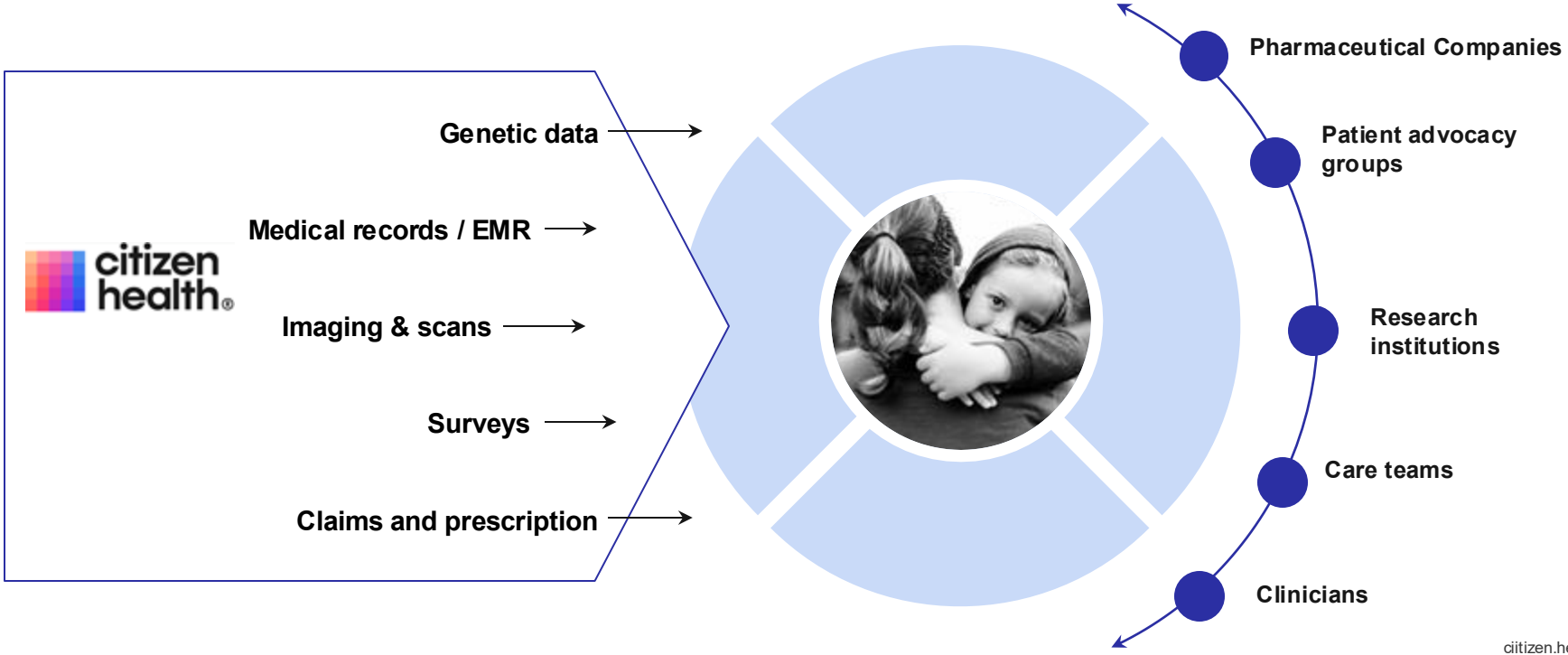


Rare Bootcamp™

How it works

How does it work?

Unlock powerful insights from rich medical data without patient burden



How does it work?

Collect Data

all patient's health data and upload it to their secure profile

Codify and Structure

the data to normalize it for research grade datasets

Drive Research

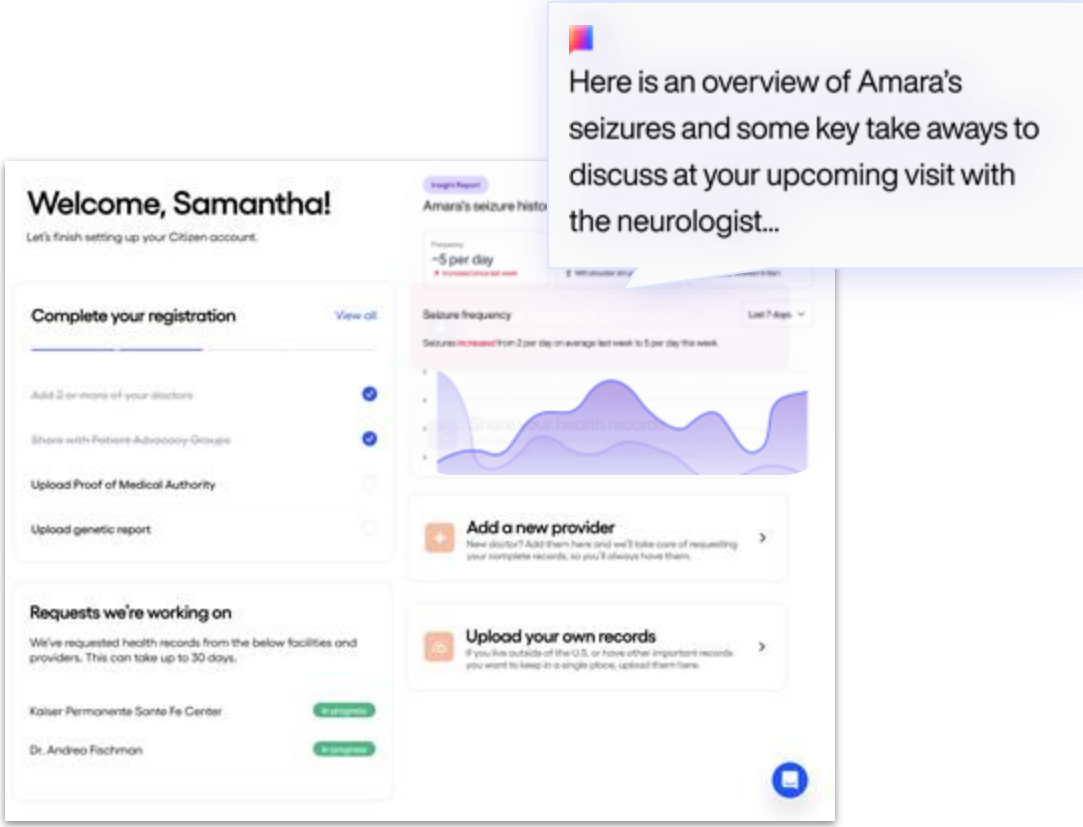
de-identify and share consented data to drive research

