HOUSE RESOLUTION

WHEREAS, First described in 1826, more than 170 years ago, Batten Disease (Neuronal Ceroid Lipofuscinoses), thought to be one of the most common neurodegenerative diseases, remains an unsolved mystery today, a puzzling disease that assures its victims of only one consistent manifestation, early death; and

WHEREAS, An inherited, degenerative, neurological disease, Batten Disease may affect persons of any age, but primarily affects infants, toddlers, and school age children, beginning unexpectedly and leading to a progressive loss of brain function that later destroys bodily functions, eventually leaving the victim totally helpless; and

WHEREAS, Whether in the case of infantile (Santavnori), late infantile (Jansky, Bielschowsky), juvenile (Batten, Spielmeyer, Sjogren), or adult type (Kuf, Parry), the early symptoms of Batten Disease are confusing ones; it strikes without warning, affecting vision, and causing seizures or convulsions; and

WHEREAS, Possibly most frustrating of all is the fact that Batten Disease is rarely diagnosed immediately, often being mistaken for epilepsy or mental retardation, even schizophrenia; and once diagnosed, there is no satisfactory

treatment and no cure; the clinical course of the disease includes a marked decline in cognitive function; personality and behavior changes; loss of communication and motor skills; poor circulation; decrease in muscle mass; hyperventilation; hallucinations, and, finally, deterioration to a vegetative state that ends in death; and

WHEREAS, Batten Disease is named after the British pediatrician who first described it in 1903; also known as Spielmeyer-Vogt-Sjogren-Batten Disease, it is the most common form of a group of disorders called Neuronal Ceroid Lipofuscinoses (or NCLs); and

WHEREAS, Although Batten Disease is usually regarded as the juvenile form of NCL, it has now become the term to encompass all forms of NCL; and

WHEREAS, The forms of NCL are classified by age of onset and have the same basic cause, progression and outcome but are all genetically different; over time, affected children suffer mental impairment, worsening seizures, and progressive loss of sight and motor skills; eventually, children with Batten Disease/NCL become blind, bedridden, and unable to communicate and it is presently always fatal; Batten Disease is not contagious or, at this time, preventable; and

WHEREAS, The first probable instances of this condition were reported in 1826 in a Norwegian medical journal by Dr. Christian Stengel, who described 4 affected siblings in a small mining community in Norway; although no pathological studies were performed on these children, the clinical descriptions are so succinct that the diagnosis of the Spielmeyer-Sjogren (juvenile) type is fully justified; and

WHEREAS, More fundamental observations were reported by F. E. Batten in 1903, and by Vogt in 1905, who performed extensive clinicopathological studies on several families; retrospectively, these papers disclose that the authors grouped together different types of the syndrome; and

WHEREAS, Furthermore Batten, at least for some time, insisted that the condition that he described was distinctly different from Tay-Sachs Disease, the prototype of a neuronal lysosomal disorder now identified as GM2-Gangliosidosis type A; around the same time, Spielmeyer reported detailed studies on three siblings, suffering from the Spielmeyer-Sjogren (juvenile) type, which led him to the very firm statement that this malady is not related to Tay-Sachs Disease; subsequently, however, the pathomorphological studies of Schaffer made these authors change their minds to the extent that they reclassified their respective observations as variants of Tay-Sachs Disease, which caused confusion lasting about 50 years; and

WHEREAS, In 1913-14, M. Bielschowsky delineated the Late Infantile form of NCL; however, all forms were still thought to belong in the group of "familial amaurotic idiocies", of which, Tay-Sachs was the prototype; in 1931, the Swedish psychiatrist and geneticist, Torben Sjogren, presented 115 cases with extensive clinical and genetic documentation and came to the conclusion that the disease which we now call the Spielmeyer-Sjogren (juvenile) type is genetically separate from Tay Sachs; and

WHEREAS, Departing from the careful morophological observations of Spielmeyer, Hurst, and Sjovall and Ericsson, Zeman and Alpert made a determined effort to document the previously suggested pigmentary nature of the neuronal deposits in certain types of storage disorders; simultaneously, Terry and Korey and Svennerholm demonstrated a specific ultrastructure and biochemistry for Tay Sachs Disease, and these developments led to the distinct identification and also separation of the NCLs from Tay Sachs Disease by Zeman and Donahue; at that time, it was proposed that the Late Infantile (Jansky-Bielschowsky), the Juvenile (Spielmeyer-Vogt), and the adult form (Kufs) were quite different from Tay-Sachs Disease with respect to chemical pathology and ultrastructure and also different from other forms of sphingolipidoses; and

