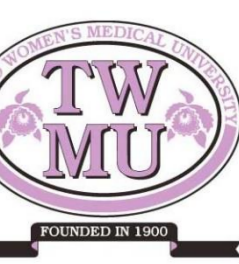


Nationwide Patient Registry of Fukuyama Congenital Muscular Dystrophy in Japan



*Keiko ISHIGAKI (a), Chikoto IHARA (b), Terumi MURAKAMI (a), Takatoshi SATO (a), Kumiko ISHIGURO (a), Minobu SHICHIJI (a), Hisanobu KAIYA (b), Makiko OSAWA (a)(b) and Satoru NAGATA (a)

(a) Department of Pediatrics, Tokyo Women's Medical University, School of Medicine, Tokyo, Japan
(b) The Japan Muscular Dystrophy Association, Tokyo, Japan

Background

- Hallmarks of Fukuyama congenital muscular dystrophy (FCMD), the second most common muscular dystrophy in the Japanese population, include severe muscular dystrophy and dysgenesis of the central nervous system.
- Most patients with typical FCMD die of respiratory dysfunction, pulmonary infections including aspiration pneumonia, suffocation and congestive heart failure around age 20 years.
- In 2013, the Japan Muscular Dystrophy Association established a registry of FCMD patients in Japan to facilitate patient recruitment for clinical trials and to provide information on the natural history of FCMD. We retrospectively analyzed this nationwide FCMD registry database.

Methods

From October 2011 through September 2013, 207 Japanese FCMD patients (104 boys and 103 girls) in total were registered. Data were summarized using descriptive statistics, including the mean, standard deviation (SD), median, range, frequency, and percentage.