Enable Communities

provide patients and PAGs tools and insights to power care, community support and research

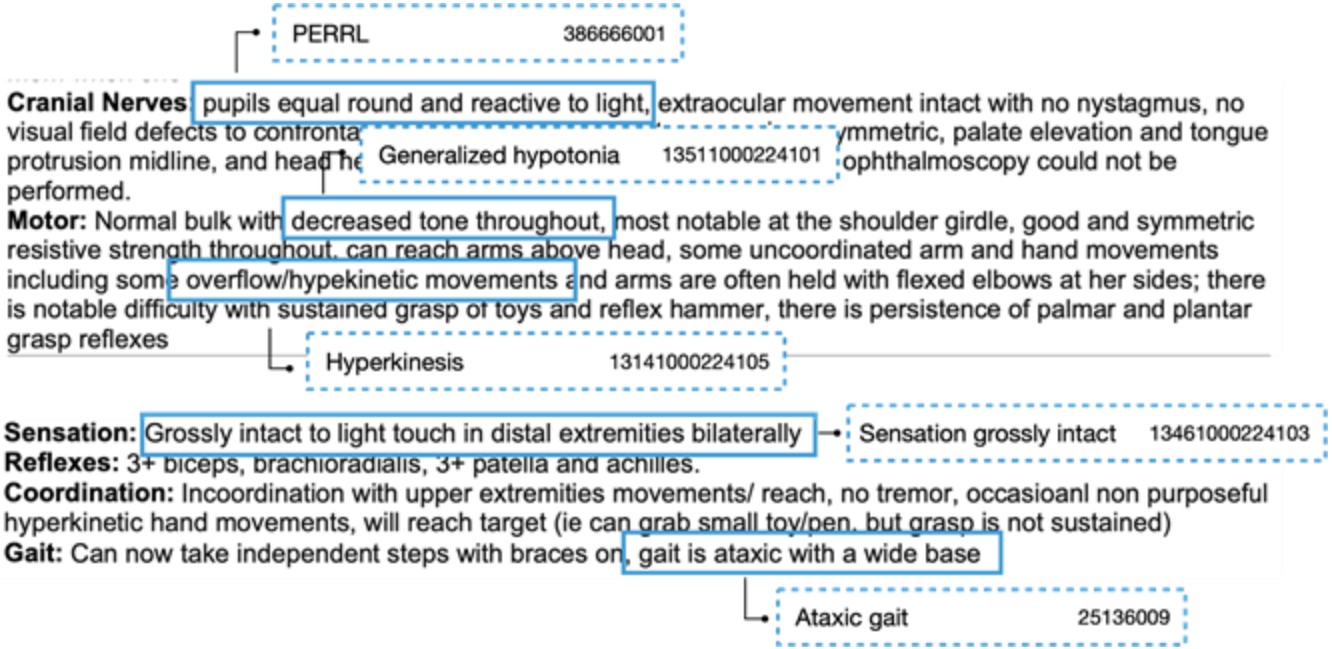
Patients

- No cost record collection
- Share easily and securely
- Real time chat with records insights and support
- Easily contribute to research without leaving home
- Revenue share



