

NATIONAL NIEMANN-PICK DISEASE FOUNDATION

FAMILY SUPPORT SUPPLEMENT

Volume 3, Issue 1, Summer 2007

It has been a number of years since NNPfD published a Family Support Supplement, so I would like to take this opportunity to welcome you and explain how this publication will work.

The Family Support Supplement will provide articles that address day-to-day management of many aspects of Niemann-Pick disease. For example, in this issue we are publishing an updated version of “Communicating with Health Care Professionals” – a document that should be useful to families as they visit physicians and other health care providers.

There’s also a glossary of clinical terms specific to NPD – consider posting it on your refrigerator for quick reference – and a Book Corner about publications of interest.

We’ll continue to publish news about awareness activities, family fundraisers and memorials, as well as information of interest to all of our members and supporters, in the NNPfD Newsletter and, of course, on our web site at **www.nnpdf.org**.

We hope that this package of printed and web-based material addresses your needs and the needs of the entire NNPfD Family.

Please let us know what you think about the NNPfD Family Support Supplement and all of our publications.

Regards,

Barb Vorpahl

Chair, NNPfD



N *National* **Niemann-Pick** **D** *Disease*
Foundation



**15th Annual NNPDF Family Support
and Medical Conference
August 16th–19th, 2007
Hilton ~ Milwaukee City Center**



www.visitmilwaukee.org

The 15th annual National Niemann-Pick Disease Foundation Family Conference is fast approaching and we have many educational and entertaining events planned for you. Things get underway on Thursday evening, August 16 at the Hilton-Milwaukee City Center with an opening appetizer reception and our “Light of Hope” commemoration.

There will be plenty of time to meet new friends and rekindle treasured connections. Friday and Saturday we hit the main course with an overview of your Foundation’s activities, research and clinical updates, our keynote speaker, and breakout sessions.

Keynote speaker and special guest Dr. Debra McLisch, a rehabilitation medicine specialist, will talk about treating the “whole child” with a chronic illness, using a multidisciplinary approach to care. We also have Dr. Elsa Shapiro from the University of Minnesota discussing memory loss and dementia in patients who have lysosomal storage disorders, an educator who will discuss the IEP process, a metabolic dietitian, a sleep disorders specialist and much more. And there will certainly be time for fun and surprises. Information about how to register for the conference and make your hotel and travel arrangements can be found at the NNPDF web site under Family Services.

(http://www.nnpdf.org/familyservices_03.html).

*Your Conference Hosts:
Barb and Gary Vorpahl
Hilton ~ Milwaukee City Center*



“Yesterday, Today, Tomorrow”

Look to this day

*For yesterday is but a dream,
And tomorrow merely a vision.*

*But today, well lived,
Makes every yesterday a dream of happiness*

And every tomorrow a vision of hope.

Look well, therefore, to this day.

“Family Conference Mentor Match”

Are you a first-time attendee to the NNPDF Family Conference? Would you like to have a familiar voice greet you as you arrive? If so, join the NNPDF Family Conference Mentor Match. We are a group of current members who have been active in NNPDF and we're anxious to welcome you to the Foundation



We would like to touch base with you prior to the meeting to answer your questions about the conference or about traveling to the conference. We'll meet with you during the new family get-together on Thursday, August 16 and would be happy to introduce you to other families and to the professionals who will be speaking at the conference.

Please let us know if you are interested by contacting the Foundation office. You'll be asked for the names of your family members and the ages of the children who will be in attendance, as well as a contact telephone number.

We look forward to seeing you soon.

First Time Family Fund Raiser Winners

It is our pleasure to announce the 2006-2007 winners of our

First Time Family Fund Raiser Contest:

The Darrile and Mark Papier Family.

The Papier's hosted a number of different and creative fund raising events throughout the year in honor of their sweet little **Dillon (age 4 ~ NPC)** and rose to the top via strong support from their families, neighbors, and community members.



