

## Abstract

### Background:

With ultra-rare neurodegenerative disorders such as Canavan disease, the research community all too often has little understanding of the patient voice. Families are faced with few opportunities to engage in meaningful dialogue about their experience with Canavan disease or raise awareness of their needs. In October 2016, Canavan Research Illinois and AltaVoice (formerly PatientCrossroads) launched the Canavan Disease Research Patient Insights Network (PIN) to unite families throughout the Canavan community and create a Canavan network that prepares the community for forthcoming research and therapeutic development.

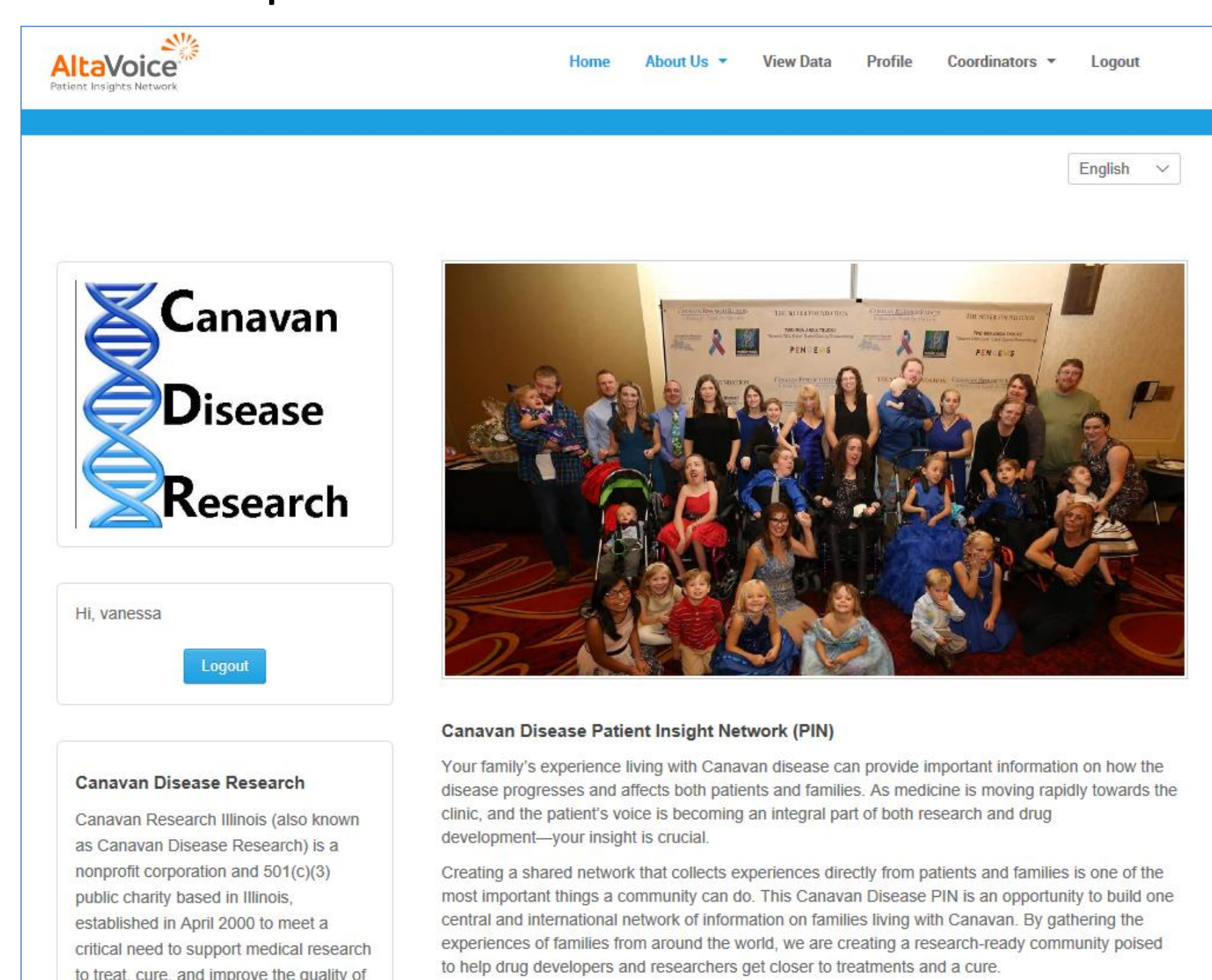


Figure 1. Homepage for the Canavan PIN (<https://connect.altavoice.org/?org=canavan>).

