

# Measuring Functional Abilities and Assistance Level in People with Niemann-Pick Disease Type C and GM2 Gangliosidoses Taking Levacetylleucine

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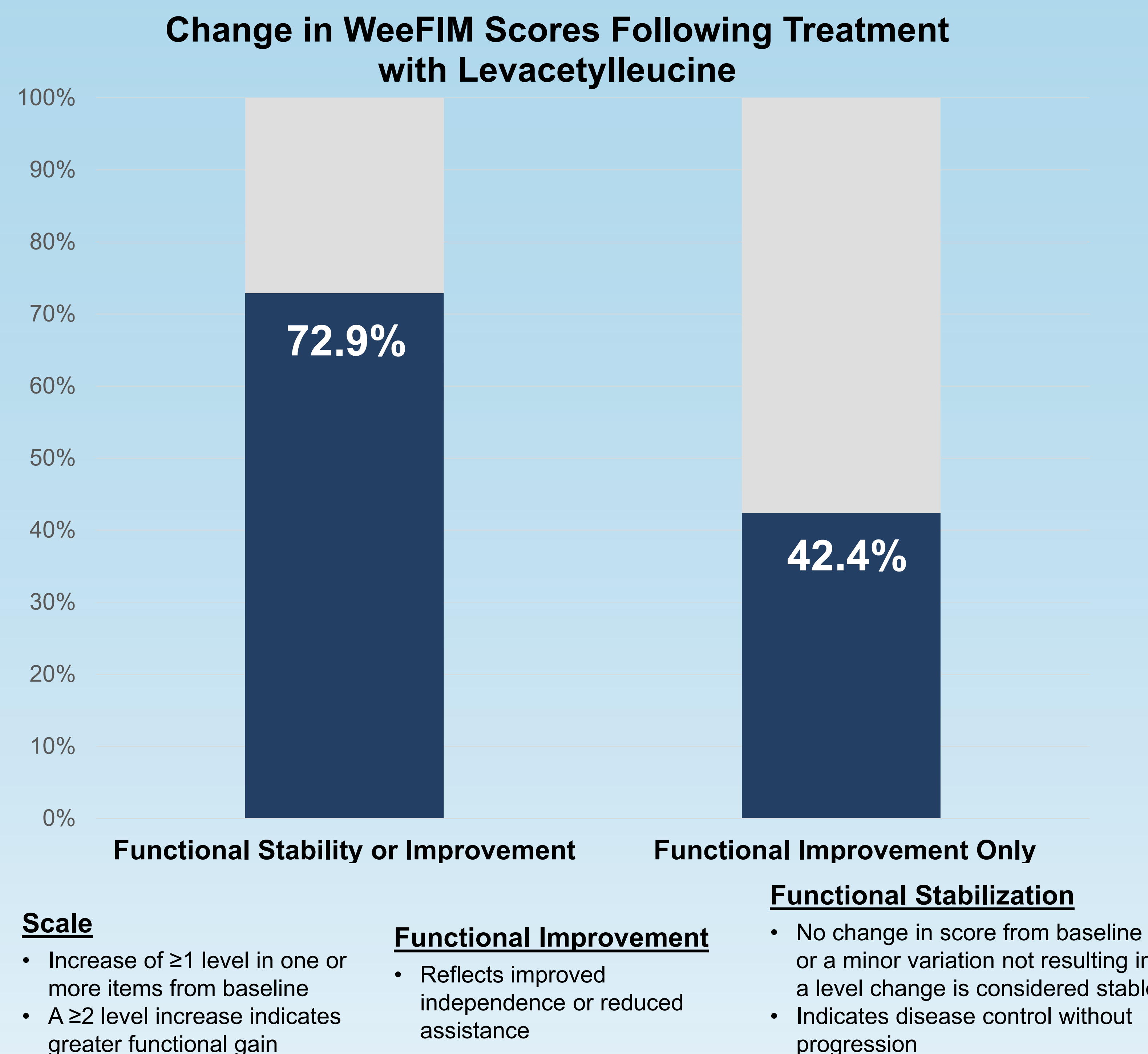
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## Background