Research

Unlock powerful insights from rich medical data without patient burden

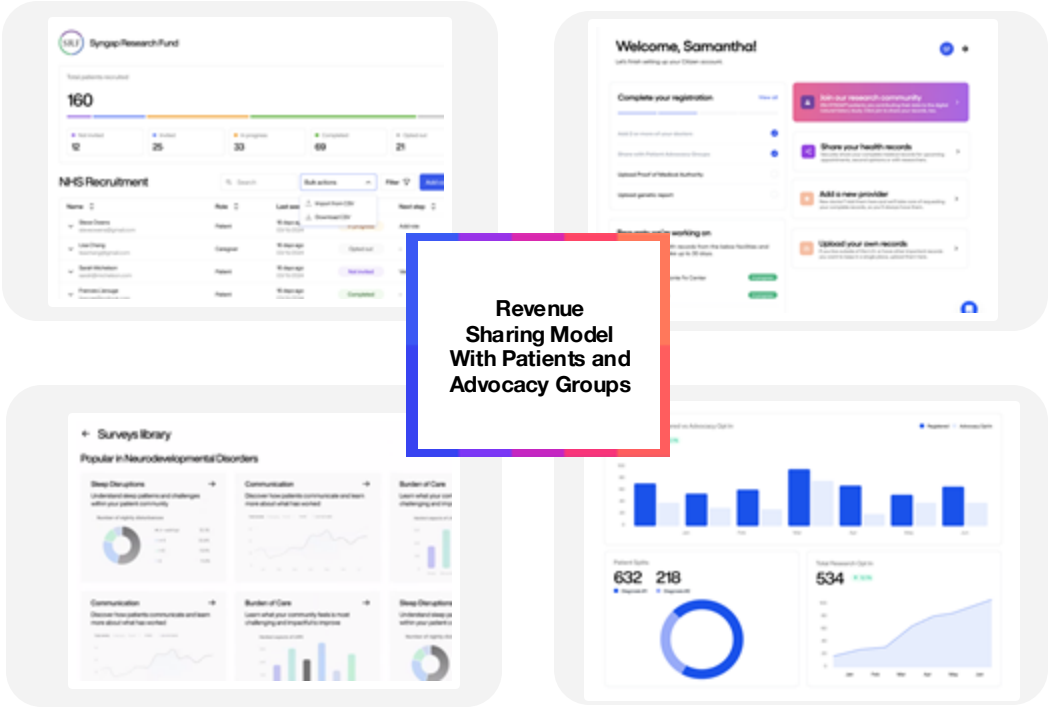


- Machine learning/NLP-assisted extraction engine that normalizes data
- Human assistance and review of pipeline generated data for QA/QC
- Terminologies such as SNOMED, RXNORM and LOINC used for normalization

Advocacy

Advocacy Group Partnership with Citizen

- **Registry platform** for community engagement
- Medical **data collection** and community **insights**
- Custom and validated **surveys**
- No patient burden, quick **Natural History Studies**

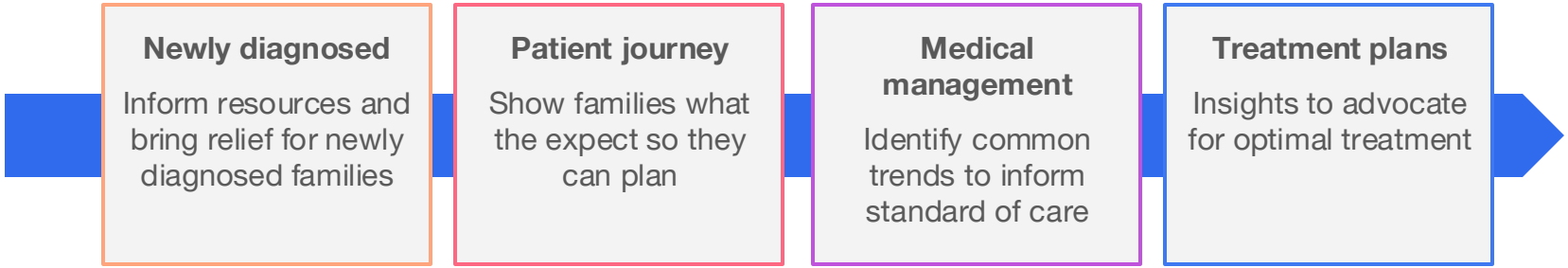


Rare Bootcamp™

The power of data

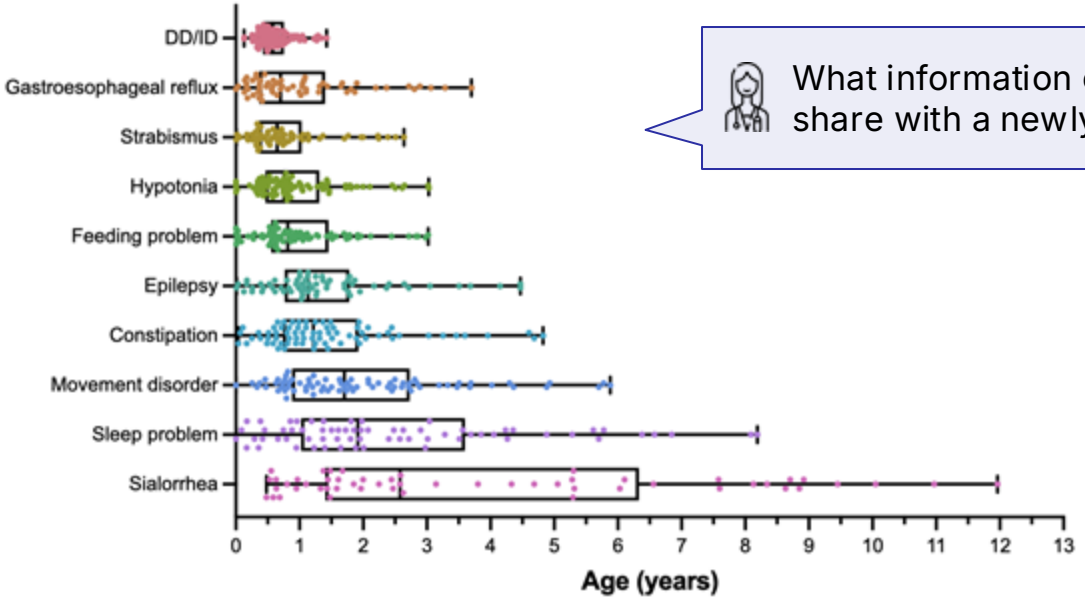
Robust patient data
is essential to support communities