As winners of the contest, the Papier Family will enjoy a complimentary suite upgrade at the family conference in Milwaukee!!

Congratulations to the Papiers, their family, their community supporters, AND all of our NNPDF members.

October 2007

Families Around the Nation Gear Up to Celebrate National Niemann-Pick Disease Awareness Month

NPD Families nationwide continue to be creative in their plans to observe National Niemann-Pick Disease Awareness month. NPD Awareness Month, observed every October, is a time to draw attention nationally to the challenges facing our NPD members and their families, advocate for more research funding, strengthen family caregiving and education, and increase community awareness and support for NPD families.

The basis and heart of the National Niemann-Pick Disease Foundation (NNPDF) is the strength of our families working together toward the common goals of family support and raising funds for vital research into all forms of Niemann-Pick Disease. As we have learned over the past fifteen years, the fundamental factors in the success of our organization are grassroots fundraising, educational and community awareness programs, and support of research to find a cure.

**This October, NNPDF will promote the “theme”, PERSEVERE,
for NPD Awareness Month 2007.**

NNPDF has developed a variety of tools, available to our families via the foundation office and our web site, to support NPD Awareness Month activities. Many NPD families have found that the “Persevere” wristbands, t-shirts, and magnets, along with the “NPD Fundraising Packet”, in addition to the new, “NPD Family Media Guide” are great resources.

These resources offer suggestions to guide families as they develop project ideas within their community. The Media Guide offers many “how to” suggestions with television and print media, political proclamation ideas and contacts along with an “NPD Fact Sheet”.

We encourage each family to “**PERSEVERE**” and look within their family structure, friends, and community support networks to create their own “team” and sponsor an awareness campaign and/or fundraising event during **October 2007**.

Please contact the foundation office or visit our web site for more information.

www.nnpdf.org

Family Services Program

The goal of NNPDF's Family Services Program is to provide services that anticipate and respond to the needs of those dealing with Niemann-Pick disease and to reduce the impact of the disease on immediate and extended families.

For 15 years, NNPDF has assisted families as they face the challenges of living with Niemann-Pick Disease. Our Family Services Program includes assistance with:

Facilitation of Family Connections

NNPDF offers help in easing the emotional burden imposed by Niemann-Pick Disease by fostering and facilitating networking and other connections among individuals and families. These connections can reduce feelings of isolation and despair as families experience the different phases of NPD.

Education

NNPDF provides medical and educational information to schools, doctors, and other health-care professionals. These materials assist with the diagnosis and treatment of NPD, and they provide referral information to those affected by the disease.

NNPDF also promotes and assists with continuing education and community awareness campaigns through informational brochures, educational videos, a public relations media guide, and many other materials.

Support, Counseling, Referral Services, and Advocacy

NNPDF provides genetic and supportive counseling and referral services through its Coordinator for Education, Referral, and Advocacy. We offer information about working with doctors, clinics, insurance companies, and other health and human service programs. NNPDF programs have been developed to assist families during a crisis and to offer practical suggestions about day-to-day care of those affected by all types of NPD.

Support of Fundraising and Awareness

NNPDF receives financial support for its Family Services and Research Programs from individuals, families, and private organizations. We are grateful for this support. We encourage families to develop and conduct fund raising initiatives on behalf of NNPDF and we have developed a specialized Fund Raising Manual, which is available to families on-line at www.nnpdf.org or from the NNPDF office.

NNPDF Contacts & Support

JEFFERY A. FORAN, PH.D.,

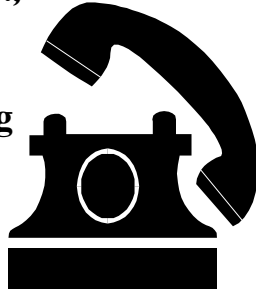
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Web Site: www.nnpdf.org

Donations may be made to:

NNPDF

Post Office Box 310

Fort Atkinson, WI 53538-0310

New NNPDF Logo

The heart and magnifying glass logo on NNPDF's web site represents our commitment to support families affected by NPD, and research to find treatments and a cure.