- A need exists for the generation of real-world patient reported outcome (PRO) data for Niemann-Pick disease type C (NPC) and GM2 gangliosidoses (GM2) therapies<sup>1</sup>
- Levacetylleucine was approved by the US FDA for the treatment of neurological manifestations of NPC in adults and pediatric patients weighing  $\geq 15$  kg<sup>2</sup>
- Levacetylleucine is an investigational drug in GM2 gangliosidoses and has not yet been approved by any health authority
- The Functional Independence Measure for Children (WeeFIM<sup>®</sup> [1]) is one of the most respected pediatric outcomes management tools in the market<sup>3</sup>
- Developed from the World Health Organization (WHO) bio-psycho-social model, the WeeFIM is a validated tool assessing functional independence in children
- Utilizing a validated, well recognized assessment provides standardized real-world insight into the treatment journey
- For this analysis, the WeeFIM<sup>®</sup> assessment was adapted for adult participants and administered to people living with NPC and GM2 gangliosidoses receiving levacetylleucine
- The WeeFIM<sup>®</sup> has conceptual validity across age groups as it assesses core functional domains that are age-agnostic<sup>4</sup>

## Results

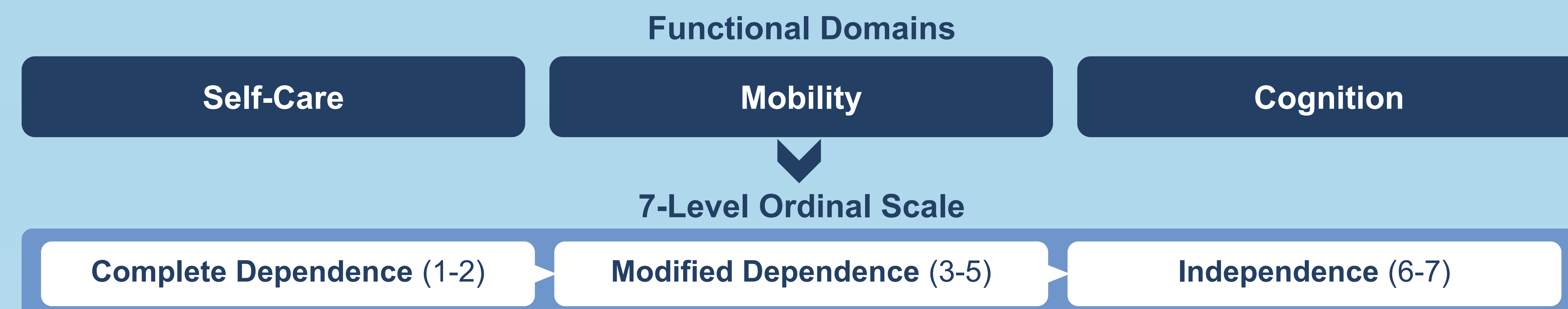
- There were a total of 33 participants (aged 2-68) with baseline and follow-up assessments; 27 were people with NPC and 6 were people with GM2
- Among the 33 participants, 45.5% were male (N=15), 54.5% female (N=18) and 51.5% (N=17) were aged <18 years
- Total scores for the assessment are calculated by adding up all domain scores (Self-Care, Mobility, and Cognition) for each assessed individual for both baseline assessment and all subsequent assessments
- Functional stability or improvement is defined in this assessment as the assessed individual's total score in the most recent assessment being the same as or better than baseline total score
- Following the initiation of levacetylleucine, 72.9% (N=24) showed either functional stability or improvement, with 42.4% (N=14) demonstrating functional improvement
- Analysis indicated improvement or stability across one or more functional domains in all individuals
- Adverse events were monitored and reported