## Design

Using AltaVoice's<sup>1</sup> proven HIPAA and FISMA compliant advocacy partner platform, Canavan Research Illinois invited families from around the globe and organizational partners to join the web-based opt-in PIN (<https://connect.altavoice.org/?org=canavan>). Once registered, participants control their data-sharing and contact preferences and contribute their de-identified data openly through a dedicated PIN Data Portal and infographic charts.

- Provides each user a password protected account
- Add dependents; extended family members linked via a family code
- Collects longitudinal survey data, links healthcare providers, allows upload of medical records
- Access to de-identified registry data by participants and professionals
- Available in English, Portuguese; other languages via Google Translate



Figure 2. Canavan PIN supporting organizations



Figure 3. Canavan PIN QR code

### Surveys:

Surveys include common data elements from the NIH GRDR Common Data Elements, RD-Connect data elements, and review of systems questions (GenomeConnect, a program of The ClinGen Resource).

- Core surveys: diagnosis, medical care, family history, general health
- Demographic data collected to characterize participants
- Asks sufficient questions to trigger supplemental surveys
- Not overly lengthy or burdensome to encourage maximum participation and completeness

## Participant Overview

To date, 118 participants have enrolled worldwide, of which 90% (106) have set up accounts. Of these participants, 97 (82%) have completed one or more surveys.

Participant ages: 0-2 years, 26 (22%); 3-5 years, 14 (12%); 6-10 years, 31 (26%); 11-20 years, 29 (25%); 21-40 years, 18 (15%)  
 Average age: 11 years; median age: 8 years



Figure 4. To date, 107 participants have provided their country, of which 44 (41%) are from the United States and 63 (59%) are from countries other than the United States.

- >91% Participants chose to share de-identified data
- 88% Participants want to be contacted for clinical trials & research studies

## History of Diagnosis

Participants provide valuable insights about their diagnosis, medical care and medical history. A selection of participant-provided data is provided.

Participants describe the history of their diagnosis:

