

# The situation of care in Canavan Patients

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## Introduction

Canavan disease (CD) is a rare hereditary disease, caused by a deficiency of the enzyme aspartoacylase, which leads to a pathological accumulation of N-acetylaspartate in the brain. Affected patients typically show a psychomotor developmental arrest within the first months of infancy. They suffer from severe neurological disabilities with helplessness in their first years and a reduced life expectancy, requiring extensive support (1). However, information about the needs of care and support in CD is sparse.

## Methods

After informed consent data from patients with confirmed diagnoses were analyzed. Information about the situation of care was collected the most recent follow-up visit of 28 CD patients using a standardized caregiver questionnaire. The data were compared with clinical parameters from the most recent follow-up visit.

The percentage of patients is shown in the Y-axis. Age distribution [years] is shown in the X-axis: 0-2 (n=9), 3-5 (n=8), 6-8 (n=5), 9-11 (n=2), 12-14 (n=3), 15+ (n=1).

## Results

Due to the severity of the disease and the symptoms that affect multiple functions of CD patients, the number of aids and supportive treatments per patient typically increases with age. Physical therapy is received by nearly all

patients across different age groups. The notable prevalence of spasticity (86%) underscores the increasing importance of orthoses and anti-spastic medication with aging. Only 25% of the 28 patients have gained the ability to walk, which is why the purchase of a wheelchair is essential. Feeding problems are common, including frequent choking, requiring speech therapy and the usage of a G-tube. Lack of or minimal verbal abilities require assistance in communication.