WHEREAS, Subsequently, it was shown by Santavuori and Haltia that an infantile form of NCL exists, which Zeman and Dyken had included with the Jansky Bielschowsky type; and

WHEREAS, There are four main types of NCL, including two forms that begin earlier in childhood and a very rare form that strikes adults; the symptoms are similar but they become apparent at different ages and progress at different rates:

Infantile NCL (Santavuori-Haltia disease): begins between about 6 months and 2 years of age and progresses rapidly; affected children fail to thrive and have abnormally small heads (microcephaly); also typical are short, sharp muscle contractions called myoclonic jerks; initial signs of this disorder include delayed psychomotor development with progressive deterioration, other motor disorders, or seizures; the infantile form has the most rapid progression and children live into their mid childhood years;

Late Infantile NCL (Jansky-Bielschowsky disease): begins between ages 2 and 4; the typical early signs are loss of muscle coordination (ataxia) and seizures along with progressive mental deterioration; this form progresses rapidly and ends in death between ages 8 and 12;

Juvenile NCL (Batten Disease): begins between the ages of 5 and 8 years of age; the typical early signs are progressive vision loss, seizures, ataxia, or clumsiness; this form

progresses less rapidly and ends in death in the late teens or early 20s, although some may live into their 30s;

Adult NCL (Kufs Disease or Parry's Disease): generally begins before the age of 40, causes milder symptoms that progress slowly, and does not cause blindness; although age of death is variable among affected individuals, this form does shorten life expectancy; and

WHEREAS, Batten Disease/NCL is relatively rare, occurring in an estimated 2 to 4 of every 100,000 births in the United States; the diseases have been identified worldwide; although NCLs are classified as rare diseases, they often strike more than one person in families that carry the defective gene; and

WHEREAS, Childhood NCLs are autosomal recessive disorders; that is, they occur only when a child inherits two copies of the defective gene, one from each parent; when both parents carry one defective gene, each of their children faces one in four chance of developing NCL; at the same time, each child also faces a one in two chance of inheriting just one copy of the defective gene; individuals who have only one defective gene are known as carriers, meaning they do not develop the disease, but they can pass the gene on to their own children; and

WHEREAS, Adult NCL may be inherited as an autosomal

recessive (Kufs) or, less often, as an autosomal dominant (Parrys) disorder; in autosomal dominant inheritance, all people who inherit a single copy of the disease gene develop the disease; as a result, there are no unaffected carriers of the gene; symptoms of Batten Disease/NCLs are linked to a buildup of substances called lipopigments in the body's tissues; these lipopigments are made up of fats and proteins; their name comes from the technical word lipo, which is short for "lipid" or fat, and from the term pigment, used because they take on a greenish-yellow color when viewed under an ultraviolet light microscope; and

WHEREAS, The lipopigments build up in cells of the brain and the eye as well as in skin, muscle, and many other tissues; inside the cells, these pigments form deposits with distinctive shapes that can be seen under an electron microscope; some look like half-moons (or comas) and are called curvilinear bodies, others look like fingerprints and are called fingerprint inclusion bodies, and still others resemble gravel (or sand) and are called granual osmophilic deposits (grods); these deposits are what doctors look for when they examine a skin sample to diagnose Batten Disease; the diseases cause death of neurons (specific cells found in the brain, retina and central nervous system); the reason for neuron death is still not known; and

WHEREAS, Because vision loss is often an early sign, Batten Disease/NCL may be first suspected during an eye exam; an eye doctor can detect a loss of cells within the eye that occurs in the three childhood forms of Batten Disease/NCL; however, because such cell loss occurs in other eye diseases, the disorder cannot be diagnosed by this sign alone; and

WHEREAS, Often an eye specialist or other physician who suspects Batten Disease/NCL may refer the child to a neurologist, a doctor who specializes in disease of the brain and nervous system; in order to diagnose Batten Disease/NCL, the neurologist needs the patient's medical history and information from various laboratory tests; diagnostic tests used for Batten Disease/NCLs include:

Skin or tissue sampling; the doctor can examine a small piece of tissue under an electron microscope; the powerful magnification of the microscope helps the doctor spot typical NCL deposits; these deposits are found in many different tissues, including skin, muscle, conjunctiva, rectal, and others; blood can also be used;

electroencephalogram or EEG; an EEG uses special patches placed on the scalp to record electrical currents inside the brain; this helps doctors see telltale patterns in the brain's electrical activity that suggest a patient has seizures;

