Advocating for Research on the RASopathies ~ Genetic Disorders of the RAS Signaling Pathway

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https://rasopathiesnet.org

RASopathies Network (RASNet) is a nonprofit advocacy organization run by parents of children with neurodevelopmental syndromes caused by germline mutations in components of the Ras-MAPK signaling pathway. These syndromes are collectively referred to as 'RASopathies' and include: Costello syndrome (CS) caused by mutations in HRAS; Cardio-facio-cutaneous (CFC) syndrome with mutations in BRAF, MAP2K1/2, or KRAS; Neurofibromatosis type 1 (NF1) caused by neurofibromin RAS-GAP mutations; Legius syndrome with SPRED1 mutations; and Noonan and Noonan-like syndromes (NS, NSML, NS-LAH) linked to >18 genes, with PTPN11 (Shp2) mutations being the most prevalent. The vast majority of RASopathy mutations result in activation of signaling. Excess signaling during development causes clinical manifestations including distinct facial features, feeding difficulties, growth delay, heart defects, sparse/coarse hair, cutaneous marks, short stature, and elevated cancer risk. RASopathies affect nearly every physiological system to various extents impacting quality of life and sometimes mortality.

RASNet provides support and information to families diagnosed with a RASopathy, raises awareness of the syndromes, and connects caregivers, researchers, and doctors, with the ultimate goal to accelerate research toward effective treatments and better quality of life. In particular, RASNet 1) convenes biennial symposia with clinicians, researchers, and RASopathy families, to discuss the current state of the field and bring attention to issues raised by patients; 2) engages stakeholders through partnerships, presentations, website and social media networks to build knowledge and community around the RASopathies; 3) encourages participation in research studies and; 4) raises funds for research projects; and 5) lays the groundwork for clinical trial participation. It is the hope of the RASopathies community that advances in the treatment of RAS-driven cancers will come to benefit individuals with RASopathies. 2021

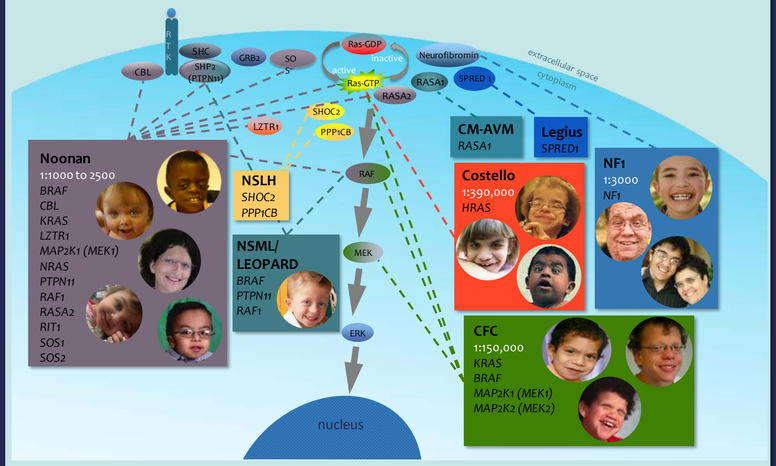
RASOPATHIES NETWORK
connect • collaborate • accelerate

RASopathies Network (RASNet), founded in 2010, is a nonprofit volunteer advocacy organization run by parents of children with genetic disorders of RAS signaling.

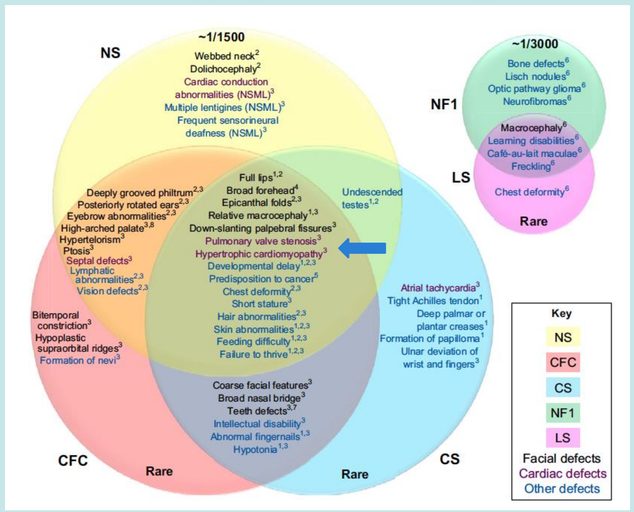
Our mission is to advance research to improve the quality of life for RASopathies families by bringing together families, clinicians and scientists.