The rainbow colors are from our original rainbow logo, which was designed by Tommy Kenny. Tommy passed away in 1995 from Niemann-Pick C. Although he knew he was seriously ill, he took strength from the rainbow as a symbol of hope and faith.

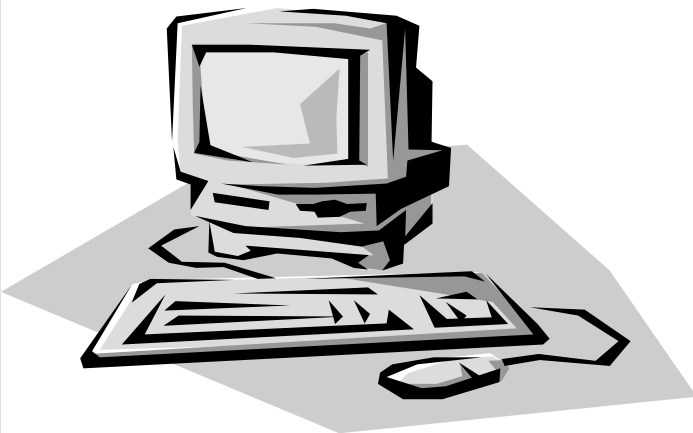
We continue to share his hope and faith with the rainbow colors, our support of NP families (the heart), and our support for research (the magnifying glass).

**N^{ational} Niemann-Pick Disease
Foundation**



Web Site Updates

The new face of NNPDF has arrived with the introduction of our web site in May 2007! We hope that you have had an opportunity to view the home page and the new drop-down menus, to explore many new items and find some old familiar ones as well. Log on to www.nnpdf.org and check out the site.



Let us know what you think and what we can add to make the NNPDF web site more user friendly and useful.

Foundation Office Contact Information

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Are you "On The Move"?

You are important to us.

Please keep in touch



We don't like losing track of our members who have moved. Keep us informed of new addresses, telephone numbers, and e-mail addresses so that we can stay in touch and keep the important flow of information on track to your doorstep.



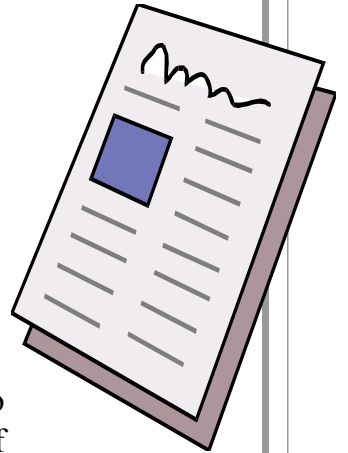
NNPDF DUES

NNPDF is a foundation that provides services to all individuals affected by Niemann-Pick Disease. We **DO NOT** charge for our services (NNPDF no longer requires payment of dues to become a member); rather, our funds come from family fund raising events, private donors, and other foundations.

We invite all interested individuals and organizations to join NNPDF in support of our efforts to raise awareness of NPD, to support individuals and families affected by the disease, and to support research toward finding treatments and a cure for Niemann-Pick Disease.

You can learn more about NNPDF's programs, and you can become an NNPDF member by visiting our web site at www.nnpdf.org, or by contacting the NNPDF office at nnpdf@nnpdf.org.

This is YOUR Family Support Supplement!



We would like to extend a note of thanks to all who contributed to this edition of the NNPDF "Family Support" Newsletter.

We really do need your input and would like to ask for your help and support in continuing this much-needed communication.

Please submit topics and information that you would like to see addressed in future issues. We will work to accommodate your requests.

Look for past copies of our Niemann-Pick News publications as well as copies of these supplements on our web site.

Thanks.