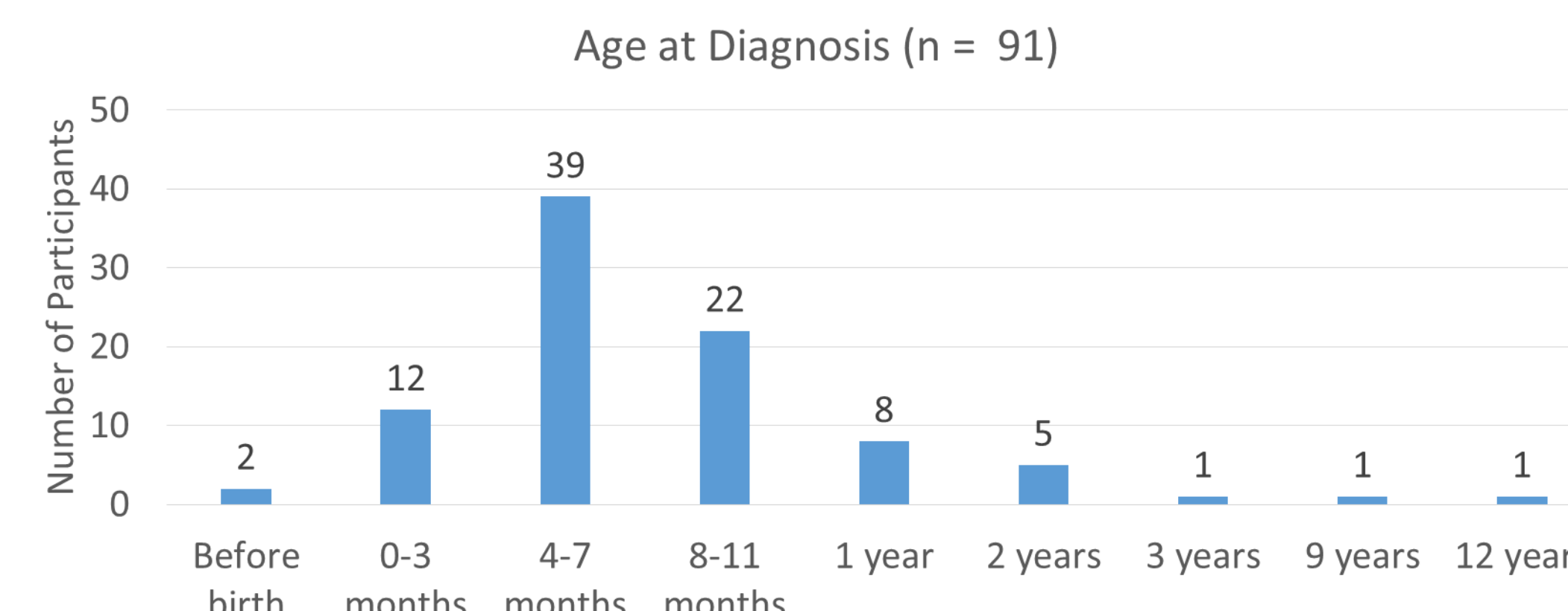


Figure 5. Participants report the age at diagnosis.