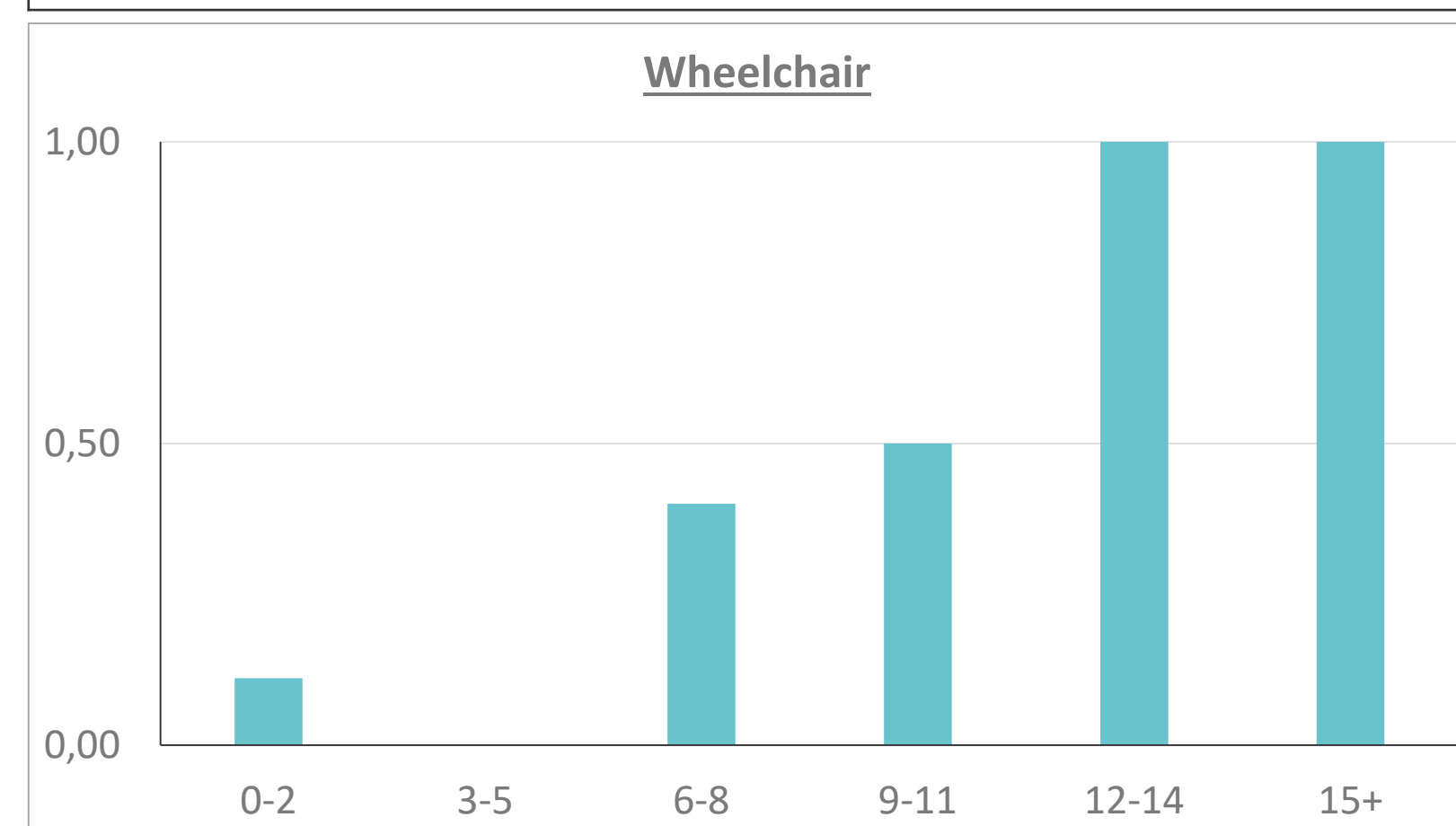


Chart 1. Percentage usage of wheelchair

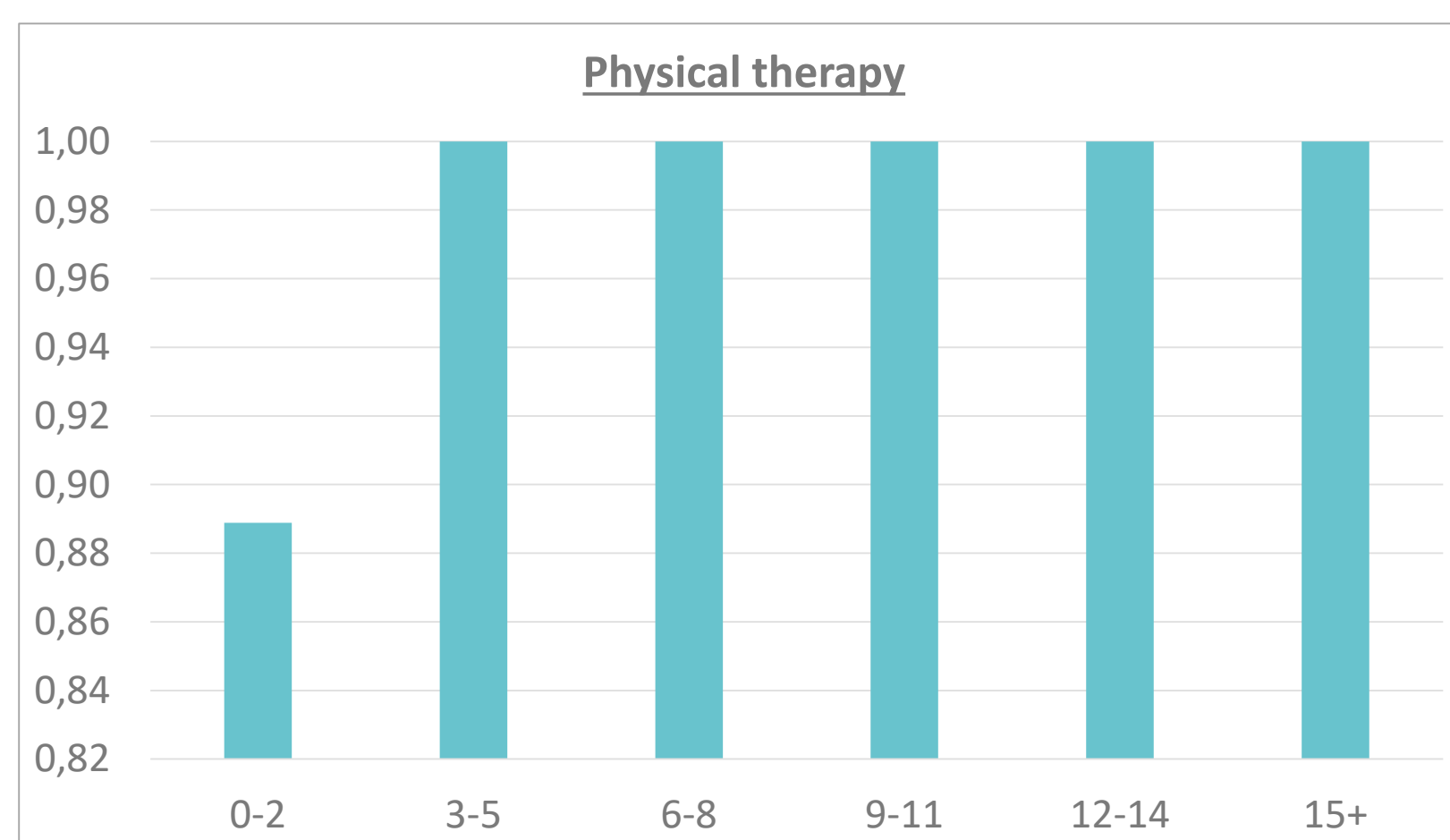