## Methods

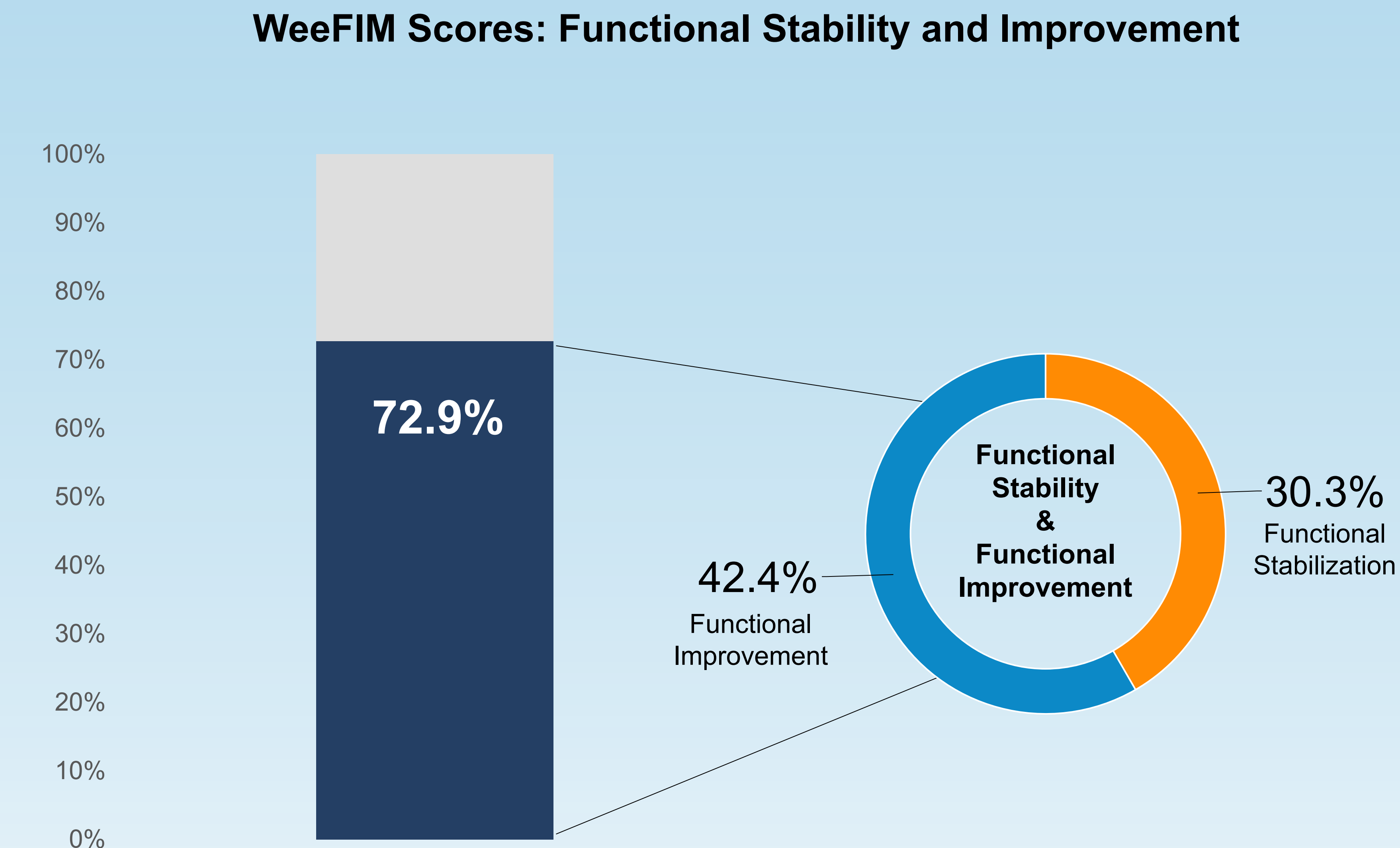
- The 18-item questionnaire is administered telephonically to the caregiver or person with NPC or GM2 assessing self-care, mobility, and cognition on a 7-level ordinal scale, and categorized as "Complete Dependence" (1-2), "Modified Dependence" (3-5), or "Independence" (6-7) (Figure 1)
- Total WeeFIM<sup>®</sup> scores range from 18 to 126, with a higher score indicating more independence, and a lower score indicating more dependence
- Participants were prescribed and received orally administered levacetylleucine 2-3 times a day based on weight-tiered dosing
- Baseline surveys were administered to caregivers or people with NPC or GM2 at Week 0 upon initiation of levacetylleucine treatment and with follow-up assessments conducted at a mean duration of 27.3 weeks post-treatment initiation of levacetylleucine

Figure 1. Overview of the WeeFIM<sup>®</sup> Instrument: Domains & Rating Scale



## Conclusion

- **42.4% (14/33) of the participants demonstrating real world symptom improvement both aggregately and across one or more domains is compelling**
- **A further 30.3% of participants (10/33) reported disease stabilization**
- **Longitudinal collection of PROs in people with NPC and GM2 will assess treatment adherence and correlation to symptom progression to further evaluate treatment effectiveness**



## References

1. Patterson MC, et al. Orphanet J Rare Dis. 2020, 15:104. 2. FDA. FDA Approves New Drug to Treat Niemann-Pick Disease, Type C. U.S. Food and Drug Administration; 2024 Sep 24. 3. Uniform Data System for Medical Rehabilitation (UDSMR). Pediatric Rehab: The WeeFIM II<sup>®</sup> System is the most respected pediatric outcomes management tool in the market. 2025. 4. Msall ML, et al. *Clinical Pediatrics*. 1994, 33(7):421-430.

## Acknowledgements

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## Disclosures

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