What is NBIA?

Neurodegeneration with Brain Iron Accumulation (NBIA), is a group of rare, inherited, neurological disorders, characterized by the progressive degeneration of the nervous system.

The common feature among all individuals with NBIA is iron accumulation in the brain along with a progressive movement disorder: This is especially seen in regions of the basal ganglia called the globus pallidus and the substantia nigra. The basal ganglia is a collection of structures deep within the base of the brain that assist in regulating movements. The exact relationship between iron accumulation and the symptoms of NBIA is not fully understood.

Individuals can plateau for long periods of time and then undergo intervals of rapid deterioration. Symptoms may vary greatly from case to case, partly because there are different genes causing NBIA and also different mutations within a gene could lead to a more or less severe presentation. The factors that influence disease severity and rate of progression are still unknown.

The movement disorders result in clumsiness, difficulty controlling the body and speech problems. Dystonia (involuntary muscle contractions that may force certain body parts into unusual and sometimes painful movements and positions), spasticity and visual disorders are common clinical signs.

Many individuals eventually lose the ability to walk, talk, or chew food and become totally dependent on others for all their needs.

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NBIA ALLIANCE

Worldwide Partners for a Cure

The Alliance is an international umbrella organization for NBIA lay advocacy groups. It started as an informal group in January 2012, in conjunction with the kick-off meeting of the EU funded FP7 project TIRCON.

The countries below are represented with member groups:

Canada
France
Germany
Italy
Netherlands
Spain
Switzerland
United States

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NBIA Disorders

Prior to 2001, all forms of NBIA were called Hallervorden-Spatz Syndrome or disease. There were no genes yet identified to help us define what was causing the clinical symptoms.

In 2001, the first genetic cause was identified, and it was found that approximately 35-50% of individuals with a clinical diagnosis of NBIA have gene mutations in *PANK2*. Those with this gene defect have Pantothenate Kinase-Associated Neurodegeneration (PKAN).

In 2006 a second NBIA gene was identified, *PLA2G6*, and affected individuals are said to have PLAN, or PLA2G6-Associated Neurodegeneration. In 2011 the responsible gene for Mitochondrial-membrane Protein-Associated Neurodege-neration (MPAN), *C19orf12*, was detected, and in 2012 mutations in the gene *WDR45* were found to cause Beta-propellar Protein-Associated Neuro-degeneration (BPAN).

The following very rare disorders have been added to the NBIA group as well: Aceruloplasminemia, COASY Protein-Associated Neurodegeneration (CoPAN), Fatty Acid Hydroxylase-Associated Neurodegeneration (FAHN), Kufor-Rakeb, Neuro-ferritinopathy, and Woodhouse-Sakati Syndrome.

As more genes are discovered, we expect the list of disorders to grow. Those individuals where the gene has not yet been found but who exhibit the clinical symptoms of NBIA are said to have Idiopathic NBIA, meaning it is of unknown origin.

NBIA Alliance

Development and Mission

TIRCON

FP/-277984-2

www.tircon.eu

In January 2012 the EU-funded NBIA project TIRCON (Treat Iron-Related Childhood-Onset Neurodegeneration) started with a kick-off meeting in Munich, Germany. Existing and developing NBIA lay advocacies from Europe and the United States launched the international NBIA Alliance in combination with and supported by this TIRCON meeting.

The Alliance members' agreements comprise the following areas of cooperation:

* While each country has their own legal entity, we share common goals in helping NBIA families coping with these rare disorders, promoting improved health care with access for all those affected, and supporting NBIA research to find more effective treatments and ultimately cures for these disorders.

* We are working together on a strategic plan for the future that we all endorse and will implement together.

* We display the Alliance logo on our websites and in literature to show we are members of a worldwide collaboration.

* We encourage the growth of NBIA organizations in other countries and strive to help newly developing groups by sharing our knowledge, and being a resource to help them flourish and join our Alliance.

* We are represented by the lay leaders of each country's NBIA organizations and a Scientific Advisory Board of leading NBIA researchers (with representatives from the TIRCON consortium) and clinicians that share our vision of collaboration and cooperation.

* We are willing to network and exchange experiences with other organizations that share our goals in the field of rare diseases.

Members of the NBIA Alliance, October 2015
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