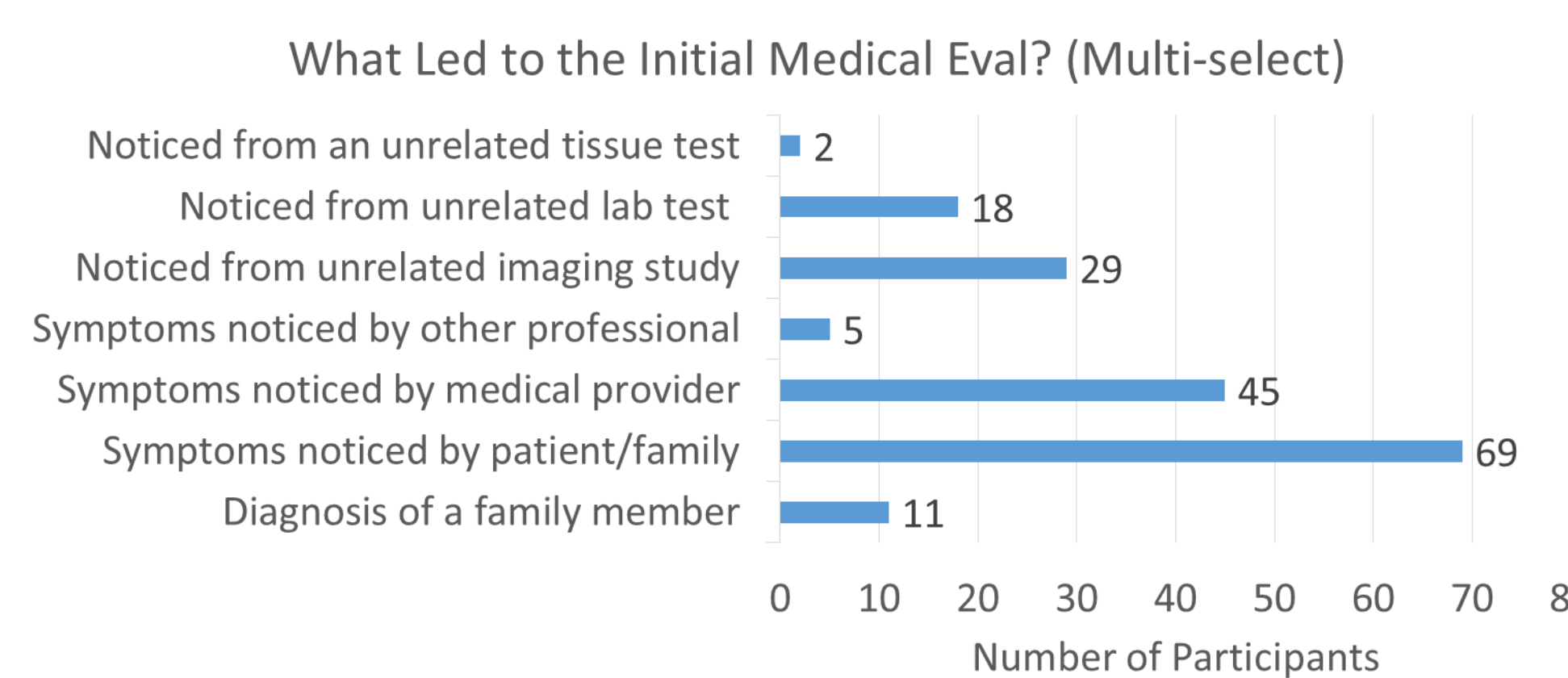


Figure 6. Participants report what led to the initial medical evaluation.

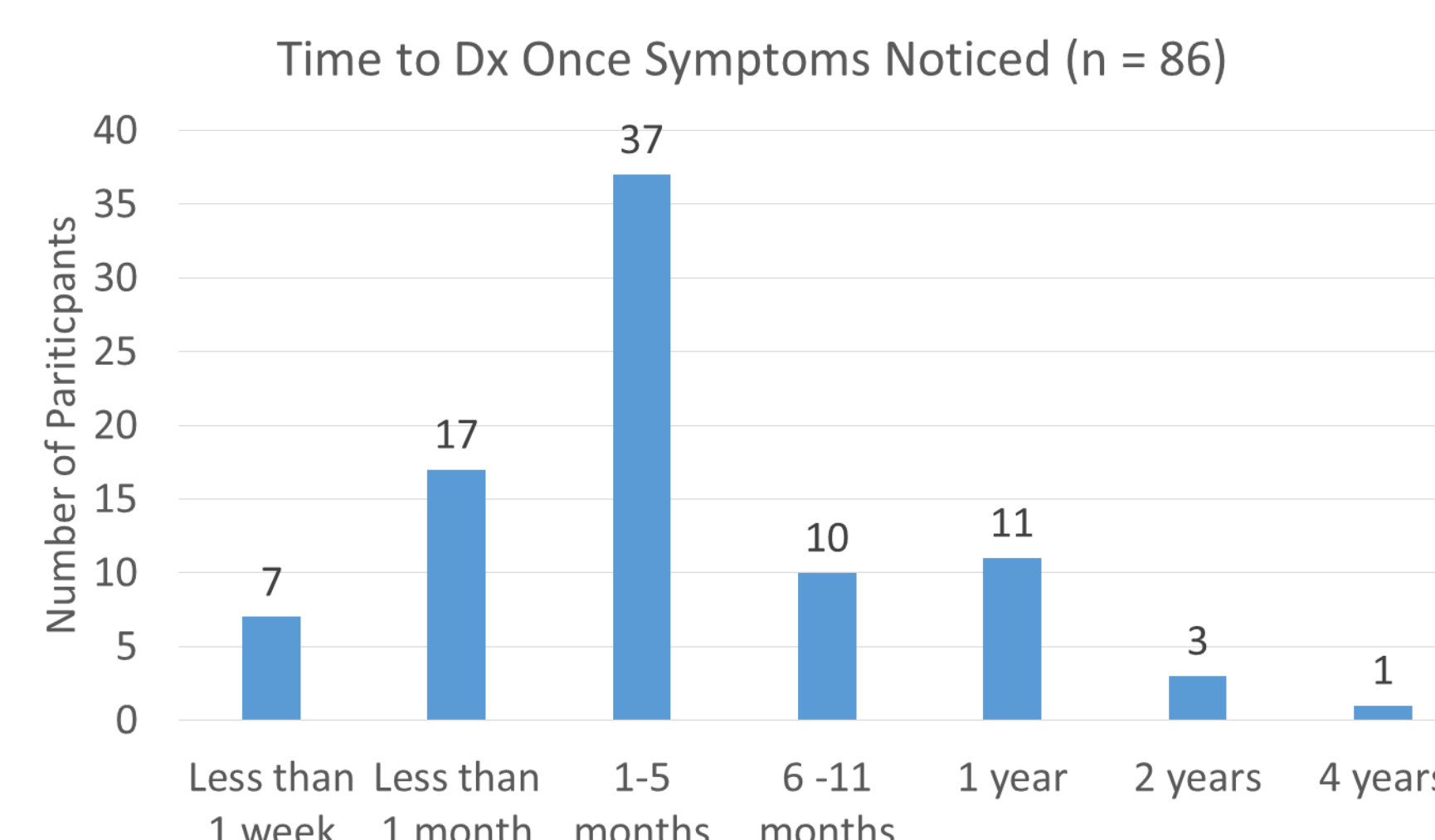


Figure 7. Participants report the time it took the provider to make a diagnosis once symptoms were first noticed.

