RASopathy Biorepository

Status: RECRUITING

Eligibility Criteria

Sex: ALL
Age:
Healthy Volunteers: This study is also accepting healthy volunteers
Inclusion Criteria:

- Patients with a suspected or known diagnosis of any of the group of disorders known as RASopathies (e.g., Neurofibromatosis, Costello Syndrome, Noonan Syndrome). Diagnosis may be made clinically and/or confirmed through genetic testing.
- Unaffected relatives of patients with a suspected or known diagnosis of any of the group of disorders known as RASopathies.

Exclusion Criteria:

- Individuals who do not have a suspected or definite diagnosis of a RASopathy.
- Individuals who do not have a relative with a suspected or definite diagnosis of a RASopathy.
- Patients who do not have the ability/capacity to undergo the informed consent process OR whose parent/legal guardian is unable to undergo the informed consent process.

Conditions & Interventions

Conditions:
RAS Mutation, Neurofibromatosis 1, Noonan Syndrome, Noonan Syndrome With Multiple Lentigines, Noonan Neurofibromatosis Syndrome, Cardiofaciocutaneous Syndrome, Costello Syndrome, Legius Syndrome, Smith-Kingsmore Syndrome, MTOR Gene Mutation, GATOR-1 Gene Mutation, SYNGAP1-Related Intellectual Disability, DLG4, MAPK1 Gene Mutation

More Information

Description:
The RASopathies are a group of developmental disorders caused by genetic changes in the genes that compose the Ras/mitogen activated protein kinase (MAPK) pathway. New RASopathies are being diagnosed frequently. This pathway is essential in the regulation of the cell cycle and the determination of cell function. Thus, appropriate function of this pathway is critical to normal development. Each syndrome in this group of disorders has unique phenotypic features, but there are many overlapping features including facial features, heart defects, cutaneous abnormalities, cognitive delays, and a predisposition to malignancies. This research study proposes to collect and store human bio-specimens from patients with suspected or diagnosed RASopathies. Once obtained, blood and/or tissue samples will be processed for: metabolic function studies, biomarkers, genetic studies, and/or the establishment of immortalized cell lines. In addition, data from the medical record (including neuropsychological evaluations) and surveys will be stored to create a longitudinal database for research conducted at CCHMC or at other research institutions.

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