Communicating With Health Care Providers

The relationship between health care providers and patients has changed significantly over the years, in part because of the pressures of the health care system. It is more important now than ever that you learn to take every advantage of the time the provider has to consider your concerns and questions. Be informed, gather information and come to appointments prepared, but be realistic about how much can be managed in a single appointment. Try not to overwhelm the provider with information from outside sources, focus on one or a few issues at each visit. Your description of your own or your child's condition, particularly in the face of a diagnosis of a complex disorder like Niemann-Pick Disease (NPD), will aid the doctor and others in planning treatment in an atmosphere of respect and cooperation. You will need to be frank and clear about the information you bring to the appointment and do your best to communicate complex symptoms that may be difficult to explain.

Identify a Primary Care Provider

It is important that you have one health care provider who knows you or your child well. This professional, your primary care provider, may be a local pediatrician, family practitioner, internist, nurse practitioner or other staff member at a clinic. The primary care provider has several functions. First, he or she is the person you turn to first, especially for routine health issues. He or she is also the person who helps you coordinate the services necessary to comprehensively deal with the disease. This may include recommending referrals to specialist care providers who will supervise a portion of your or your child's care.

How to Choose a Primary Care Provider

Begin your search for a primary care provider by thinking about what you most need from a health care professional. You may, for example, want to find someone who is easy to talk to or who has a particular interest in the condition. At your first visits to doctors or other professionals, observe their reactions to you/your child.

- ~ Do they make eye contact with you/your child?
- ~ Do they treat your child who has NPD with the same concern as they treat your other children?
- ~ Do they really listen and hear what you have to say?
- ~ Do they accept your feelings?

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Communicating With Health Care Providers (cont.)

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- ~ Are they interested in your questions?
- ~ Do they respect your experience and skill as your child's primary caretaker?
- ~ Do they ask appropriate questions?

Your Relationship with Your, or Your Child's Doctor

When you have chosen a primary care provider, make sure that the doctor or professional knows that you want them to take on this role. Establish an understanding on several key issues in the relationship. For example, it helps to agree on what constitutes an emergency and what you should do when one occurs. Think of your primary care provider as someone you can talk to:

- ~ about your child's growth and development and monitoring of overall health in addition to other medical issues
- ~ about your child's progress as well as his or her problems
- ~ about your fears and anxieties and about any changes in your family that might affect your or your child's health status
- ~ about appointments just to talk about serious concerns you have about your child; ask about fees for such consultations

Make the Most of Your Time with the Primary Care Provider

Prepare for routine visits by thinking about the things you want to ask and/or tell the physicians. Write these down! Be sure your own or your child's medical history is up to date so that you can answer any questions the provider may have. Keep a health history form, including significant past medical history and surgeries, allergies, current medications, supplements, herbs, vitamins and over the counter medications as well as those that have been recently discontinued.

The following suggestions may help you be more prepared to ask questions:

- ~ Identify your concerns. Are you worried about specific things?
- ~ Be specific; provide details in an ordered fashion

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Communicating With Health Care Providers (cont.)

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- ~ Be focused; turn off your cell phone, leave other children at home with a spouse, other family member, or sitter if possible
- ~Keep a running list of questions that occur to you between appointments
- ~Make a list of any questions your child might have. This is a good way to reinforce your child's role in his/her health care
- ~After you and your child have written down your concerns, consider them in order of importance. You usually have a limited amount of time for questions.
- ~Make sure you understand the doctor's answers. Feel free to write down what the doctor says and ask for repeat of answers if necessary.
- ~ Ask about what is the best way to communicate If you have to leave with some of your questions unanswered, ask if it is permissible for you to call or e-mail and ask the remaining questions.
- ~Ask for a written summary of the discussion if one is not routinely sent.
- ~Ask for written materials about any devices or medications if that would be helpful.
- ~Ask for information about trusted web sites if applicable, or for sources of additional information.

Concerns

Parents often have concerns in the areas listed below. You may have concerns that do not appear on this list, and all of these concerns may not be of equal importance to you.