## Current Health and Impact

Participants share their current health and medical care. They report seeing a healthcare provider with the following frequencies: 1 or more times per week (7%); 1-3 times per month (31%); 2-6 times a year (36%); 7-11 times a year (19%); annually (2%).

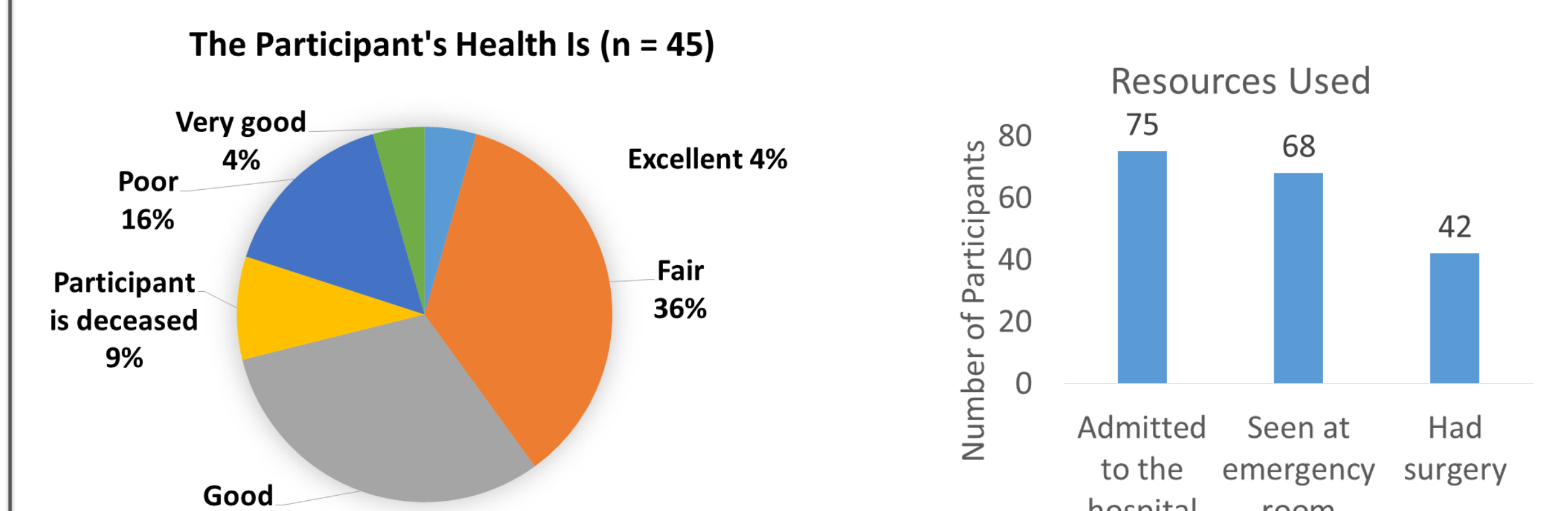


Figure 8. Participants report their current health.

Figure 9. Participants report medical events.

