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:**
http://www.parseghian.org/aboutniemannppickc.html
http://www.nnpdf.org/npdisease_01.html
Pediatric Clinical Support: Niemann-Pick Type C

<table>
<thead>
<tr>
<th>Type A and Type B (Type I)</th>
<th>Type C and Type D (Type II)</th>
<th>Type E</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Age of Onset</strong></td>
<td>Symptoms usually begin between 3-6mo of age for Type A and during late childhood and early adolescence for Type B</td>
<td>Symptoms usually appear around 5 years of age, though they can occur at anytime in life</td>
</tr>
<tr>
<td><strong>Description</strong></td>
<td>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.</td>
<td>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.</td>
</tr>
<tr>
<td><strong>Symptoms</strong></td>
<td>Type A</td>
<td>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</td>
</tr>
<tr>
<td><strong>Diagnosis</strong></td>
<td>Type A</td>
<td>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.</td>
</tr>
<tr>
<td><strong>Treatment</strong></td>
<td>Type A</td>
<td>There is no known treatment at this time.</td>
</tr>
<tr>
<td><strong>Prognosis</strong></td>
<td>Type A</td>
<td>Most children die in infancy however some live up to age 4.</td>
</tr>
</tbody>
</table>

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