Diagnosis

- ~ Is a firm diagnosis possible at this time? If not, when or why not?
- ~ Is there anything to be gained from a second opinion?
- ~ What are the immediate concerns? The longer-term concerns?
- ~ Will this condition get better, get worse, or stay the same?

Medications

- ~ What is the drug meant to do?
- ~ Are there possible side effects?

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Communicating With Health Care Providers (cont.)

(Continued from page 11)

- ~ How much should be taken? At what times? With or between meals?
- ~ Should the medication be taken until symptoms disappear, or until the medication is gone, or ongoing?
- ~ What are the effects of combining this drug with other medications now being taken, including over-the-counter medications like cough syrup, Tylenol or ibuprofen?
- ~ Is there anything, such as food, activities, etc., that should be avoided while using this medication?

Tests

- ~ What do we expect to learn from the test?
- ~ How is the test done?
- ~ Do I need to do anything to prepare for the test?
- ~ What stresses or side effects can the test cause?
- ~ How long will it be before we get the results of the test, and how will they be relayed to me?

Referral to a Specialist

- ~ Why is the referral to the specialist being made?
- ~ What will the specialist do?
- ~ Will visits to the specialist be a one-time event, for a limited time, or ongoing?
- ~ Who will get the reports of the evaluation?
- ~ How will the findings be coordinated with the individual's overall health care?

Hospitalization

- ~ How long can we expect to stay in the hospital?
- ~ What are the names of the attending and consulting doctors?
- ~ Will a family member be able to stay with the hospitalized individual?
- ~ Does the hospital have a program to prepare children before they are hospitalized?

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Communicating With Health Care Providers (cont.)

(Continued from page 12)

- ~What will be done during the hospitalization?
- ~Is it possible to estimate a cost for some or all of the hospitalization and testing?
- ~Can we meet with someone to help us understand what will or will not be covered by insurance?

Surgery

- ~ Explain the surgery in everyday terms
- ~ What will happen if the surgery is not done?
- ~ How long will it take to recuperate?
- ~ What are the possible complications?
- ~ Is there anything to be gained from a second opinion?
- ~ Will therapy or follow-up surgery be necessary?
- ~ What will be the long-term effects of the surgery?

Insurance

- ~ Request referral for pre-registration and pre-authorization/pre-approval
- ~ Know your annual deductible (per individual and for the family as a unit)
- ~ Know your lifetime cap on coverage
- ~ Ask about protections provided by HIPAA (Health Insurance Portability and Accountability Act) and GINA (Genetic Information Non-discrimination Act).
- ~ Stay sensitive to privacy issues. Who needs to know the information? If in doubt, don't disclose.
- ~ Never give a blanket disclosure authorization

Remember

You know your child better than anyone else. You are the expert on the needs of your child. Take a pro-active role in any medical decision. Arm yourself with as much knowledge as possible to make informed decisions with any medical recommendations or procedures.

Visit WWW.NNPDE.ORG for links to additional information.

NNPDF Book Corner

The Complete IEP Guide: How to Advocate for Your Special Ed Child

Comments from one Amazon.com customer: "Whether you are new to the world of "special ed" or you are a seasoned veteran, attorney, Lawrence M. Siegel's book will guide you through the advocacy maze with ease. It is a valuable resource of information, including, but not limited to: an overview of special ed and the IEP process; sample IEP forms and letters; organization and planning tips; a listing of support groups, advocacy organizations and federal and state departments of education. Laws and procedures change frequently, and regulations can be different from state to state, so new editions are printed periodically. You can even take advantage of an "update service" that offers a 35% discount on any future purchases."

By Lawrence M. Siegel, NOLO; 5th edition (January 22, 2007), English, ISBN-10: 1413305105, ISBN-13: 978-1413305104.