Results

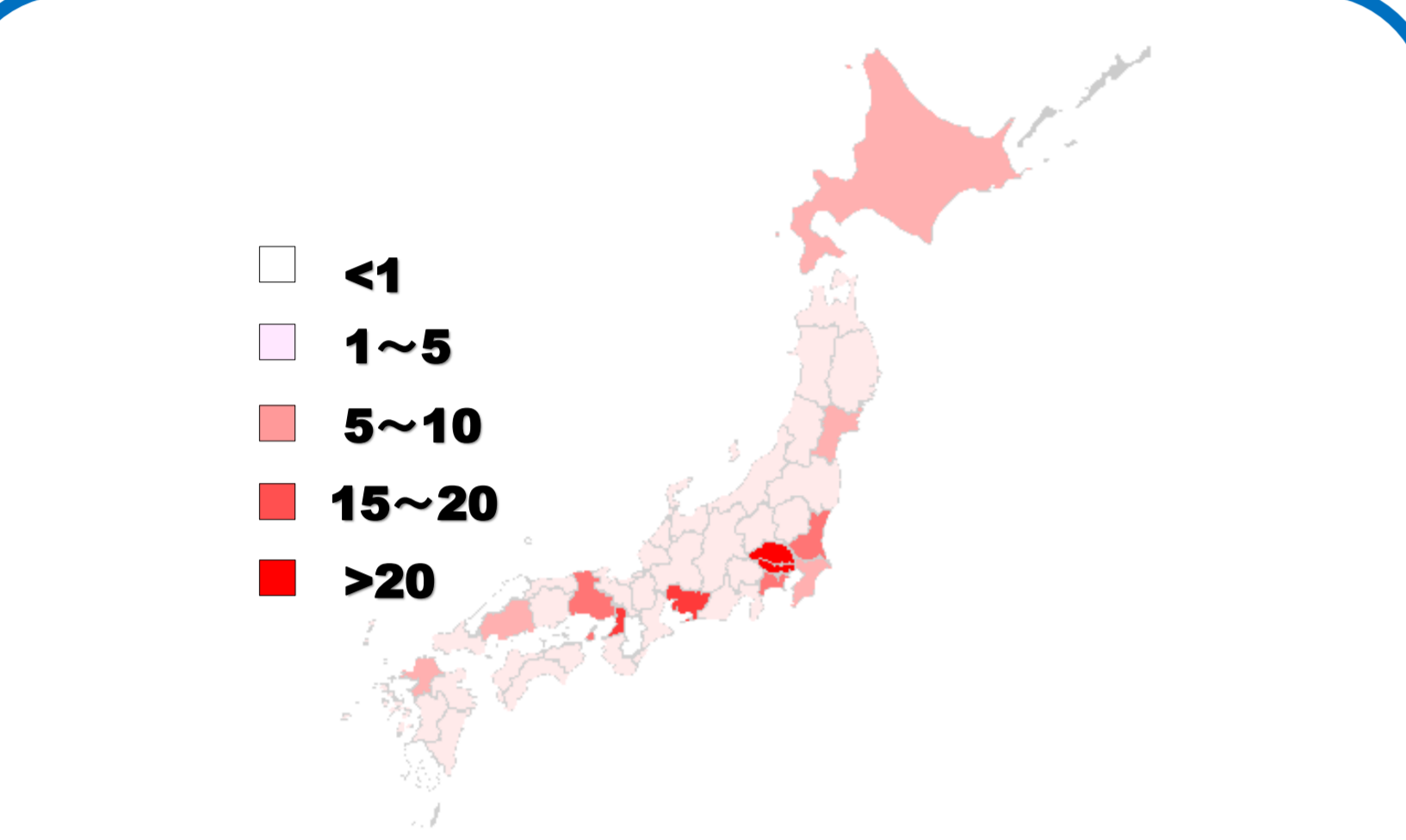


Figure 1: Participant distribution
207 Japanese FCMD patients (104 boys and 103 girls) in total from throughout Japan (41/47 prefectures) were registered.

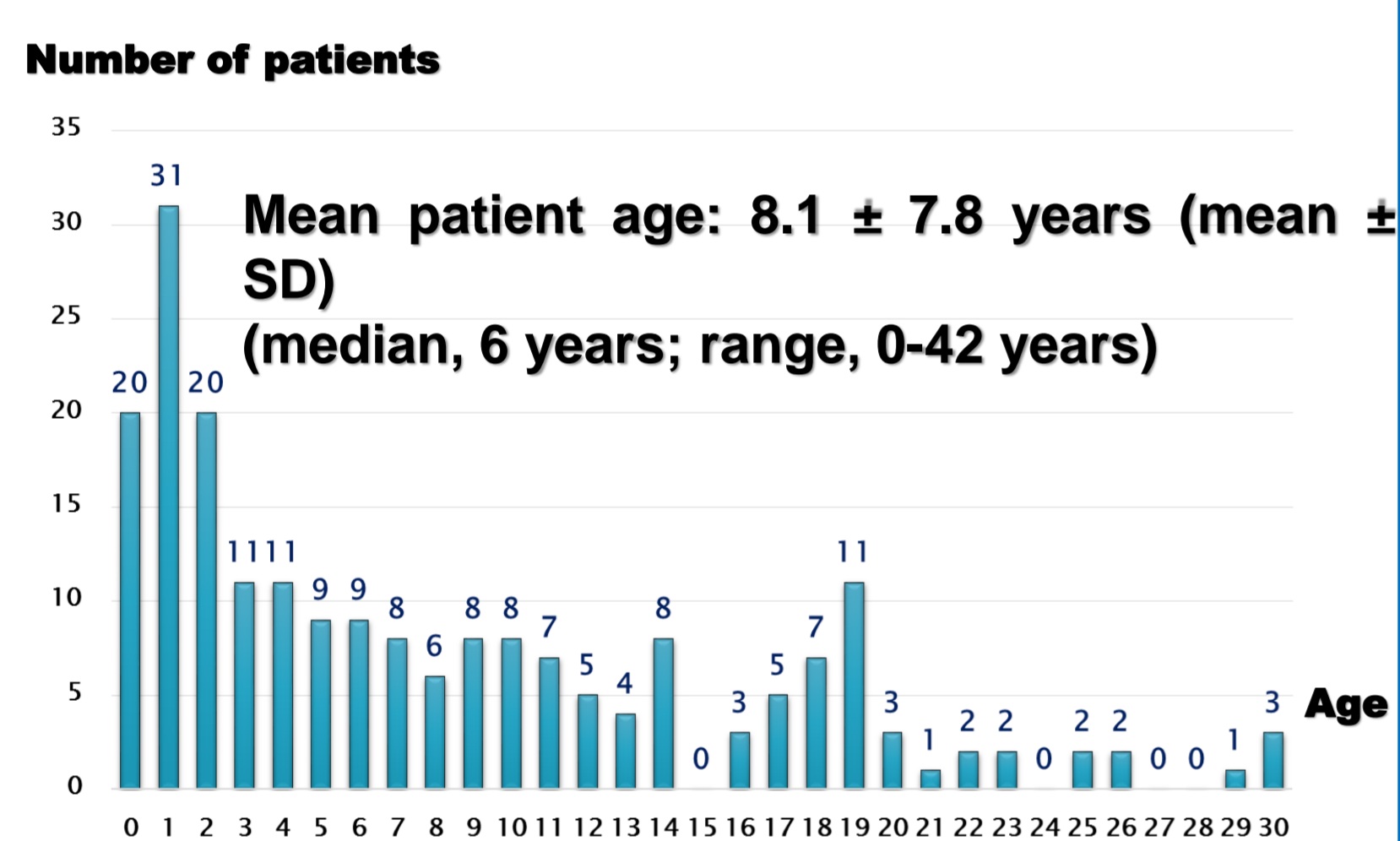


Figure 2: Age distribution of participants
Young patients (under age 5 years) accounted for nearly half of the registrants.