Data that supports patient communities



Clinician & Community Support

We create **rich longitudinal** datasets that describe how symptoms **present** and **evolve** to power meaningful insights for patients, clinicians, and advocates.

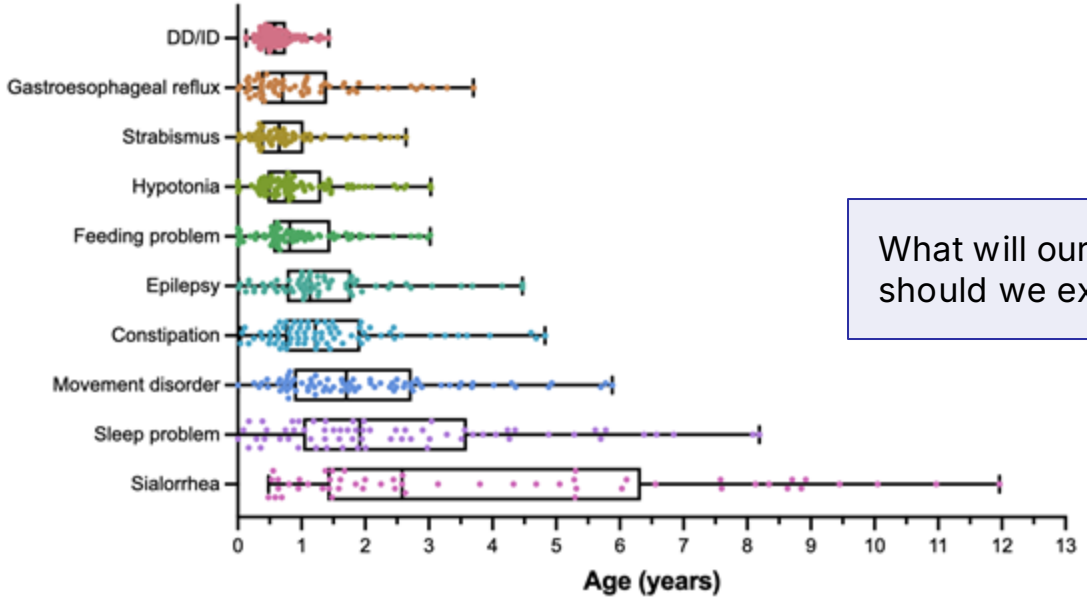


What information or resources can I share with a newly diagnosed family?

Newly diagnosed → Patient journey → Medical management → Treatment plans

Clinician & Community Support

We create **rich longitudinal** datasets that describe how symptoms **present** and **evolve** to power meaningful insights for patients, clinicians, and advocates.



What will our future look like? What should we expect now? In 10 years?

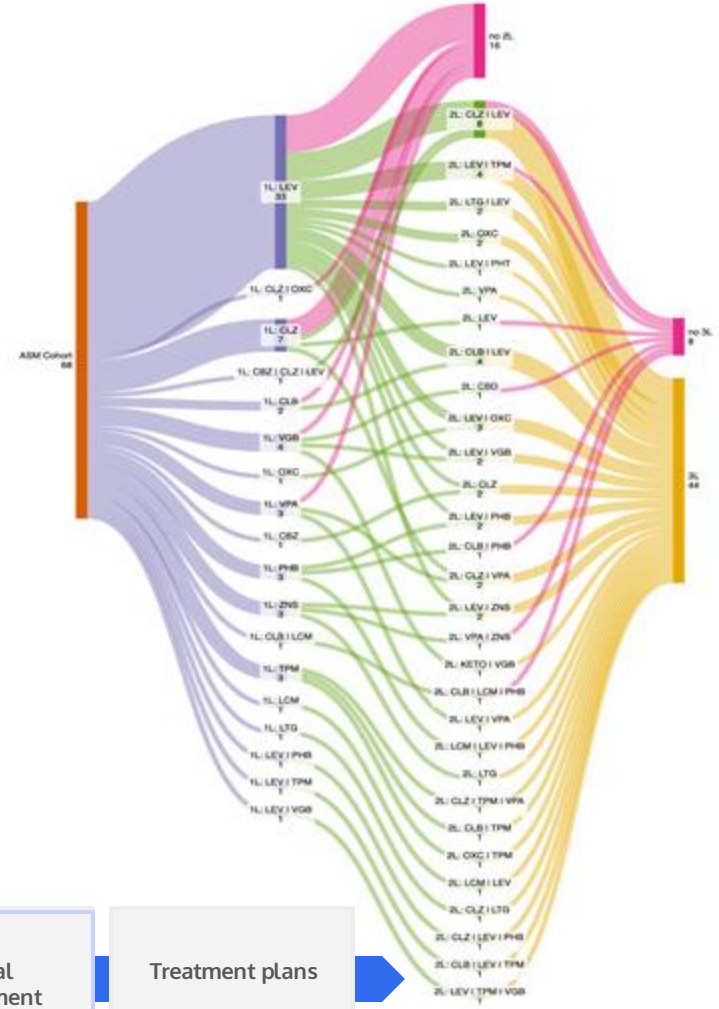
Newly diagnosed | Patient journey | Medical management | Treatment plans