Easy for You to Say: Q's and A's For Teens Living With Chronic Illness or Disabilities

"...this very unusual book, aimed exclusively at teens who are disabled or who have a chronic illness, focuses on individual needs. Written by a Canadian physician who works with adolescents, it is filled with very personal, even courageous questions from teens with varied medical conditions--from spina bifida to cystic fibrosis, to kidney disease. There are a few fairly general chapters--on family dynamics, friendship, and recreation. But the best sections concern medical issues and sexuality. Here, teens ask about drug interactions and side effects (Kaufman has included several useful appendixes) and broach some very specific sex-related concerns. At times, Kaufman sounds a little too much like an all-wise counselor, but she's always frank and

careful to remind teens to act safely and responsibly, especially in terms of sex. Kaufman might have given teens stronger encouragement to communicate with their doctors or parents, but as it is, this is loaded with the kind of information disabled teens often need but may be too embarrassed to ask for."

By Miriam Kaufman, Firefly Books; Revised edition (August 6, 2005), English, # ISBN-10: 1554070783, # ISBN-13: 978-155407078.



The 36-Hour Day: A Family Guide to Caring for Persons with Alzheimer Disease, Related Dementing Illnesses, and Memory Loss in Later Life

While this text is directed to those dealing with Alzheimer disease, many of the tips and suggestions are important for all people dealing with an illness that leads to memory loss and dementia. From the Library Journal regarding this new, 3rd edition, "The heart of the guide remains unchanged, focusing on helping families cope with this progressive and irreversible disease. Besides tips on how to care for the demented during the various stages of the disease (for example, place a picture of a toilet on the bathroom door), the text discusses the different kinds of help available and how to seek it. Financial and legal issues are well covered, while sections on nursing homes and other alternative living arrangements provide advice and practical suggestions. Appendixes list recent books, videos, web sites, and U.S. and international organizations."

3rd Edition By Nancy L. Mace and Peter V. Rabins, Wellness Central (November 1, 2006), English, ISBN-10: 0446618764, ISBN-13: 978-0446618762.

NIEMANN-PICK DISEASE GLOSSARY OF CLINICAL TERMS

Acid Sphingomyelinase (ASM): This is a lysosomal enzyme that breaks down a substance called Sphingomyelin. This enzyme has decreased function to a greater or lesser extent in NPA/B disease.

Ascites: An abnormal accumulation of serous fluid in the abdominal cavity.

Ataxia: failure of muscular coordination; irregularity of muscle action or movement.

Atrophy: A wasting or decrease in size of a body organ, tissue, or part owing to disease, injury, or lack of use.

Autosomal: Refers to chromosomes 1 to 22; i.e. any chromosome other than the sex chromosomes.

Blood Brain Barrier: A highly selective barrier formed by blood vessels and a type of brain cell called astrocytes that allow only appropriate molecules to cross from blood to brain; e.g. oxygen and sucrose. It aims to keep harmful molecules out of the brain.

Brainstem: The 'stalk' of brain tissue that connects the spinal cord, cerebellum and cerebral hemispheres. Control of eye movements, breathing, heart rate, blood pressure and the nerves of the head and neck resides in the brainstem. Brain death is defined by irreversible loss of the brainstem function.

Carrier: an individual heterozygous for a single recessive gene.

Cataplexy: A sudden loss of muscle tone, usually evoked by a strong stimulus such as laughter or anger. Cataplexy is believed to represent a fragment of rapid eye movement (REM) sleep that intrudes into otherwise normal consciousness. It correlates with dysfunction of the upper brainstem.

Cerebellum: Part of the brain located behind the brain stem under the main mass of the brain. Is known to be responsible for the coordination of movement and contains Purkinje cells which are badly affected in NPC.

Cherry-red Spot or Macula: The appearance of the retina in some neuro-metabolic disorders. The normal central retina is red and appears as a spot surrounded by grey to white appearing retinal cells which are pale due to accumulation of metabolic storage materials.