Chart 2. Percentage of patients receiving physical therapy

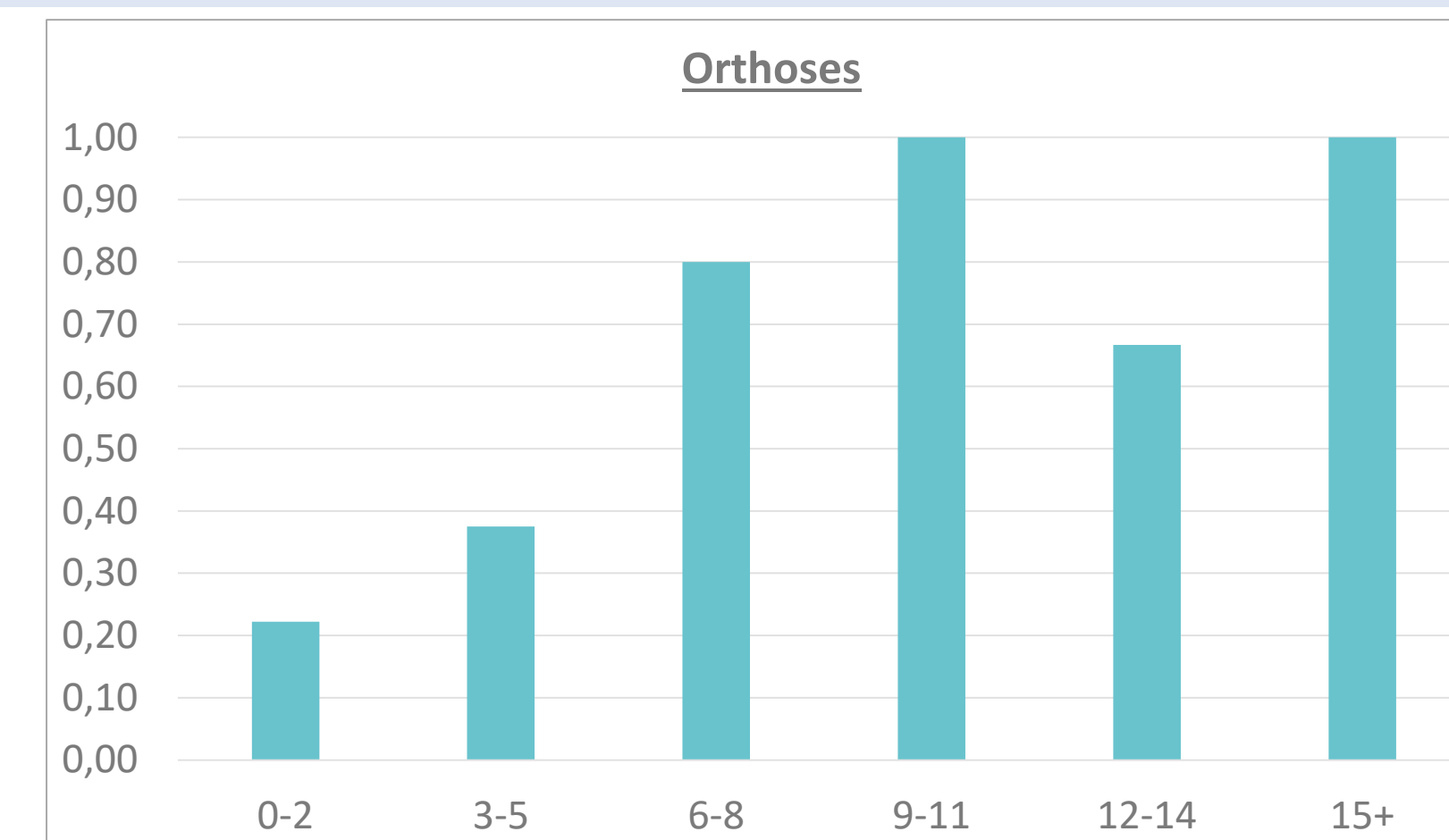


Chart 3 . Percentage of