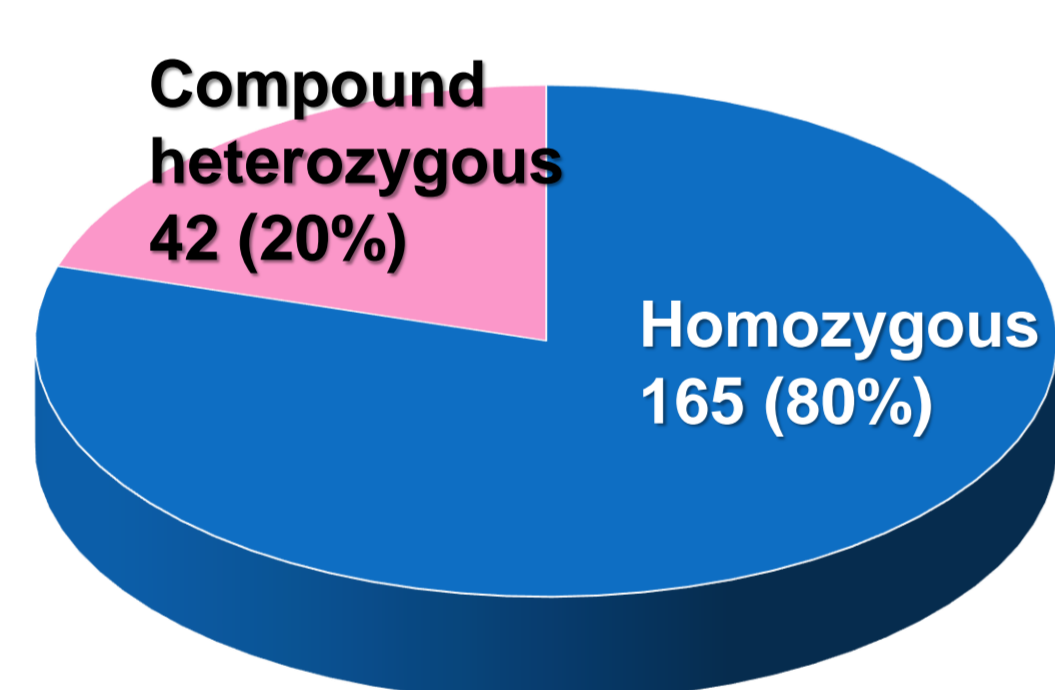


Figure 3: FKTN gene mutation
A homozygous founder 3-kb insertion mutation in the FKTN gene was present in 80% of registrants, while 20% had a compound heterozygous mutation.

