What is Niemann-Pick Disease?

Niemann-Pick diseases are a group of rare inherited lysomal storage disorders that can affect both children and adults.

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 which means that it does not perform the function that 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 per cent chance that they will both pass on this gene mutation to their child.

With a recessive condition, a person may be a carrier of a disease gene, but with no noticeable effect in their everyday lives and health.

There is currently no cure for Niemann-Pick disease, and treatments focus on managing symptoms. However, the landscape is rapidly changing, and clinical trials are actively recruiting in multiple countries, including the UK.

NPUK is a charitable organisation dedicated to making a positive difference to the lives of those affected by Niemann-Pick diseases. We raise much needed awareness, provide practical and emotional support, advice and information and facilitate research into potential therapies.

Please visit
npuk.org
for the most up-to-date information

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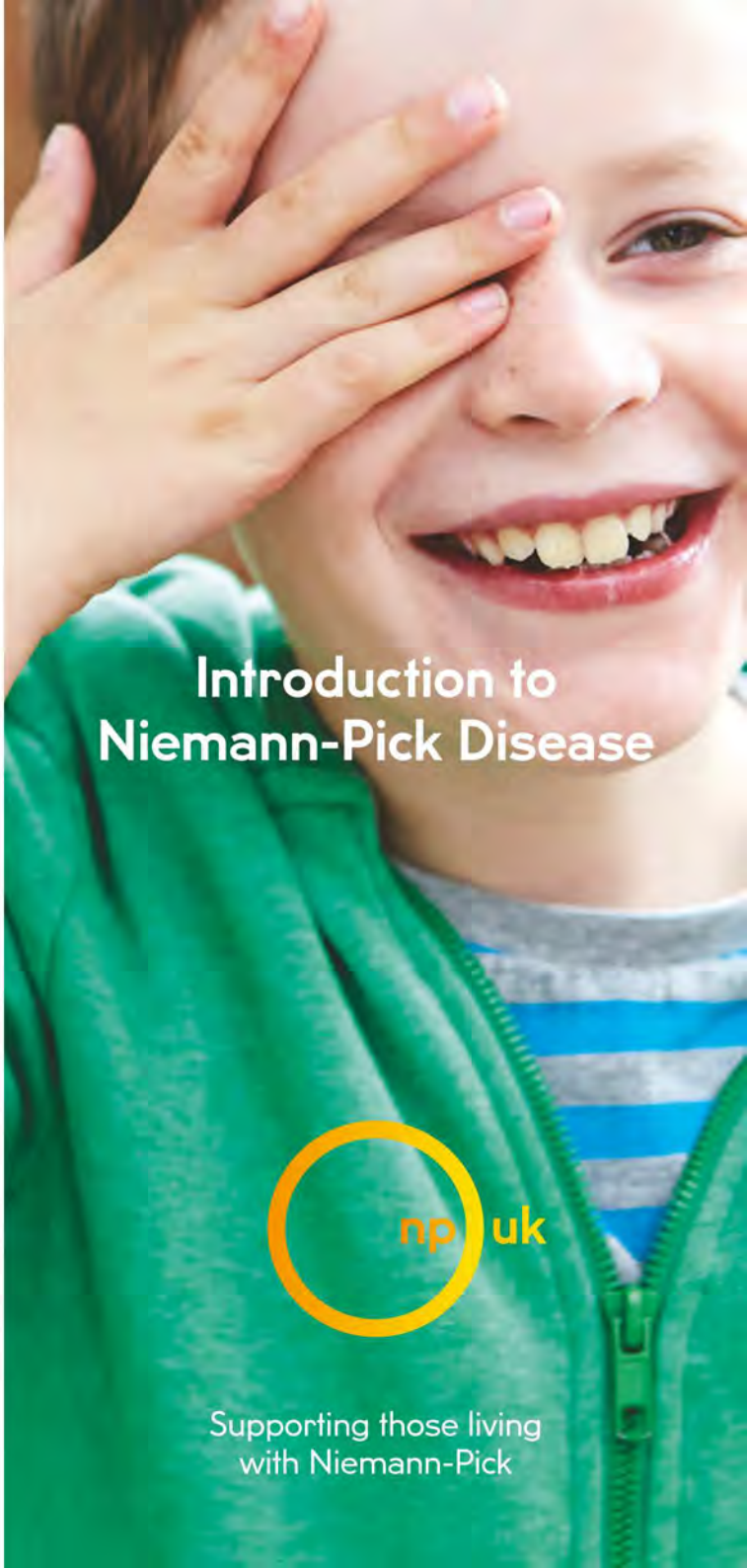

Introduction to Niemann-Pick Disease

Supporting those living with Niemann-Pick

There are two recognised forms of Niemann-Pick disease; they have similar clinical presentations, but quite separate causes:

1 Acid Sphingomyelinase Deficiency (ASMD)
ASMD — also known as Niemann-Pick Disease Type A and Type B — represent opposite ends of a spectrum of the same disease, characterised by a deficiency of an enzyme which causes a build-up of toxic materials in the body's cells. Many variations exist within this spectrum, in terms of clinical symptoms and rate of progression.

2 Niemann-Pick Disease Type C (NP-C)
NP-C is caused by a protein deficiency, not an enzyme deficiency, but the end result is the same; an accumulation of materials (lipids) in the body's cells. The age of onset and rate of disease progression can greatly vary between individual patients.

- **You may also read about Niemann-Pick Disease Type D**
This refers to a certain NP-C mutation occurring only in Nova Scotia.

Acid Sphingomyelinase Deficiency (ASMD)

Niemann-Pick Disease Type A (NP-A) and Type B (NP-B) are caused by a lack of the enzyme Acid Sphingomyelinase, leading to a build-up of toxic materials in the body's cells and organs.

- **NP-A** is a rapidly progressing neurological disease. Symptoms may include early feeding difficulties, failure to thrive and an abnormally large abdomen. Life expectancy rarely exceeds three years of age.
- **NP-B** does not affect the brain and, although growth may be slow, those affected will survive into adulthood, with many being able to lead a full and active life.
- A small number of 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.
- There are three common mutations that account for NP-A in the Ashkenazi Jewish population, and the estimated incidence is ~1 in 40,000. The incidence in other populations is not known, but it is considered extremely rare and estimated to be about 1 in 10 million.
- The incidence of NP-B is estimated as 1 in 250,000 in the general population.

Niemann-Pick Disease Type C (NP-C)

is a devastating neurodegenerative disease that affects infants, children and adults. It is caused by an accumulation of lipids (fats) in the liver, brain and spleen.

- The brain and other organs are affected, leading to progressive intellectual decline, loss of mobility and motor skills and seizures.
- Speech can become slurred and swallowing problems may develop.
- 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.
- A symptom that is particularly suggestive of NP-C is difficulty with upward and downward eye movement.
- Where onset is later, psychological problems and dementia are often experienced.
- The incidence of NP-C is widely reported at 1 in 120,000, although recent evidence suggests this may be an under-estimate.