patients treated with orthoses

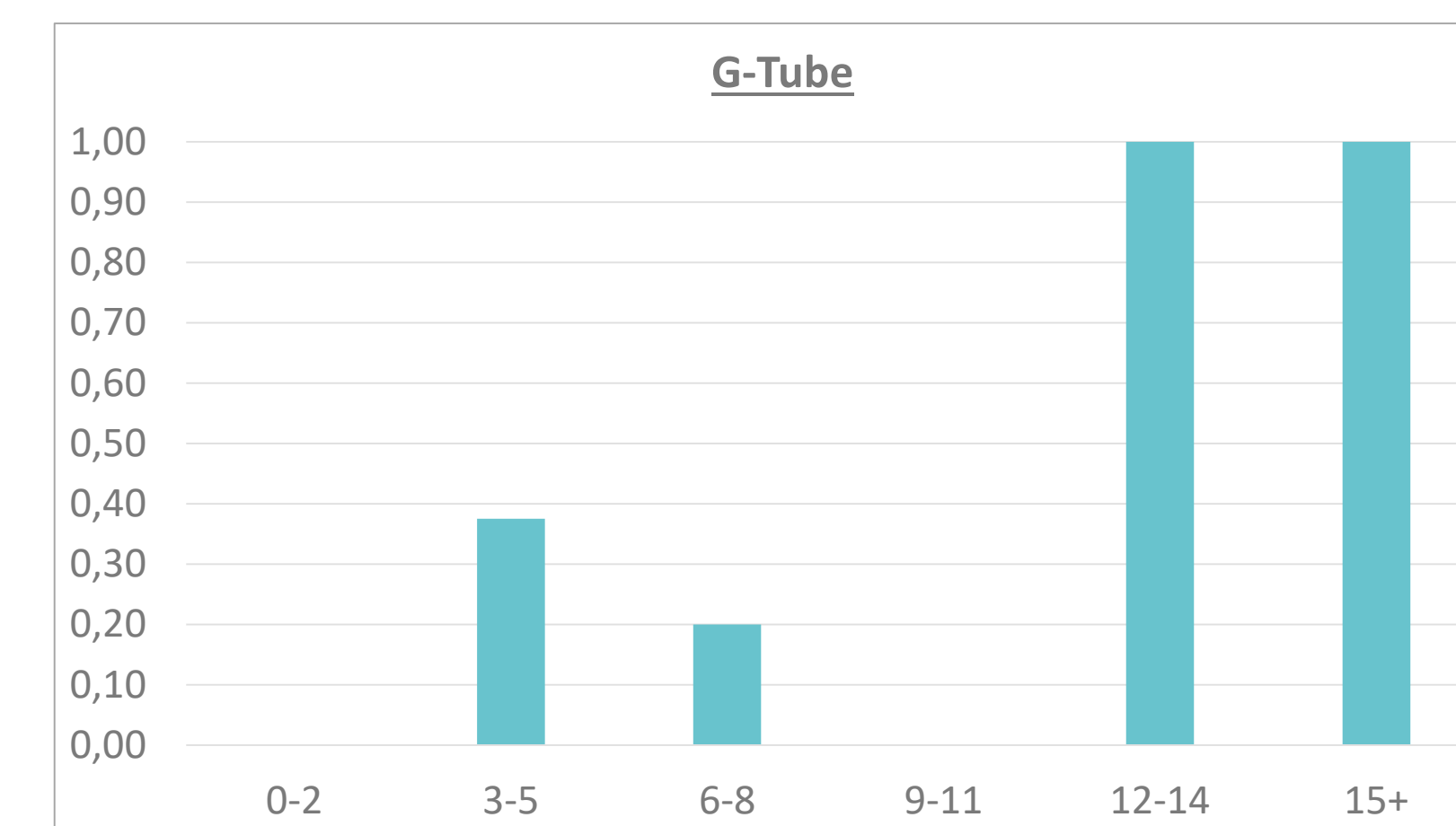


Chart 4. Percentage of patients treated with a G-tube