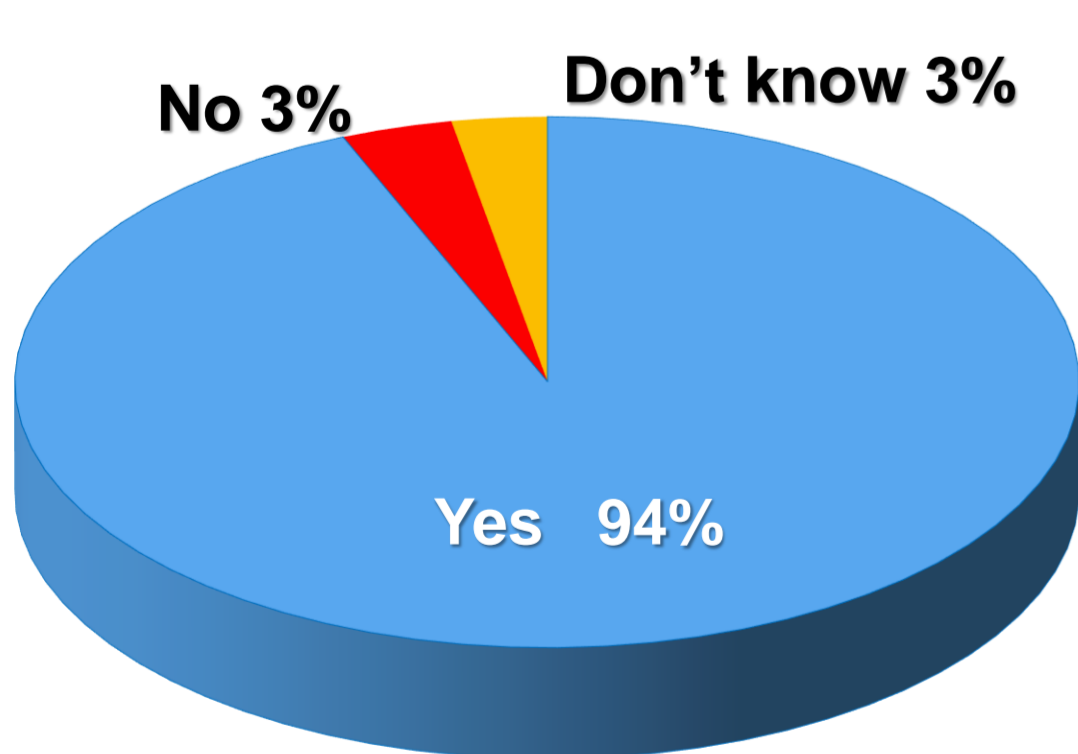


Figure 4: Interest in clinical trial information
Whether the family wanted to obtain information about trials.