Clinician & Community Support

Characterize treatment patterns and outcomes to inform medical management

We can explore treatment patterns across cohorts to understand the following:

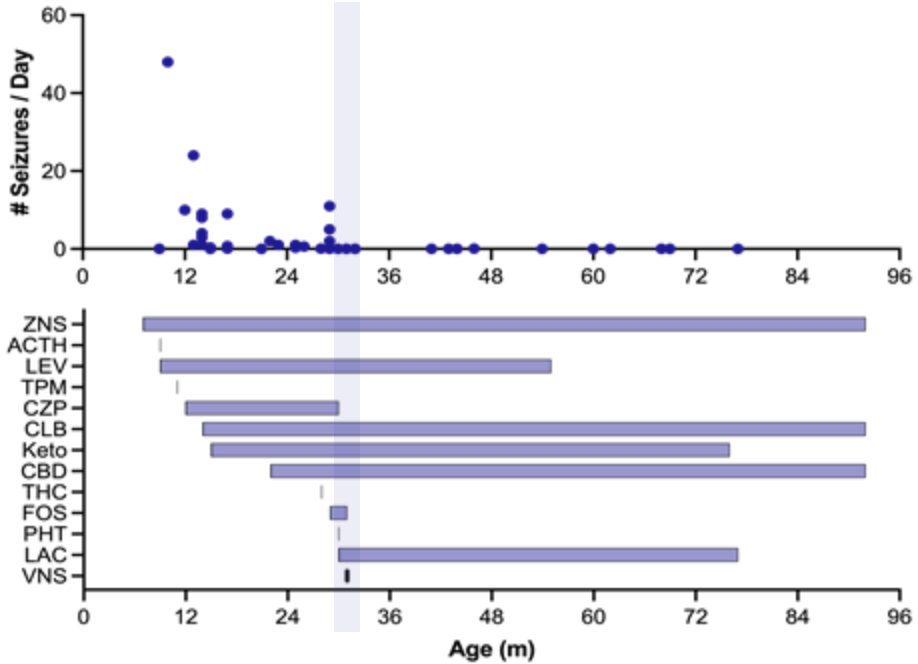
- Are there opportunities to drive better standardization in seizure management?
- Are certain ASMs or combinations of ASMs used more commonly?
- How many ASMs do patients trial?
- What is the use of polypharmacy in the population?



Newly diagnosed → Patient journey → Medical management → Treatment plans

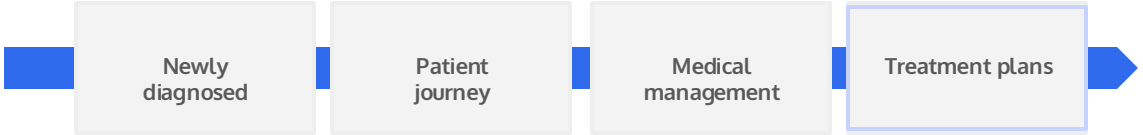
Clinician & Community Support

Characterize treatment patterns and outcomes to inform medical management



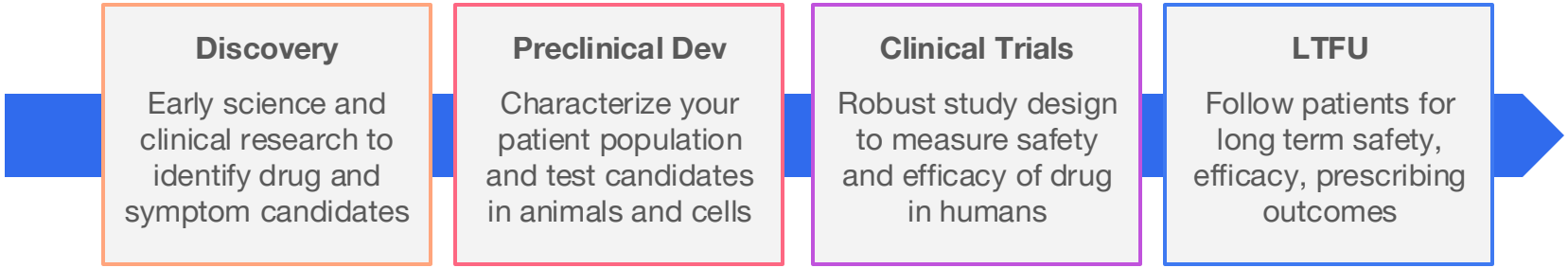
We can overlay key events to describe **patient journeys** for individuals and across cohorts:

