

Title of Study: A Registry Study to Characterize Genetic and Pathway Biomarkers in Noonan Syndrome and Other RASopathy Patients

We would like to let you know about research being conducted for patients with a diagnosed RASopathy disorder and hypertrophic cardiomyopathy by Dr. Bruce Gelb at the Icahn School of Medicine at Mount Sinai in New York, NY.

The goal of this study is to observe and track disease progression in people with hypertrophic cardiomyopathy (HCM) due to a RASopathy syndrome. Study procedures will include bloodwork and cardiac imaging.

Eligibility:

To be eligible, an individual needs...

- a clinical or molecular diagnosis of a RASopathy syndrome (Noonan, Noonan with Multiple Lentigines/LEOPARD, Costello and cardiofaciocutaneous syndromes)
- be in generally stable health
- have HCM

Time commitment:

- There will be three assessments over a one year period: baseline, 6 months, and 1 year

For additional information, please contact the study coordinator:

Meghan Mac Neal, MS, CGC, MPH

Meghan.macneal@mssm.edu

(212) 241-6012



12/30/2013 – 4/29/2014