Pediatric Clinical Support:
Niemann-Pick Type C

Also known as Juvenile Niemann-Pick Disease, Niemann-Pick Type C (NPC) is a lysosomal storage disease, which affects the way fats, lipids, and cholesterol is eliminated from the body. When a buildup of fats, lipids, and cholesterol occur in the organs, the result can be critical, often affecting the liver, spleen, bone marrow, and the brain. This can also lead to a shortened life expectancy. There are several different types of Niemann-Pick syndrome (see Table 1 on reverse side). Type C presents with sensorineural hearing loss, in addition to abnormal lipid metabolism (Healthline overview, 2012), and can be diagnosed as early as in infancy or as late as the sixth decade of life.

Characterized by:
Usually, a diagnosis of lysosomal storage disease is confirmed at a young age (excluding Type E); swelling of body organs, including the spleen and liver; lung and brain damage; poor muscle tone and/or body movements; seizures. See Table 1 on reverse side for a fuller understanding of all the types of Niemann-Pick Disease.

Genetic manifestation:
Niemann-Pick Type C affects 1 out of 150,000 individuals. Mutations occur in NPC1 and NPC2 genes located on Chromosome 18. Nearly half of all people diagnosed with Type C present before age 10, however, symptoms may be identified as late as the sixth decade of life.

Audiological Considerations: Hearing evaluations can determine severity of loss and should be constantly monitored, as hearing loss progresses with this disease. Appropriate amplification or cochlear implantation can be chosen based off of the individual's audiogram. Assistive listening devices can be used in addition to or in conjunction with amplification and/or cochlear implants.

Educational and Professional Considerations: If the child is well-enough to attend school, NPC should be listed in the child's IEP. If the child with NPC is diagnosed with hearing loss, it should be indicated and addressed as well. The IEP should include access to information, effective communication strategies, and other approaches for easier communication. Quality of life considerations should be addressed, including those related to hearing loss as well as diet, with all professionals who interact with the child.

Online Support Sources:
http://www.nnpdf.org/npdisease_09.html
http://www.parseghian.org/aboutniemannppickc.html
http://niemannpick.nd.edu/
http://www.bripardun.com/npc.html

Online and other References:
Erickson Gabby, A (2012). Niemann-Pick Disease. Retrieved from: http://www.healthline.com/health/niemann-pick-disease#Overview1
Patterson M (2013). Niemann-Pick Disease Type C. Retrieved from: http://www.ncbi.nlm.nih.gov/books/NBK1296/
http://www.parseghian.org/aboutniemannppickc.html
https://www.counsyl.com/services/family-prep-screen/diseases/niemann-pick-disease-type-c/
http://www.nnpdf.org/npdisease_01.html

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	Type A and Type B (Type I)		Type C and Type D (Type II)		Type E
Age of Onset	Symptoms usually begin between 3-6mo of age for Type A and during late childhood and early adolescence for Type B		Symptoms usually appear around 5 years of age, though they can occur at anytime in life		Symptoms appear in adulthood
Description	Acid sphingomyelinase (ASM) is an enzyme that helps eliminate fat from the body. In this type of Niemann-Pick, ASM is not properly produced by the body and allows the fats within the body to build up, which causes cells to die and can lead to organ failure.		Type C and Type D (Type D is now considered a variation of Type C) cannot effectively remove cholesterol and other lipids from the body, causing buildup in the liver and spleen, and excessive fat buildup in the brain.		More research is needed to better understand Type E; however, this type primarily occurs in adults.
Symptoms	**Type A** Enlarged liver and spleen leads to abdominal swelling; swollen lymph nodes; red spot inside of eye; difficulty feeding; poor muscle tone; brain and lung damage; frequent respiratory infections	**Type B** Enlarged liver and spleen leads to abdominal swelling; delayed growth/failure to achieve developmental milestones on time; poor coordination, mental retardation, and psychiatric disorders; respiratory infections	**Type C and Type D** Difficulty moving limbs/loss of muscular function; enlarged spleen or liver (or both); jaundice; difficulty learning and speaking; decline of intellect (dementia); seizures and tremors; difficulty moving the eyes, especially when moving up and down; difficulty walking or being unsteady/clumsy; hearing or vision loss; brain damage		**Type E** Swelling of the spleen and brain; Neurological problems due to swelling in the nervous system
Diagnosis	**Type A and Type B** The amount of ASM in white blood cells, either in blood or bone marrow, will determine a Niemann-Pick diagnosis. Genetic testing is also recommended to determine if a parent is a carrier of the syndrome.		**Type C and Type D** A skin biopsy will be taken and then cell growth will be analyzed to determine diagnosis of this type of Niemann-Pick. Movement and storage of cholesterol in the cells is also monitored.		
Treatment	**Type A** There is no known treatment at this time.	**Type B** Bone marrow transplants; gene therapy; enzyme replacement therapy. Research is ongoing.	**Type C** Miglustat, an enzyme inhibitor, prevents the body from producing cholesterol, reducing fatty buildup	**Type D** There is no known treatment at this time.	
Prognosis	**Type A** Most children die in infancy however some live up to age 4.	**Type B** Children may live to into late childhood or early adulthood; quality of life is poor	**Type C and Type D** Type C is always fatal, but outlook depends on onset of diagnosis. Prognosis varies but the child is likely to live into late teens/early 20s.		

Table 1. ▸ Breakdown of Niemann-Pick Disease by type.

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