- Illustrate **patterns** in key outcomes over time
- Explore associations between interventions and outcomes
- Explore relationships between phenotypes of interest (i.e. seizure frequency and developmental outcomes)



Robust patient data
is essential to drive
research

Data supports across all stages of drug development



Data supports across all stages of drug development

Accelerating the research journey

Answers you need

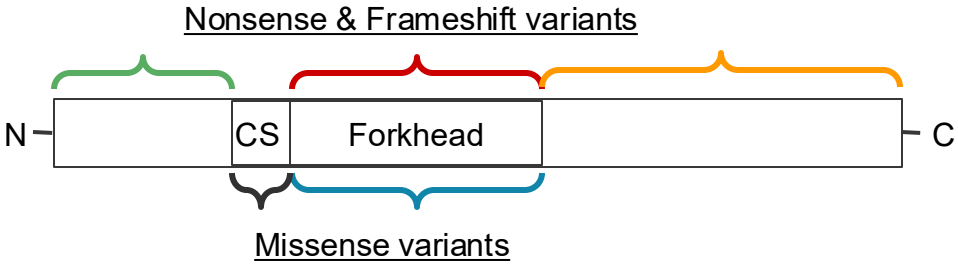
- What are the different variants?
- Who makes up this condition? What are the key demographics?
- What are the most common diagnosis and symptoms?
- What are the most frequent medications and are they working?
- Can I demonstrate the burden on the family and health system?

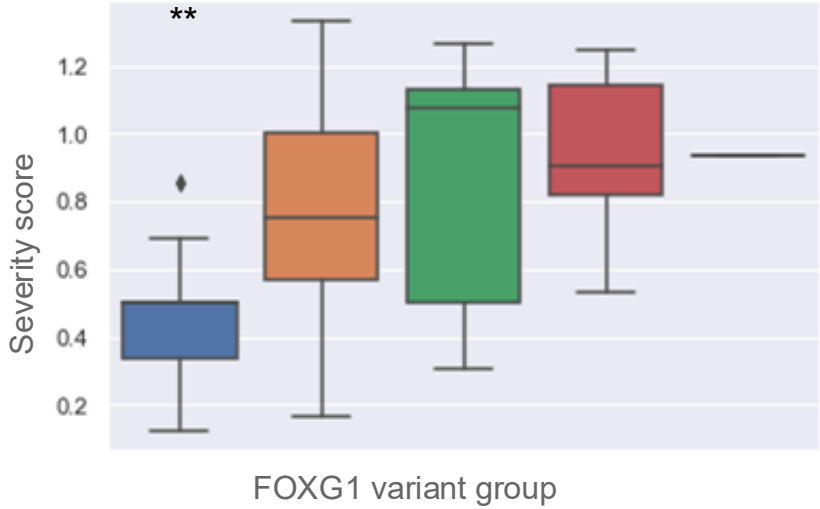
To drive better, faster research

- Identify the right clinical end points
- Choose the most promising drug development opportunity
- Design a more effective trial
- Include the right study questions and tests to minimize patient burden and increase success of the study
- Choose the right I/E criteria

Accelerate Preclinical Development

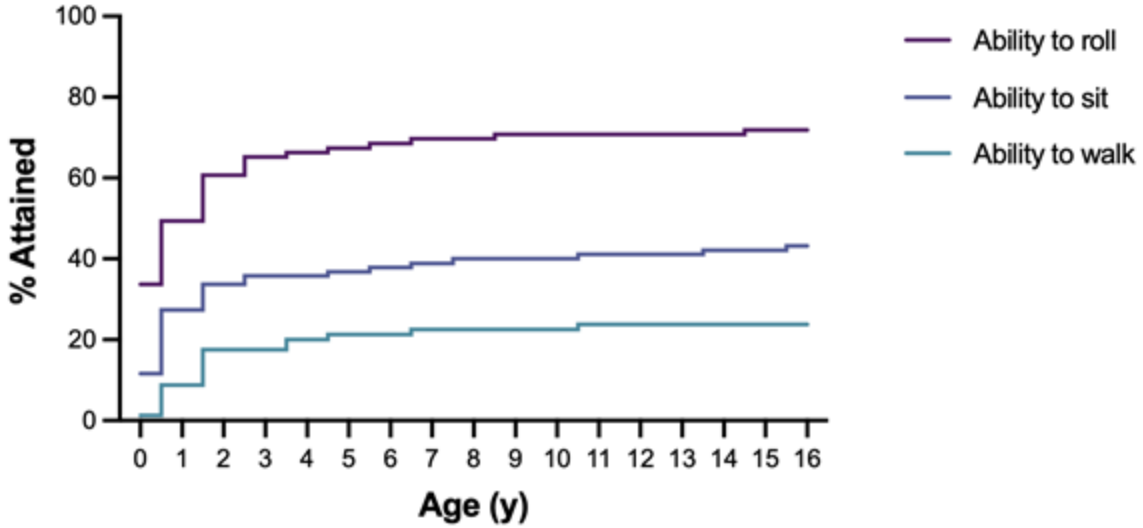
Genotype/Phenotype correlation work led to understanding of which models to build





