NEWS

# Large genetic registry may uncover autism’s diverse origins

BY **ANN GRISWOLD**

21 APRIL 2016

Autism researchers are reaching out to families to build the nation’s biggest repository of genetic information to date from individuals with the condition.

Unveiled today, the ambitious new project, called **SPARK**, aims to gather information from 50,000 people with autism, along with their relatives. (SPARK is funded by the Simons Foundation, *Spectrum*'s parent organization.) About 500 families signed up to participate during the project's pilot phase, launched in December.

The participants can mail in saliva samples for genetic testing — a convenience that could attract people who are currently **underrepresented** in research studies, such as minorities and those who have severe symptoms, says lead investigator **Wendy Chung**, assistant professor of pediatrics at Columbia University in New York.

Some of these groups tend not to participate in research because of the time commitment, cost of travel or stress involved. But without a large and diverse pool of participants, researchers cannot identify the various causes of autism, says **Jeremy Parr**, clinical senior lecturer at Newcastle University in the United Kingdom, who is not involved with the project. Parr works with the **Autism Spectrum Database-UK**, a registry of clinical information from more than 1,000 families that have a child with autism.

Even when recruitment is complete, the project will represent only a small fraction of the estimated 3.5 million Americans who have autism[1]. Still, the project's size is a "significant step forward," Parr says.

## Simple sign-up:

To participate in SPARK, individuals with autism or their caregivers must provide demographic information, describe their extended family's medical and psychiatric histories and complete the **Social Communication Questionnaire**, which measures social skills, all via an online portal. The family also uploads each participating member's health records and confirms, by checking a box, that the person with autism has a clinical diagnosis. Participants can also register in person at one of the project's 21 sites.

"The ability of so many individuals to get online, either on their smart phones or from work or home, has allowed us to do things that we could not have done five years ago," says Chung.

However, not everyone has access to this technology, and the project may end up leaving out some of the participants whom researchers most want to reach. "It may miss patients who have a socioeconomic disadvantage," says **Nalin Payakachat**, associate professor of pharmacy at the University of Arkansas for Medical Sciences in Little Rock, who is not involved in the project.

Although uploading medical histories online is convenient, "it may not be as reliable as enrolling patients from clinics and obtaining their medical information from medical records," she adds.

To meet their interim goal of enrolling 10,000 participants by next year, the researchers are using

recruitment tactics ranging from hosting events to posting on social media. Once enrolled, participants ship saliva samples (using a kit) to one central lab for analysis. Researchers then extract DNA and sequence the protein-coding portion of each genome.

“It’s about as easy as anyone could make it,” says **Dianna Varady**, director of the Arkansas Autism Resource and Outreach Center and mother of a 12-year-old boy with autism. The researchers plan to share the genetic results with families, which might help them make decisions about treatment, Varady says.

## Revealing relationships:

A key aspect of the project is scientists’ ability to recruit registered members for future studies. The participants update their information each year, which should create a “richer, deeper and more dimensional” resource over time, says Chung.

Because of the group’s size, researchers may also be able to probe relationships between genes and clinical symptoms. For example, **Stephen Kanne**’s team at the University of Missouri plans to analyze the data to understand how autism genes might contribute to language development or responses to treatment. And **Robin Kochel**’s team at Texas Children’s Hospital aims to use demographic information from the participants to examine how a family’s cultural beliefs and geographical location might influence its members’ approach to intervention.

Another registry, the **Interactive Autism Network** (IAN), has also collected behavioral and clinical information through online surveys of 20,000 people with autism plus their family members. However, most IAN members have not provided any genetic samples.

Some of the participants are likely to be members of other registries, such as IAN or the **National Database for Autism Research**. The researchers can pinpoint these participants because each carries a **unique identifier**, or GUID.

By cross-referencing data from these registries for each participant, researchers might be able to piece together a complete portrait of families with autism. Using such a strategy, Payakachat says, “can catalyze relationships not envisioned by the original data collectors.”

*With additional reporting by **Nicholette Zeliadt**.*

**REFERENCES:**

1. Buescher A.V. *et al. JAMA Pediatr.* **168**, 721-728 (2014) **PubMed**