

Clinical, Genetic, and Epidemiologic Study of Children and Adults With RASopathies

NCT04888936

Status	RECRUITING
Sponsor	National Cancer Institute (NCI)
Enrollment	500 participants

Key Eligibility Criteria

Inclusion (16)

- Carriers: An individual who meets any of the following criteria will be eligible to participate in this study:
- Individuals with a clinical diagnosis of a RASopathy, including Costello syndrome, Noonan syndrome, Noonan syndrome with multiple lentigines, Cardiofaciocutaneous syndrome, Legius syndrome, capillary arteriovenous malformation syndrome, or others, are eligible. Published clinical diagnostic criteria exist for most of the clinical RASopathy syndromes and differ by syndrome. It will be uncommon for individuals to have a clinical diagnosis and not have had molecular genetic testing. All individuals considered by the study team to be at risk for a RASopathy who have not had prior genetic testing will have this completed as part of the study. The rare individuals with a clinical diagnosis of a RASopathy who are not found to carry a corresponding pathogenic or likely pathogenic variant in a known RASopathy gene will be considered for exome analysis for identification of potentially novel RASopathy germline variation.
- Individuals with a germline variant (P/LP or a variant of uncertain significance but predicted bioinformatically to be damaging) in a RASopathy-associated gene are eligible. These include but are not limited to: BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MAP3K8, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RASA1, RASA2, RIT1, RRAS, SHOC2, SOS1, SPRED1. From herein, we refer to 1) individuals with germline pathogenic variation in a RAS pathway gene AND 2) individuals with a clinical RASopathy diagnosis but in whom a genetic variant has not yet been identified as "carriers." The first member of a family to be identified is termed a "proband."
- Individuals with NF1 only are not eligible for the study. However, individuals with a dual diagnosis of both NF1 and another RASopathy (via genetic testing and/or clinical diagnosis) are eligible for the study.

... and 11 more (see full listing online)

Exclusion (5)

- Carriers: An individual who meets any of the following criteria will be excluded from participation in this study:
- Individuals with only a diagnosis of NF1, or a newly identified germline pathogenic germline variant in NF1, and first-degree relatives of these patients are ineligible. However, individuals with a dual diagnosis of both NF1 and another RASopathy (via genetic testing and/or clinical diagnosis) are eligible for the study.
- Individuals who, in the opinion of the investigator, are not able to return for follow-up visits or obtain required follow-up studies will be excluded from participation in the NIH Clinical Center Cohort.
- Controls: An individual who meets any of the following criteria will be excluded from participation in this study:
- Individuals who, in the opinion of the investigator, are not able to return for follow-up visits or obtain required follow-up studies will be excluded from participation in the NIH Clinical Center Cohort.

Locations (2 total)

National Institutes of Health Clinical Center, Bethesda, Maryland, United States
National Cancer Institute - Shady Grove, Rockville, Maryland, United States

<https://clinicaltrials.gov/study/NCT04888936>

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