Resource Summary Report

Generated by RRID on May 5, 2025

NBIA Disorders Association

RRID:SCR_005382
Type: Tool

Proper Citation

NBIA Disorders Association (RRID:SCR_005382)

Resource Information

URL: http://www.nbiadisorders.org/

Proper Citation: NBIA Disorders Association (RRID:SCR_005382)

Description: The NBIA Disorders Association, formerly known as Hallervorden-Spatz Syndrome Association, (HSSA) was originally founded in 1996 by President, Patricia Wood. The goals of the association are to raise funds to support research pertinent to NBIA; to provide emotional support to those afflicted with NBIA and their families; and to raise public awareness of NBIA. The NBIA Disorders Association is accepting applications for one-year grants for clinical and translational research studies related to the early detection, diagnosis, or treatment of patients with NBIA. Neurodegeneration with Brain Iron Accumulation (NBIA) is a group of rare, genetic, neurological disorders characterized by the accumulation of iron deposits in the brain and progressive degeneration of the nervous system. It typically first appears in childhood. Presenting signs and symptoms may include difficulty walking, loss of balance, and problems related to speech. Those affected suffer a progressive loss of muscle control, sudden involuntary muscle spasms, and uncontrolled tightening of the muscles. Symptoms may also include disorientation, seizures, and deterioration of intellectual ability. Approximately half of the cases diagnosed have been linked to a mutation of a gene known as PANK2. At the present time, symptoms may be treated but there is no cure. The purpose of the NBIA Disorders Association Research Grant Program is to encourage meritorious research studies designed to improve the diagnosis or treatment of NBIA. The research can be conducted in the United States, countries of the European Union, Canada, Australia, New Zealand, Brazil, Argentina, Chile, South Africa, Japan, or Israel, and in other countries where adequate supervision of grant administration is possible. Grants will be awarded to qualified researchers to initiate pilot studies, the results of which are intended to be used to obtain larger multi-year grant funding. Evaluation of proposals will follow NIH guidelines and include careful consideration of experimental or protocol design, objectivity or relevance of parameters measured, and statistical analysis plan. Proposals that address the following areas will be given priority: * Therapeutics Development: ** Development of pantethine and

its derivatives ** Development of other rational therapeutics * Animal & Cellular Models: ** Development of a new rodent disease model by targeted insertion of a "human disease" mutation into Pank2 ** Development of induced pluripotent stem cell lines. *** Development of animal and cellular models will be considered for multi-year funding with adequate budget justification. Proposals should detail a research plan and a budget for the initial phase of the work, with the option to contract further work out to a commercial enterprise. * Biomarker Discovery and Assay Development: ** Metabolomics ** Coenzyme A / acyl coenzyme A measurement using accessible (peripheral and central) tissue/fluid * New NBIA gene discovery

Abbreviations: NBIA Disorders Association

Synonyms: NBIA Disorders Association: from discovery to cure, Hallervorden-Spatz Syndrome Association, HSSA

Resource Type: institution

Keywords: hallervorden-spatz disease, neurodegeneration with brain iron accumulation, rare disease, pantothenate kinase-associated neurodegeneration, genetic, neurological disorder, brain, neurodegeneration, pank2

Funding:

Resource Name: NBIA Disorders Association

Resource ID: SCR_005382

Alternate IDs: Crossref funder ID: 100009582, grid.469792.7, nlx_144453

Alternate URLs: https://ror.org/008421332

Record Creation Time: 20220129T080229+0000

Record Last Update: 20250420T014250+0000

Ratings and Alerts

No rating or validation information has been found for NBIA Disorders Association.

No alerts have been found for NBIA Disorders Association.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 4 mentions in open access literature.

Listed below are recent publications. The full list is available at RRID.

Jeong SY, et al. (2019) 4'-Phosphopantetheine corrects CoA, iron, and dopamine metabolic defects in mammalian models of PKAN. EMBO molecular medicine, 11(12), e10489.

Orellana DI, et al. (2016) Coenzyme A corrects pathological defects in human neurons of PANK2-associated neurodegeneration. EMBO molecular medicine, 8(10), 1197.

Shumar SA, et al. (2015) Induction of Neuron-Specific Degradation of Coenzyme A Models Pantothenate Kinase-Associated Neurodegeneration by Reducing Motor Coordination in Mice. PloS one, 10(6), e0130013.

Schiessl-Weyer J, et al. (2015) Acanthocytosis and the c.680 A>G Mutation in the PANK2 Gene: A Study Enrolling a Cohort of PKAN Patients from the Dominican Republic. PloS one, 10(4), e0125861.