

International Niemann-Pick Disease Registry

Patient Information Sheet: Acid Sphingomyelinase Deficiency Niemann-Pick Disease

What is Niemann-Pick Disease?

Niemann-Pick diseases are a group of rare, inherited, metabolic conditions that can affect children and adults. These conditions are caused by specific genetic mutations and are pan-ethnic. The commonly recognised forms of the disease are:

- Acid Sphingomyelinase Deficiency (ASMD) Niemann-Pick disease type A and type B, represent opposite ends of a spectrum of the same disease, which is characterised by a deficiency of an enzyme which causes a build-up of toxic materials in the body's cells.
- Niemann-Pick disease type C is not caused by an enzyme deficiency, but the end result is the same; an accumulation of materials (lipids) in the body's cells.

What is Acid Sphingomyelinase Deficiency (ASMD) Niemann-Pick Disease?

ASMD Niemann-Pick Disease types A & B, rather than being two separate forms of the disease, actually represent the opposite ends of a spectrum of the same disease, both caused by a deficiency of the enzyme Acid Sphingomyelinase. Many variations exist within this spectrum, in terms of clinical symptoms and rate of progression:

- ASMD Niemann-Pick Disease Type A (NP-A):
ASMD NP-A is a rapidly progressive neurological disease that usually reveals itself within the first few months of a baby's life. Symptoms may include early feeding difficulties, failure to thrive and an abnormally large abdomen. Life expectancy rarely exceeds five years of age.
- ASMD Niemann-Pick Disease Type B (NP-B):
ASMD NP-B does not usually affect the brain and although growth may be slow, those affected will usually survive into adulthood, with many being able to lead a full life.
- Some patients may be described as having A/B variant, falling in the middle of the spectrum and exhibiting neurological problems which may become more apparent over time.

What causes ASMD Niemann-Pick Disease?

ASMD Niemann-Pick type A and B are genetically inherited conditions caused by an enzyme deficiency, which results in a build-up of toxic materials in the body's cells. All types of Niemann-Pick disease are acquired through **autosomal recessive inheritance**; this means that both parents have to be carriers of the faulty gene (mutation). A mutation is a change or fault on a normal gene, causing it not to function as it should do. Parents seldom know that they are carriers of the disease and have no control over whether the disease will be transmitted to their child. In each pregnancy of a carrier couple, there is a 25% chance that they will both pass on this gene mutation to their child.

How common is it?

ASMD Niemann-Pick disease types A and B are very rare: it is estimated that they affect 1 in every 250,000 individuals.

What may happen in the condition?

ASMD Niemann-Pick type A begins in the first few months of life. Symptoms may include a combination of those listed below:

- feeding difficulties
- prolonged jaundice
- a large abdomen within 3 to 6 months
- progressive loss of early motor skills
- failure to thrive
- cherry red spot in the eye (not noticeable to parents)
- (generally) a very rapid decline leading to death by two to five years of age

The first symptoms of ASMD Niemann-Pick Disease type B are usually an enlarged liver and/or spleen in early childhood. Symptoms can include a progressive enlargement of organs, poor growth, susceptibility to respiratory infections, bleeding problems, bone pain and increased stress on the heart, but usually no neurological involvement. Most patients with ASMD NP-B will survive into adulthood, but not without experiencing health problems.

What is the outlook for a patient affected by ASMD Niemann-Pick Disease?

There is presently no cure for ASMD Niemann-Pick disease; however, recent advances in medical science have brought the hope of potential treatments. Patients will benefit from symptomatic treatments - individual medications that can help to ease the symptoms of the disease.

What are the chances of having another affected child?

In each pregnancy of a carrier couple, there is a 25% chance that they will both pass on this gene mutation to their child. Once a child has been diagnosed with ASMD NPD, it is usually possible to test future pregnancies, if the mutation carried by the parents and child has already been identified.

Who can I go to for help?

In the first instance, speak to your doctor or a member of your healthcare team, who will be able to provide you with a list of local support services/organisations. In addition you may wish to visit the website of the International Niemann-Pick Disease Alliance (INPDA), where you will find a list of Niemann-Pick specific support groups around the world: www.inpda.org

If you have specific questions relating to this leaflet, please contact:

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