

Advancing care for Niemann-Pick disease type C (NPC):

## Key domains help measure NPC's progression and severity at any point in time

### NPC can be challenging, including its progression

NPC is an ultra-rare and relentlessly progressive disease that causes irreversible damage. Delaying disease progression is critical, and an accurate diagnosis is crucial for early intervention and effective treatment. Diagnosis can be difficult, because NPC symptoms may seem to be related to other conditions. However, once NPC is diagnosed, there are NPC-specific scoring systems that doctors may use to help assess disease severity and progression.

### Tracking the impact of NPC by using key domains

Doctors will assess symptoms by focusing on domains or areas impacted by NPC, within a validated scale or measuring system. The use of a validated scale helps assess how serious symptoms are and how they have changed over time.

It also provides a way to evaluate the impact of treatment and see how you or your loved one may be responding to treatment.



### 4 KEY DOMAINS

These domains have been identified as **some of the most important** by NPC expert clinicians, caregivers, and patients, and allow for an assessment of NPC symptom progression.

NPC disease progression can be identified by observing symptoms within each domain, on a scale from mild to severe\*



#### AMBULATION

1 Clumsiness >>> Wheelchair dependence 5



#### FINE MOTOR SKILLS

1 Slight dysfunction >>> Severe muscle impairment 5



#### SWALLOW

1 Cough while eating >>> Feeding tube dependence 5



#### SPEECH

1 Slurred or slow speech >>> No communication 5



A 1-category worsening in any domain, corresponding to a 1- to 2-point change in total score, is considered **clinically meaningful**.

\*A score of 0 is normal for all domains.

# A validated scale helps provide a benchmark for NPC progression—and treatment progress

Your doctor may refer to the **Niemann-Pick type C Clinical Severity Scale (NPCCSS)** to measure how the disease is progressing. This scale was developed specifically to measure NPC.

The validated 17 domain NPCCSS was developed to assess NPC disease severity and progression but was considered too complex for doctors to assess patients in their everyday practice.



When you consider how effective a treatment may be, focus on improvement across the domains identified as **most important** by **NPC expert clinicians, patients, and caregivers**.



## THE 5-DOMAIN NPCCSS

Focuses on the domains considered most important by caregivers, patients, and doctors, which includes cognition.

- ✓ Allows a comprehensive assessment of symptoms and their impact by focusing on the **most relevant areas**
- ✓ Includes the domains **most relevant to clinicians, patients, and caregivers**
- ✓ Change in any domain, indicating **further progression of NPC**, was seen as **meaningful** by caregivers and patients



## THE RESCORED 4-DOMAIN NPCCSS

Revised to more accurately assess changes in the course of NPC over a 12-month period.

- ✓ The **cognition domain was removed** from the 5-domain NPCCSS and the scoring for the **swallowing domain was rescored** to more accurately measure swallow function and NPC progression
- ✓ Caregivers, patients, and healthcare providers consider the domains of swallow, ambulation, and speech to be the **most clinically important symptoms to measure**



Observe symptoms closely and watch for changes to assess disease progression. Share the details with your doctor to inform a plan for treatment.