



RASopathy Associated Traits California (RasCal) Study

Dr. Lauren Weiss is enrolling families to a new federally-funded research study at UCSF. This exciting study will help us understand some of the medical and behavioral problems associated with different RASopathies. RASopathies are a class of developmental syndromes caused by mutations in genes that encode protein components of the Ras/MAPK signaling pathway, such as neurofibromatosis, Costello syndrome, cardio-facio-cutaneous syndrome, and Noonan syndrome).

WHY is this study being done?

We want to understand why some RASopathy associated features are different among affected people. In order to study this, we will collect genetic information on RASopathy subjects and their family members. We will use this information to determine if there is a change at genetic locations other than the disease genes that may interact with a Ras-MAPK pathway mutation to contribute to the different risk that we see among subjects for developmental problems, cancer risk, and other features.

WHO are we looking for?

We are looking for families with someone affected by neurofibromatosis type 1, Costello syndrome, cardio-facio-cutaneous syndrome, or Noonan syndrome. The affected person, both biological parents and a healthy sibling can participate.

WHAT will be asked of you?

We ask the family to complete several brief questionnaires over the mail or phone. We will also collect saliva samples. This can be done at home and returned via mail.

Are there benefits to participating in research?

Your participation in this research study is very important because it can help identify people at higher risk for some different features of RASopathies and who might benefit from additional medical testing or therapies. This research may also help us understand related traits in the general population, so it may ultimately benefit not only families affected by RASopathies, but also those affected by common disorders like autism and cancer.

If you would like to learn more about this study or to participate, please contact the study coordinator Dina Bseiso at 415-476-6988 or email dina.bseiso@ucsf.edu

THANK YOU FOR YOUR INTEREST IN THIS STUDY!