Optimize Clinical Trial Design

- **Background:** Should gross motor development be a clinical endpoint?
- **Planned approach:** Assess gross motor function by age to understand trajectory of disease
- **Findings:** Gross motor function stays stable and can serve as a solid clinical endpoint



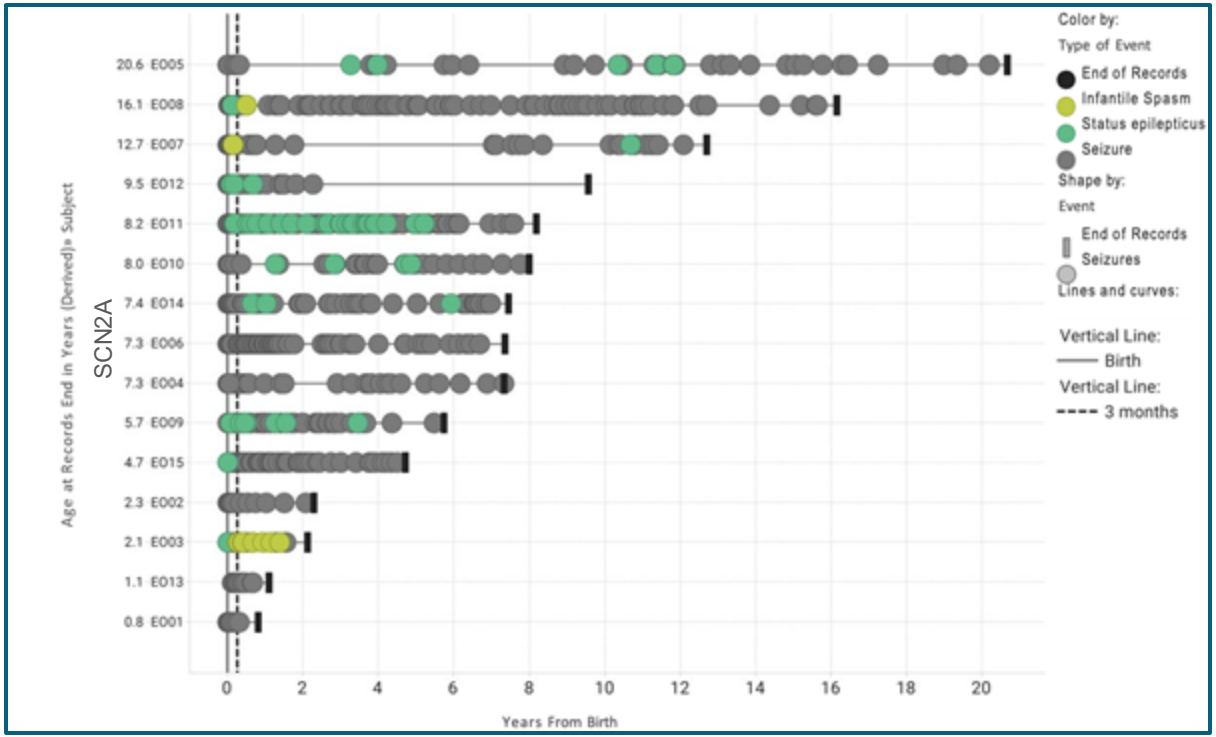
Discovery
Early science and clinical research to identify drug and symptom candidates

Preclinical Dev
Characterize your patient population and test candidates in animals and cells

Clinical Trials
Robust study design to measure safety and efficacy of drug in humans

LTFU
Follow patients for long term safety, efficacy, prescribing outcomes

IND Enabling: Select the right endpoints



- Literature suggests early onset seizures remit
- Data found seizure frequency persists through life
- Data helped confirm **primary endpoint** and **age criteria**

