We are seeking participants affected by undiagnosed, rare disease.

We are a research study called the GREGoR ("Genomics Research to Elucidate the Genetics of Rare Disease") Consortium supported by the National Institutes of Health (NIH) and taking place at Stanford University. Our goal is to improve diagnosis and care for people with undiagnosed, rare diseases.

We evaluate multiple scientific approaches for rare disease diagnoses. Through this study, we hope to enhance the development, prioritization, and application of these approaches to allow for their routine use for patient diagnoses in the future.

What does your participation involve?

- Review of your medical records
- Answer questionnaires about your and your family's health
- Collect blood and possible skin sample for various genetic and molecular tests
- May take photographs

How may you benefit from our study?

- Possible diagnosis to explain your or your family member's disease
- Improve care for others with similar conditions
- Help researchers understand the function of these genes in everyone

If you are interested in learning more, please contact us at gregorsite@stanford.edu or visit our website at gregor.stanford.edu.