Electrical studies of the eyes; these tests, which include

visual-evoked responses (VER) and electro-retinagrams (ERG), can detect various eye problems common in childhood Batten Disease/NCLs;

Brain scans; imaging can help doctors look for changes in the brain's appearance; the most commonly used imaging technique is computed tomography (CT), which uses x-rays and a computer to create a sophisticated picture of the brain's tissues and structures; a CT scan may reveal brain areas that are decaying in NCL patients; a second imaging technique that is increasingly common is magnetic resonance imaging, or MRI; MRI uses a combination of magnetic fields and radio waves, instead of radiation, to create a picture of the brain;

Enzyme assay; a recent development in diagnosis of Batten Disease/NCL is the use of enzyme assays that look for specific missing lysosomal enzymes for Infantile and Late Infantile only; this is a quick and easy diagnostic test;

Genetic/DNA testing; each form of Batten disease is the result of a different gene; genes for eight or the ten forms have been identified; testing for these is available for diagnosis as well as carrier and prenatal; and

WHEREAS, As yet, no specific treatment is known that can halt or reverse the symptoms of Batten Disease/NCL; however, seizures can be reduced or controlled with anticonvulsant drugs, and other medical problems can be treated appropriately

as they arise; at the same time, physical and occupational therapy may help patients retain function as long as possible; and

WHEREAS, Some reports have described a slowing of the disease in children with Batten Disease who were treated with vitamins C and E and with diets low in vitamin A; however, these treatments did not prevent the fatal outcome of the disease; and

WHEREAS, Support and encouragement can help children and families cope with the profound disability and losses caused by NCLs; the Batten Disease Support and Research Association enables affected children, adults, and families to share common concerns and experiences; meanwhile, scientists pursue medical research that will someday yield an effective treatment; and

WHEREAS, Within the federal government, the focal point for research on Batten Disease and other neurogenetic disorders is the National Institute of Neurological Disorders and Stroke (NINDS); the NINDS, a part of the National Institutes of Health (NIH), is responsible for supporting and conducting research on the brain and central nervous system; the Batten Disease Support and Research Association and the Children's Brain Diseases Foundation also provide financial assistance for research; and

WHEREAS, Through the work of several scientific teams, the search for the genetic cause of NCLs is gathering speed; in September 1995, The International Batten Disease Consortium announced the identification of the gene for the juvenile form of Batten Disease; the specific gene, CLN3, located on Chromosome 16, has a deletion or piece missing; this gene accounts for 73% of all cases of Juvenile Batten Disease; the rest are the result of other defects of the same gene; and

WHEREAS, Also, in 1995, scientists in Finland announced the identification of the gene responsible for the infantile form of Batten Disease; the gene, CLN1, is located on Chromosome 1; in September 1997, scientists at the Robert Woos Johnson Medical School and the Institute for Basic Research, New York, announced the identification of the gene for the "classic" Late Infantile form of Batten Disease/NCL; the gene, CLN2, is located on chromosome 11; and

WHEREAS, Scientists have also identified the genes responsible for Finnish Late Infantile (CLN5), variant Late Infantile (CLN6), EPMR (CLN8), and Congenital/CTSD (CLN10); research also continues toward identification of the gene for the adult form of Batten Disease/NCL, also known as Kufs Disease; and

WHEREAS, Identification of the specific genes for Infantile, Late Infantile, Variant Late Infantile, and Juvenile Batten Disease/NCL has led to the development of DNA diagnostics, carrier, and prenatal tests; and

WHEREAS, Scientists have discovered that the Infantile and Late Infantile diseases are missing key lysosomal enzymes, i.e. Palmitoyl Protein Thioesterase 1 (PPT1) for Infantile and Tripeptidyl Peptidase 1 (TPP1) for Late Infantile; knowing that these enzymes are missing is now leading to the development of gene replacement and stem cell transplantation therapies; and

WHEREAS, Recent studies have shown a link between the Juvenile form and the body's autoimmune system; although this link is not yet fully understood, it may eventually lead to a treatment; therefore, be it

RESOLVED, BY THE HOUSE OF REPRESENTATIVES OF THE NINETY-SIXTH GENERAL ASSEMBLY OF THE STATE OF ILLINOIS, that we declare June 6-7, 2009 Batten Disease Awareness Weekend in the State of Illinois and ask people of the State to look at ways in which they may help to combat this terrible disease; and be it further

RESOLVED, That a suitable copy of this resolution be presented to the Batten Disease Research and Support

Association as a symbol of our support.