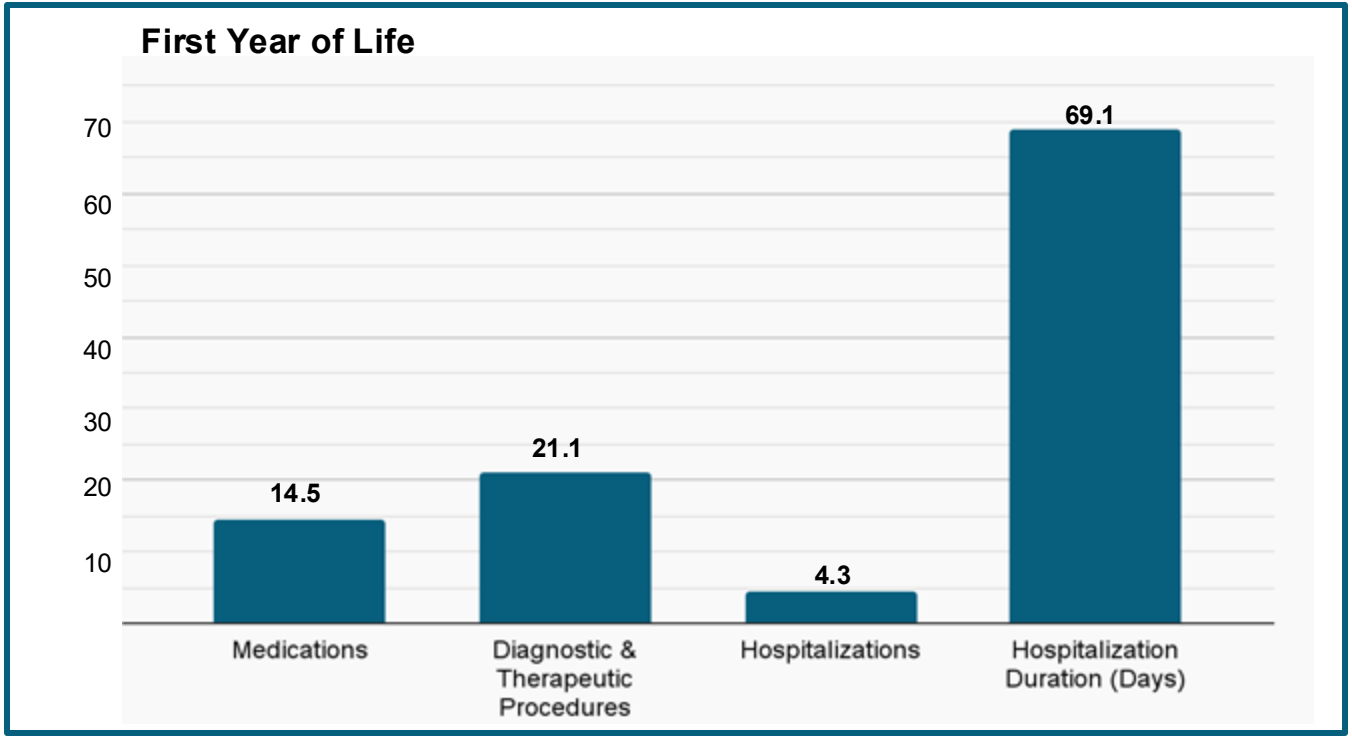
Discovery
Early science and clinical research to identify drug and symptom candidates

Preclinical Dev
Characterize your patient population and test candidates in animals and cells

Clinical Trials
Robust study design to measure safety and efficacy of drug in humans

LTFU
Follow patients for long term safety, efficacy, prescribing outcomes

IND Enabling: Demonstrate Disease Burden



Discovery
Early science and clinical research to identify drug and symptom candidates

Preclinical Dev
Characterize your patient population and test candidates in animals and cells

Clinical Trials
Robust study design to measure safety and efficacy of drug in humans

LTFU
Follow patients for long term safety, efficacy, prescribing outcomes

IND Enabling: Demonstrate Disease Burden

Praxis obtains Investigational New Drug clearance from FDA 4 years early with Citizen Data

- Citizen partnered with Praxis in December 2020 and delivered full natural history data **within 6 months,** all digital and no site visits required
- Citizen data was the **single source of natural history data** used in FDA submission package
- FDA approved use of RWD to completely replace in-person NHS for first-ever time in rare diseases

A 4-year acceleration is life-changing for children who now get access to first-ever treatment

In less than 3 years, Citizen enabled immense research acceleration for rare diseases

11 Manuscripts

- Delineating clinical and developmental outcomes in STXBP1-related disorders. *Brain*. (2023)
- Comprehensive phenotypes of patients with SYNGAP1-related disorder reveals high rates of epilepsy and autism. *Epilepsia*. (2023)
- Computation of longitudinal phenotypes in 466 individuals with a developmental and epileptic encephalopathy enables clinical trial readiness. *Epilepsia*. (2023)
- Increasing clinical trial participation of Black women diagnosed with breast cancer. *J. Racial Ethn. Health Disparities*. (2023)

25 Abstracts

- Real world patterns of oncogenomic testing in cholangiocarcinoma: pilot of a novel study approach. *Mayo Clinic Individualizing Medicine Conference*.
- Feasibility and preliminary findings of a novel, patient-centric registry model to address real-world care questions for patients with hematologic malignancies. *American Society of Hematology Annual Meeting*.
- A Novel Relationship between Interictal Epileptiform Discharge Burden and Gross Motor Developmental Delay in SCN2A Developmental and Epileptic Encephalopathy. *American Epilepsy Society Annual Meeting*.
- Data integration across healthcare resources reveals the unique phenotypic landscape of SYNGAP1-related disorders. *American Epilepsy Society Annual Meeting*.

Rare Bootcamp™

Thank You

Kate.Haldeman@citizenhealth.com
Phone: 410 852-1192
www.citizenhealth.com

Appendix

Natural History Data

Core NHS (no cost)

- Year of birth
- Gender
- Zip-3
- Self-reported diagnosis
- Genetics
- Conditions (unique)
- Symptoms (unique)
- Medications (unique)

Extended NHS (fee based)

- Conditions (longitudinal)
- Symptoms (longitudinal)
- Medications (longitudinal)
- Procedures
- Hospitalizations
- Growth parameters
- Development
- Standardized tests
- Seizure history
- Physical exam findings
- Laboratory studies