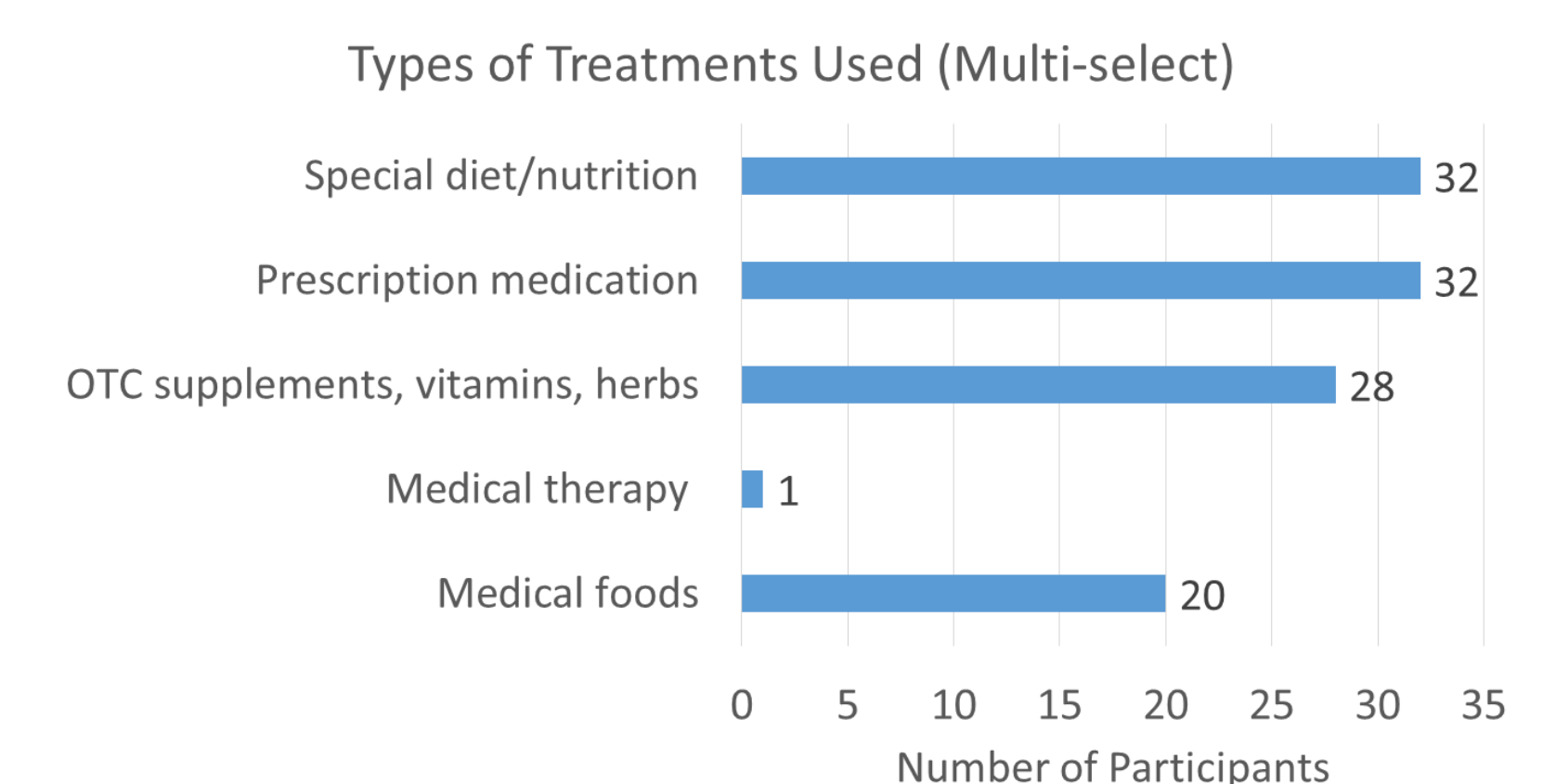


Figure 10. Participants report treatments used for Canavan disease.

Participants report the financial impact of caring for a loved one with Canavan disease.

Estimated YEARLY out-of-pocket expense	Number of Participants
None	9
Less than \$500	5
\$500 - 1,000	2
\$1,000 - 2,500	4
\$2,500 - 5,000	4
\$10,000 - 25,000	1
\$25,000 - 50,000	1
\$50,000 - 75,000	2
Greater than \$100,000	1
Prefer not to answer	3
Unsure	10

Figure 11. Participants report yearly out-of-pocket expense for Canavan treatment and care.

