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**Is the CADASIL Family Registry Research Ready?**

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**BACKGROUND**

In 2015, an online global family registry was created for the neurodegenerative Cerebral Autosomal Dominant Arteriopathy with Telangiectasia (CADASIL) disorder. The CADASIL Family Registry is a collaboration between the nonprofit patient organization, cureCADASIL, and Invitae (formerly Genomic Medicine), using their CONNECT platform. Invitae has more than 80 data registries across all rare and common diseases.

**OBJECTIVES**

This study assesses whether the registry data can be used to support cross-sectional or longitudinal research studies, genetic studies, and clinical trials.

**METHODS**

Registry and Data Sets

This study assesses whether the registry data can be used to support clinical trial enrollment.

- General Health
- Medical Care
- Family History
- Guardian can legally make medical decisions

Participants most commonly reported being white (33.5%) and are female (58.8%). Nearly half of participants did not provide contact info only with a median age of 50 years (Table 1) (Figure 3).

**RESULTS**

Contact Database

The registry includes 370 contacts worldwide, with 284 participants providing data to the de-identified survey study with research use.

Survey completion was between June 2015 and June 2018. Among registry participants, the response rate was highest for the Diagnosis survey, allowing for identification of participants who meet broad inclusion criteria.

**CONCLUSIONS**

More than 265 individuals have provided multiple surveys in the registry, allowing the identification of participants that meet clinical trial enrollment criteria.

- Questions on genotypic, phenotypic, diagnostic, or medical criteria can be submitted in the registry survey (under “research study” or “other research studies”).
- Limited patient reported clinical data specific to CADASIL (e.g., time since sympotms onset) or comorbidities (e.g., available that could be used for study enrollment).
- General health survey data from patient medical records is available. In addition, it can be made available to investigators only. These are curated only when extensions of the registry are developed and in place with patient consent.

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**AUTHOR DISCLOSURES**

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