Cholesterol: A fatty substance known as a sterol that is a major component of cell membranes, especially the main (plasma) membrane. It is also required for digestive bile acids and sterol hormones. Some cholesterol is taken in from certain foods but most is produced by the cells of the body, notably the liver. A constant supply of cholesterol is needed for cell growth and maintenance and is distributed in the blood stream by LDL

Cholesteryl Esterase: A lysosomal enzyme that cuts (hydrolyses) LDL cholesterol molecules free from their fatty acid tails. Wolman's disease results from defects in this enzyme.

Chromosome: one of the threadlike structures consisting of chromatin/DNA and carry genetic information arranged in a linear sequence.

Common mutation: A mutation in a gene that occurs in a high percentage of patients affected by a specific disease. E.g. I1061T in NPC.

Congenital: Any trait or condition that exists from birth.

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Consanguinity: genetic relationship. Consanguineous individuals have at least one common ancestor in the preceding few generations.

Cortex: The outer layer of any organ, as in the cerebral and cerebellar cortex. In the brain, the cortex consists of neurons (grey matter). It is subdivided into Paleocortex, archicortex, and neocortex, reflecting its complexity and similarities to other animals (e.g., paleocortex, with only three cell layers is present in the least complex organisms, such as reptiles, where as neocortex, with six layers, is best developed in humans and primate apes).

Deletion: the loss of a segment of the genetic material from a chromosome.

DNA (deoxyribonucleic acid). The molecule that encodes genetic information for protein and for cellular functioning. The four nucleotides in DNA contain the bases: adenine (A), guanine (G), cytosine (C), and thiamine (T) held together by weak bonds between base pairs of nucleotides. In nature, base pairs form only between A and T and between G and C; thus the base sequence of each single strand can be deduced from that of its partner (like a mirror image).

Dominant (allele): Only one copy of the allele is needed to produce a certain characteristic. E.g. if you have one allele for brown eyes and one for blue, brown is dominant and you will have brown eyes.

Dysarthria: a speech disorder that is due to a weakness or incoordination of the speech muscles. Speech is slow, weak, imprecise or uncoordinated.

Dysphagia: difficulty swallowing or pain with swallowing.

Dyspraxia: the partial loss of the ability to coordinate and perform certain purposeful movements and gestures in the absence of motor or sensory impairments.

Dystonia: disordered muscle tone

Enzyme: A protein that acts as a catalyst, speeding the rate at which a biochemical reaction proceeds but not alternating the direction or nature of the reaction. Example, sphingomyelinase

Esterification: A type of chemical reaction carried out by an enzyme. In NPC it relates to the attaching of long chain fatty acids to cholesterol and renders the cholesterol molecule

chemically inactive. A molecule of water is given off in this reaction. Reversing this process and separating cholesterol from the fatty acid is called hydrolysis.

Extrapyramidal: the extrapyramidal system is a neural network located in the brain that is part of the motor system involved in the coordination of movement. The extrapyramidal system can be affected in a number of ways, which are revealed in a range of extrapyramidal symptoms such as akinesia (inability to initiate movement) and akathisia (inability to remain motionless).

Fibroblasts: Cells found in connective tissue associated with healing wounds. Skin fibroblasts often grown to be used extensively in laboratory testing.

Filipin: A blue substance used to stain fibroblast cells. Appears fluorescent under a special microscope.

Founder effect: A gene mutation observed in high frequency in a specific population due to the presence of that gene mutation in a single ancestor or small number of ancestors

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Gene: The fundamental physical and functional unit of heredity. A gene is an ordered sequence of nucleotides located in a particular position on a particular chromosome that encodes a specific functional product (i.e., a protein or RNA molecule). See gene expression. Example NPC1, NPC2

Genetic counseling: the educational process that helps individuals, couples, or families to understand genetic information and issues that may have an impact on them.

Genotype: the genetic makeup of an individual; usually refers to gene structure of a particular gene being analyzed.

Hepatomegaly: enlargement of the liver beyond its normal size.

Hepatosplenomegaly: enlargement of the liver and the spleen.