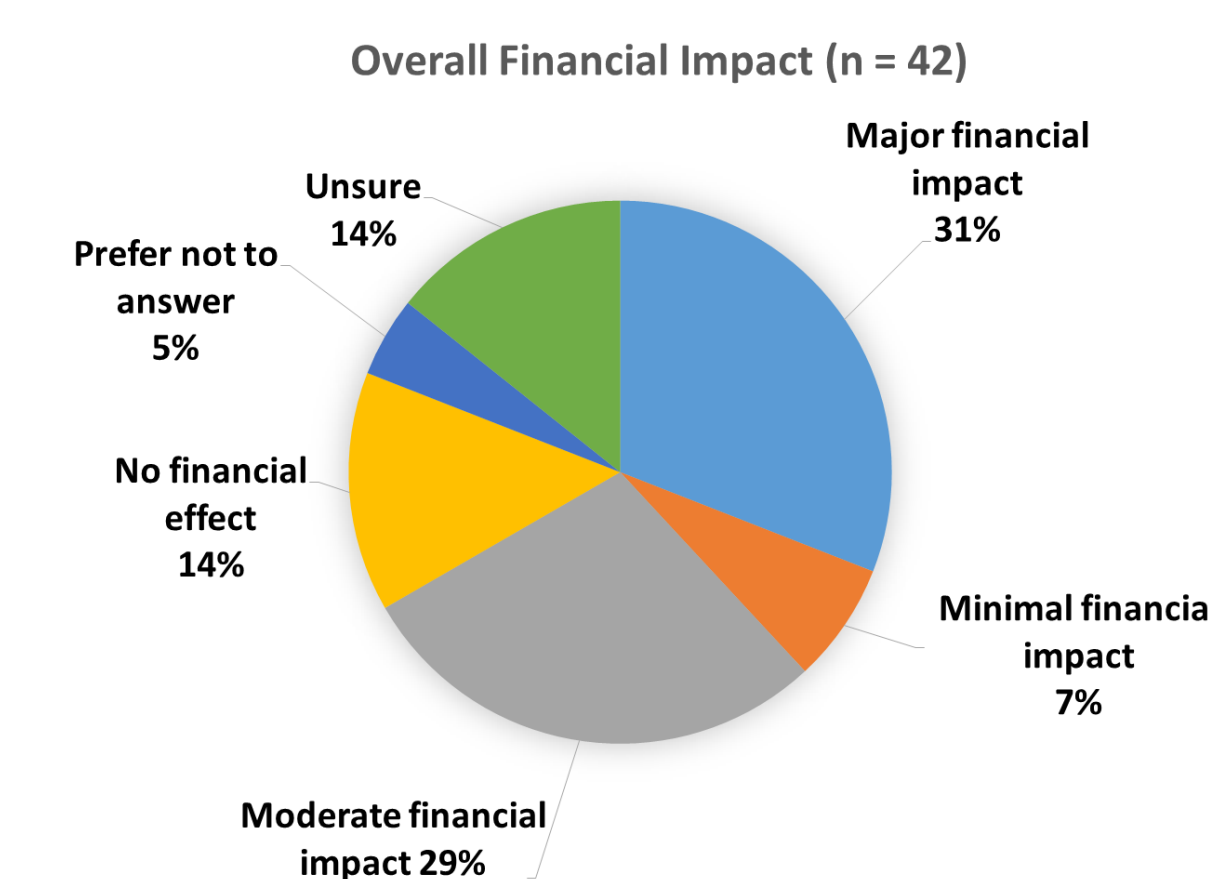


Figure 12. Participants report the overall financial impact of Canavan disease.

## Conclusions

This successful recruitment represents the largest patient-centered cohort known to be established for Canavan disease, and it has demonstrated the willingness of participants to share their health data, engage in the formation of an online resource, and request communications about clinical trial and research activities. As therapeutic development in Canavan disease expands, including gene therapy and biologic trials, a research-ready community will be needed. By organizing the community, this Canavan disease PIN offers a first ever shared resource that can amplify the voice of patients and families, to raise awareness and accelerate interest in therapeutic development.

### Acknowledgements:

We gratefully acknowledge the PIN participants and their families and the support of the advocacy organization partners [Canavan Research Illinois, National Tay-Sachs and Allied Diseases Association, Canavan Foundation, Rare Diseases Foundation]

<sup>1</sup>AltaVoice, formerly PatientCrossroads, is a wholly owned subsidiary of Invitae Corporation