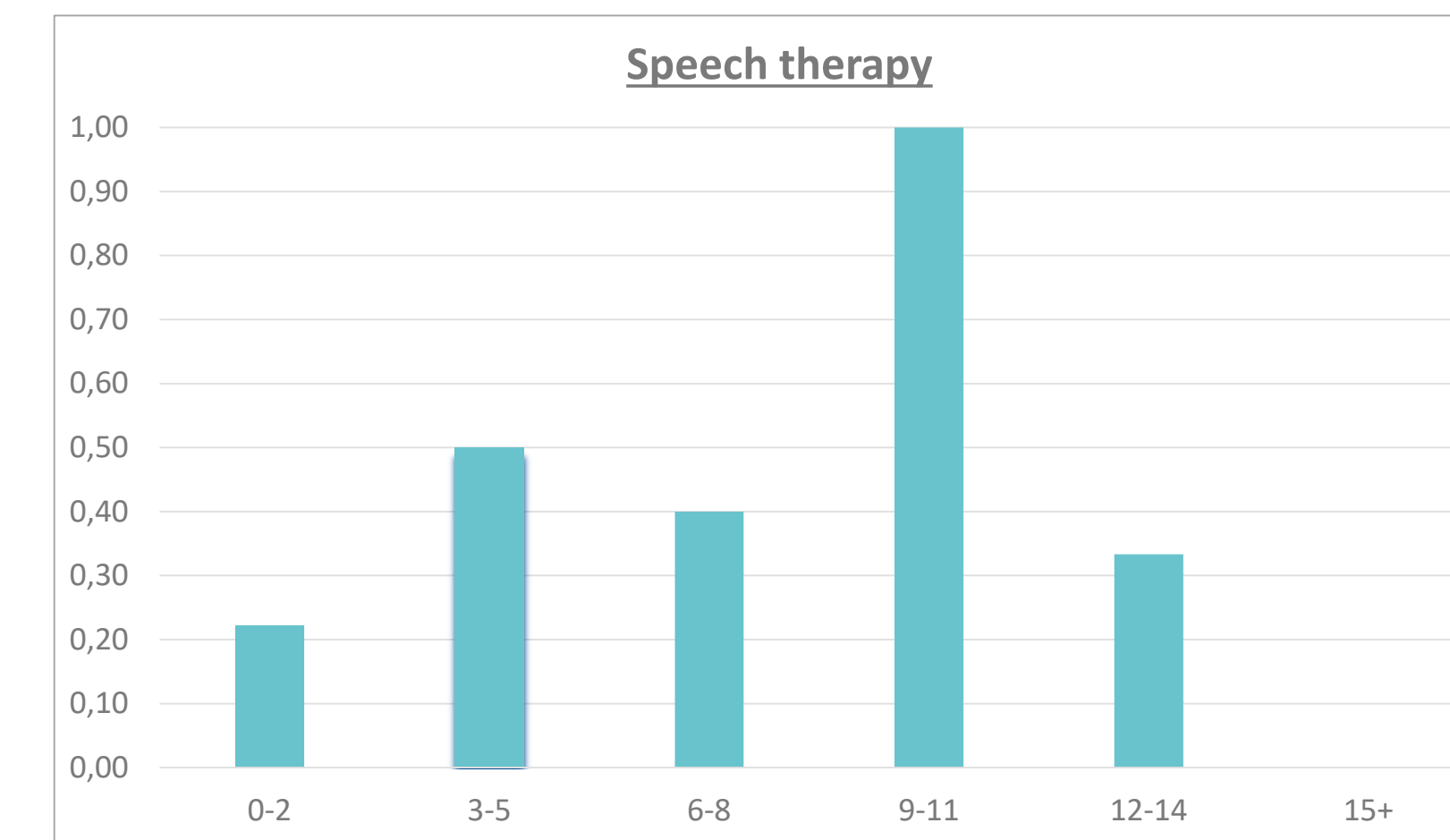


Chart 5. Percentage receiving speech therapy (which is also focused on swallowing)