RASopathies are disorders caused by mutations in genes that result in excess Ras signaling activity during embryonic development and throughout the lifespan.

RAS Pathway, Syndrome Associations and Prevalence



Overlapping Clinical Phenotypes



Jindal *et al.*, 2015, *Disease Models & Mechanisms*

Partnering Syndrome Family Support Groups

NOONAN SYNDROME FOUNDATION
NEUROFIBROMATOSIS NETWORK NF
Costello Syndrome FAMILY NETWORK
Noonan UK
ICSSG International Costello syndrome Support group
CHILDREN'S TUMOR FOUNDATION ENDING NF THROUGH RESEARCH
Noonan Syndrome Association
CFC International Cardio-Facio-Cutaneous Syndrome

How Does RASNet Work to Advance Research?

Biennial RASopathies Conferences for Families, Doctors, Researchers, and Biopharma

Fundraise to Support Research Projects

Collaborate to Facilitate Natural History Studies & Future Clinical Trials

Am J Med Genet A. 2020 Jan 8. doi: 10.1002/ajmg.a.61485. [Epub ahead of print]

Advancing RAS/RASopathy therapies: An NCI-sponsored intramural and extramural collaboration for the study of RASopathies.

Gross AM[1], Frone M[2], Gripp KW[3], Gelb BD[4,5], Schoyer L[6], Schill L[6], Stronach B[6], Biesecker LG[7], Esposito D[8], Hernandez ER[1], Legius E[9], Loh ML[10], Martin S[1], Morrison DK[11], Rauen KA[12], Wolters PL[1], Zand D[13], McCormick F[7], Savage SA[2], Stewart DR[2], Widemann BC[1], Yohe ME[1].

RASopathies Network Contact Registry Contents

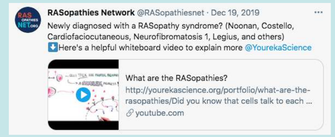
Affected person information
Name
Birth year
Street address
City, State, Zip code, Country
Contact info (e-mail, mobile phone)
Diagnosis
Gene mutation if known
If deceased, date and cause of death

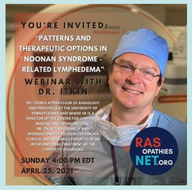
1st Caregiver (parent or other)
Name
Relationship
Contact (e-mail, mobile phone)

2nd Caregiver (parent or other)
Name
Relationship
Contact (e-mail, mobile phone)

Please contact info@rasopathiesnet.org if you are interested in joining!

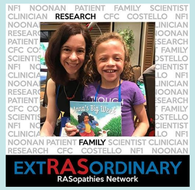
Educate, Engage, & Spotlight Current Topics





New Opportunity!

Participate in the first ever study of brain and cognitive development in boys and girls with Noonan syndrome



2nd Annual
RAS-Targeted Drug Development
Finally Bring an End to RAS Driven Cancers

Raise Awareness & Drive Innovation

Advocate For Our Loved Ones

We gratefully acknowledge the RASopathies community, RASNet Board Members: Lisa Schill, Lee Johnson, Elisabeth Parker, Bruce Deckman, Cara Borian; Scientific Advisory Board and Advocate Advisory Board members; Intramural partners: Brigitte Widemann, Andrea Gross, Marielle Yohe, Pediatric Oncology Branch, CCR, NCI; and Mentor: William Timmer, NCI.