

# International Niemann-Pick Disease Registry

## Patient Information Sheet: Niemann-Pick Disease Type C (NP-C)

### 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 Niemann-Pick Type C Disease (NP-C)?

NP-C is an extremely rare disease that affects multiple body organs and systems. The age of onset and rate of disease progression can greatly vary between individual patients. Children who develop neurological symptoms in early childhood are thought to have a more aggressive form of the disease and may not survive to adolescence, while others may remain symptom-free for many years.

Based on molecular genetic testing, NP-C is divided into two subtypes – NP-C1 and NP-C2 – with each caused by a different gene mutation. Approximately 95 per cent of NP-C cases are caused by genetic mutations in the NP-C1 gene, with the other five per cent caused by mutations in the NP-C2 gene.

The variable onset and progression of NP-C can make diagnosis challenging.

### What causes NP-C?

NP-C is a genetically inherited condition, caused by a protein 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?**

The incidence of disease is estimated as 1:120,000 (Source: Marie T.Vanier, Orphanet Journal of Rare Disease, 2010 Jun 3;5 (1):16.)

### **What may happen in the condition?**

Symptoms of NP-C vary with age of onset and from patient to patient, they may include:

- Jaundice at (or shortly after) birth
- An enlarged spleen and/or liver (hepatosplenomegaly)
- Difficulty with upward and downward eye movements (Vertical Supranuclear Gaze Palsy)
- Unsteadiness of gait, clumsiness, problems in walking (ataxia)
- Difficulty in posturing of limbs (dystonia)
- Slurred, irregular speech (dysarthria)
- Learning difficulties and progressive intellectual decline (Cognitive dysfunction - "dementia")
- Sudden loss of muscle tone which may lead to falls (cataplexy)
- Tremors accompanying movement and, in some cases, seizures
- Swallowing problems (dysphagia)

### **What is the outlook for a patient affected by NP-C?**

There is presently no cure for NP-C; however, recent advances in medical science have brought the hope of potential treatments. Patients will benefit from symptomatic treatments - individual medications that may help to slow progression and 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 NP-C, 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](http://www.inpda.org)

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

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