



Simons VIP Connect is collaborating with Roche Pharmaceuticals to pilot an exciting research opportunity for families with *SCN2A* gene changes.

Why is Roche partnering with Simons VIP to collect data on *SCN2A* families?

Scientists at Roche Pharmaceuticals were impressed by the integrity and quality of the family-centered approach to research used in earlier genetic studies led by the Simons Foundation. As a result, the scientists at Roche wanted to partner with the Simons VIP team to work together with families to collect natural history data in a way that will ensure that the data is accessible to other researchers. By partnering with Simons VIP, Roche can collect quality information on individuals with *SCN2A*, which ultimately would be used to aid in the development of treatments.

The purpose of this pilot study is to gather information about behavior and brain functioning in children and families with *SCN2A* gene changes and show that this study model works to collect useful information. We are seeking ten families to participate in this initial study. Depending on the outcome of this pilot study, we hope that this or other opportunities will be available to additional families in the future.

What is expected of my family for participation in this study?

Your specific visit schedule will be tailored to your family, depending on how many family members will be coming and how old each child is.

Two sample itinerary cards are shown, each with a dark blue header and a light yellow body. The cards are titled "Sample Itinerary: Day 1" and "Sample Itinerary: Day 2".

Day	Child	Parents
Day 1	<ul style="list-style-type: none">- Developmental/cognitive assessment (2 hours of tasks)- Play-based social communication assessment (0.5-1 hour)- Short breaks/lunch break, as needed- Language assessment (0.5-1 hour)	<ul style="list-style-type: none">- EEG study (1 hour)
Day 2	<ul style="list-style-type: none">- EEG study (1 hour)	<ul style="list-style-type: none">- Participate in a cognitive assessment (~1.5 hours)- Throughout the two-day visit, complete several interviews regarding your child's adaptive skills, as well as developmental, medical and treatment histories

More Detailed Overview of Study Assessments:

The behavioral assessment portion of the study for children lasts 3-4 hours and will span across one or two days depending on your children's attention span and energy level, breaks needed, and scheduling requirements.

Assessment includes developmental/cognitive assessment, play-based social communication assessment, and language assessments.



The EEG portion of the study involves your child wearing an EEG cap and sitting still while watching videos and listening to sounds. The EEG process lasts approximately an hour.

Parents are also very involved with the research study. Participation involves completing a cognitive assessment (~1.5 hours) and an EEG assessment, as well as several interviews regarding your children's adaptive skills and developmental, medical, and treatment histories. Depending on your children's needs and comfort, it may be helpful for you to also assist the research staff by providing support during your children's assessments.



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Important Information about Qualifying for the Study:

1. **Genetics:** You must submit a qualifying genetic test report to Simons VIP Connect.
 - a. The *SCN2A* variant identified on your lab report must be considered “*pathogenic*” (disease-causing) based on the interpretation by the lab and the study team.
 - b. The child with an identified *SCN2A* mutation does not have any other *pathogenic* findings on his/her lab report.

2. **Family Structure:** We are trying to understand the effect of a *SCN2A* mutation in the context of a family.
 - a. It is important that both biological parents participate, as well as a sibling, if available.

3. **Travel:** In-person evaluations will take place at two sites in the United States.
 - a. Your family will need to be able to travel to either the University of Washington (in Seattle) or Geisinger’s Autism and Developmental Medicine Institute (in central Pennsylvania).
 - b. International families are eligible to participate. However, travel expenses to the United States are not covered.

4. **Timing:** We expect the majority of in-person evaluations to be scheduled between April - August 2016.
 - a. Your family will need to be able to travel during this timeframe.

5. **Other Diagnoses:**
 - a. If your child has any other diagnoses that may cause autism or developmental delay, in addition to *SCN2A*, s/he may not be eligible. For example, if your child has had a traumatic brain injury as well as an *SCN2A* mutation, s/he may not be eligible.



If you are interested in participating and are eligible, we will arrange for your family to travel to an evaluation site. All travel and accommodation expenses will be paid by the study. Your child’s overall health, behavior, and learning ability will be evaluated. You will be given detailed reports with the results of each of these assessments.

If you are interested in participating in this pilot study, please complete the following form.

<https://www.surveymonkey.com/r/39B5WTV>