Heterozygous: A heterozygous state exists if the corresponding genes at a particular location on each of a related pair of chromosomes are different from each other. The different genes may be normal or disease causing.

Homozygous: A homozygous state exists if the corresponding genes at a particular location on each of the related pair of chromosomes are the same. The genes may both be normal or disease causing.

Hypotonia: A condition of abnormally low muscle tone (the amount of tension or resistance to movement in a muscle), often involving reduced muscle strength.

Inborn errors of metabolism: inherited diseases resulting from alterations in genes that code for enzymes.

Interstitial lung disease: Interstitial Lung Disease (ILD) is a general term that includes a variety of chronic lung disorders. When a person has ILD, the lung is affected in three ways. First, the lung tissue is damaged in some known or unknown way.

(Interstitial lung disease—cont.)

Second, the walls of the air sacs in the lung become inflamed. Finally, scarring (or fibrosis) begins in the interstitium (or tissue between the air sacs), and the lung becomes stiff.

Jaundice: a yellowing of the skin, conjunctive (a clear covering over the sclera, or whites of the eyes) and mucous membranes caused by increased levels of bilirubin in red blooded animals.

Lipid: Fat. Molecules that prefer to be in chloroform rather than water (hydrophobic, or won't dissolve in water). This property is important for the formation of membranes (very high lipid content) which allow separation of different biologically active solutions.

Lysosomal storage disease: a group of over thirty disorders that result from defects in lysosomal function leading to accumulation of metabolic by products in organs and tissues.

Lysosome: A membrane-bound organelle (soma = body) within the cell that contains enzymes that lyse (cut up) proteins, lipids, nucleic acids, and sugars. The cell's recycling center.

Mutations: Heritable changes in the DNA of cells, generally thought of as changes that could be detrimental to the operation of the cell.

Neurodegenerative: Causing loss of function of a system due to the loss of or damage to cells in the nervous system.

Neuropathic: having to do with damage to a nerve.

Newborn screening: examining blood samples from a newborn infant to detect disease-related abnormalities or deficiencies in gene products.

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Nystagmus: A rapid, involuntary, oscillatory motion of the eyeball; can occur be up-and-down (vertical) or back-and-forth (horizontal).

Obligate heterozygote: (synonym: obligate heterozygote) An individual who may be clinically unaffected but who must carry a gene mutation based on analysis of the family history; usually applies to disorders inherited in an autosomal recessive and X-linked recessive manner

Phenotype: the outward appearance or observable physical characteristics of an individual caused by that individual's genes interacting with the environment

Polymorphism: Natural variations in a gene, DNA sequence, protein, or chromosome that have no adverse effect on the individual and occur with fairly high frequency in the general population

Prenatal diagnosis: examining fetal cells taken from the amniotic fluid, the primitive placenta (chorion), or the umbilical cord for biochemical, chromosomal, or gene alterations.

Presymptomatic diagnosis: diagnosis of a genetic condition before the appearance of symptoms.

Proband: individual in a family whose diagnosis brought the family to medical attention.

Prognosis: prediction of the course and probable outcome of a disease.

Purkinje Cells: Large neurons of the cerebellum where they form a single layer. They are thought to be the only cells of the cerebellum with outgoing connections to the rest of the brain. These cells are vulnerable to damage and death in NPC.

Recessive: The affects of a gene that are masked by the activity of the same gene on the other chromosome (see dominant).

Saccade: an eye movement in which the eyes jump from one point to another, as from one word to the next while reading or around a room when searching for something.

Splenomegaly: Enlargement of the spleen.

Systemic: pertaining to or affecting the body as a whole.

Targeted mutation analysis: (synonym: allele-specific mutation analysis) Testing for one or more specific mutations, particularly when there are know founder mutations in a given population.

Thrombocytopenia: A lower than normal number of platelets in the blood.

Vertical Supranuclear Gaze Palsy (VSGP): an inability to look up or down a result of cerebral impairment.

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