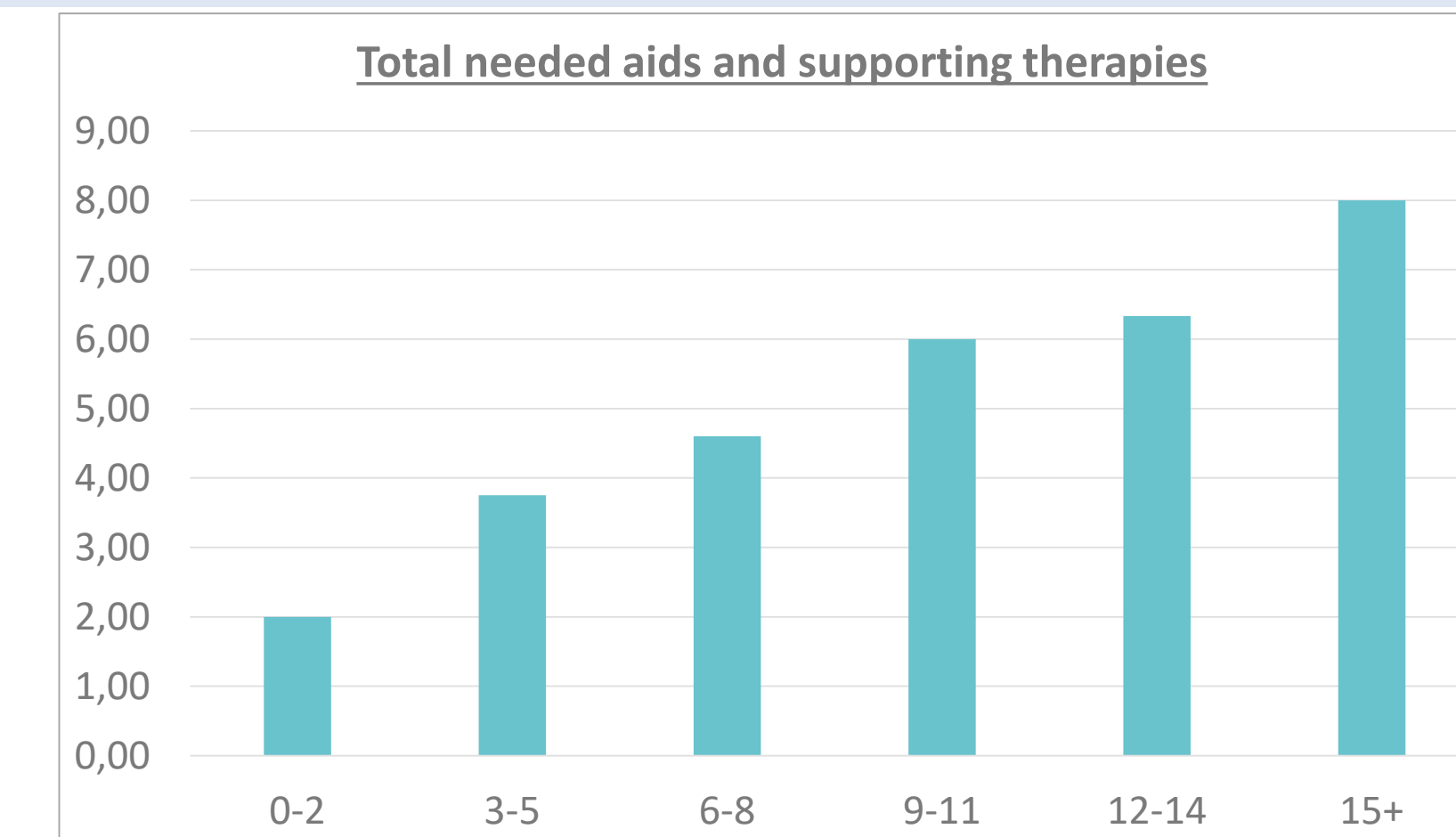


Chart 6. Total number of necessary aids and supportive therapies

## Discussion & Conclusion

CD patients have a high demand of medical aids and support. The need of physical therapy, orthoses, wheelchairs, support for feeding and the number of medical aids increases with age. This reflects the progressive nature of the course of the disease with comprehensive deficits leading to necessity of multidisciplinary collaboration of healthcare professionals and lifelong support.

## Literature

(1) The natural history of **Canavan** disease: 23 new cases and comparison with patients from literature. Bley A, Denecke J, Kohlschütter A, Schön G, Hischke S, Guder P, Bierhals T, Lau H, Hempel M, Eichler FS. Orphanet J Rare Dis. 2021 May 19;16(1):227. doi: 10.1186/s13023-020-01659-3. PMID: 34011350