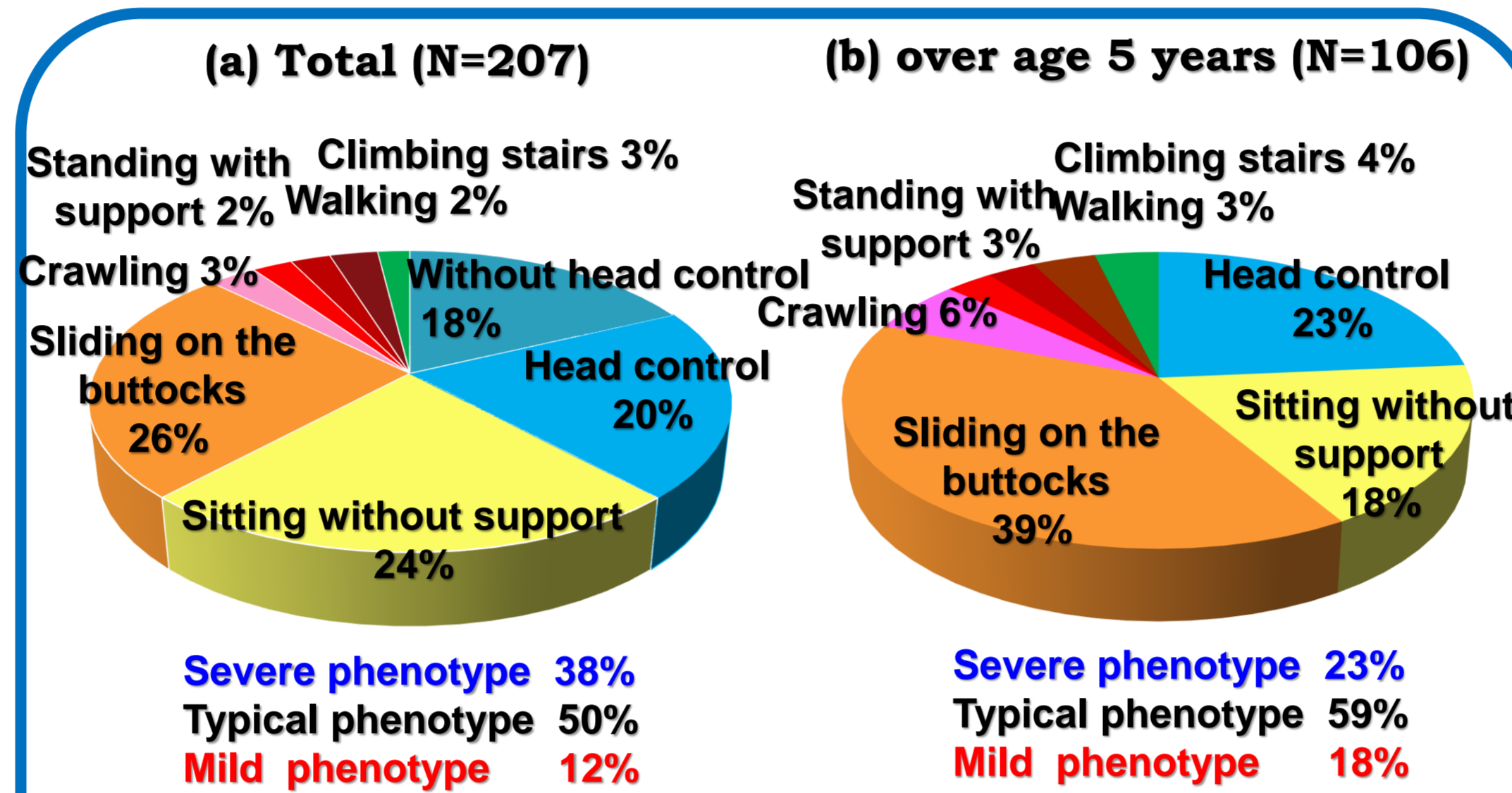


Figure 5: Maximum motor development

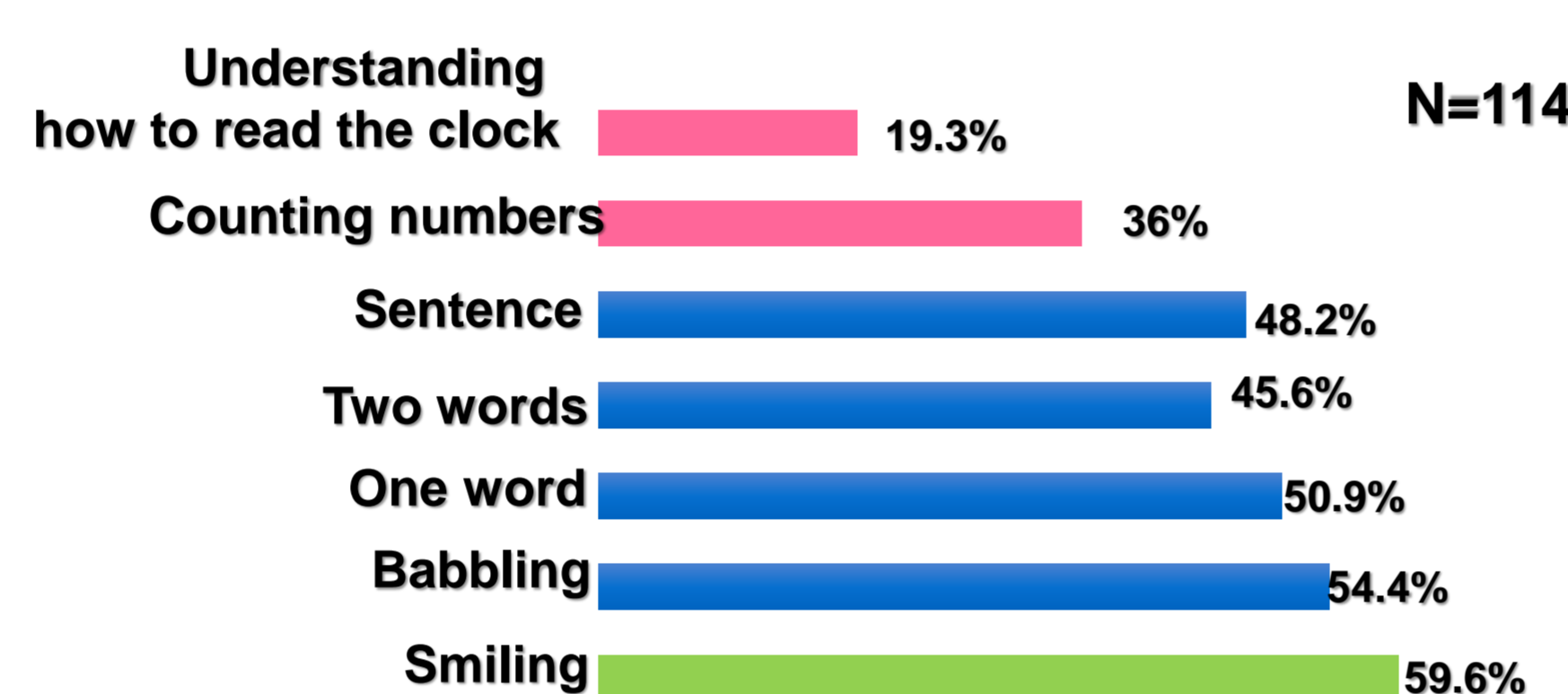


Figure 6: Maximum intellectual development (over age 5 years)
Nearly half of patients acquired the ability to speak two words after age 5 years.

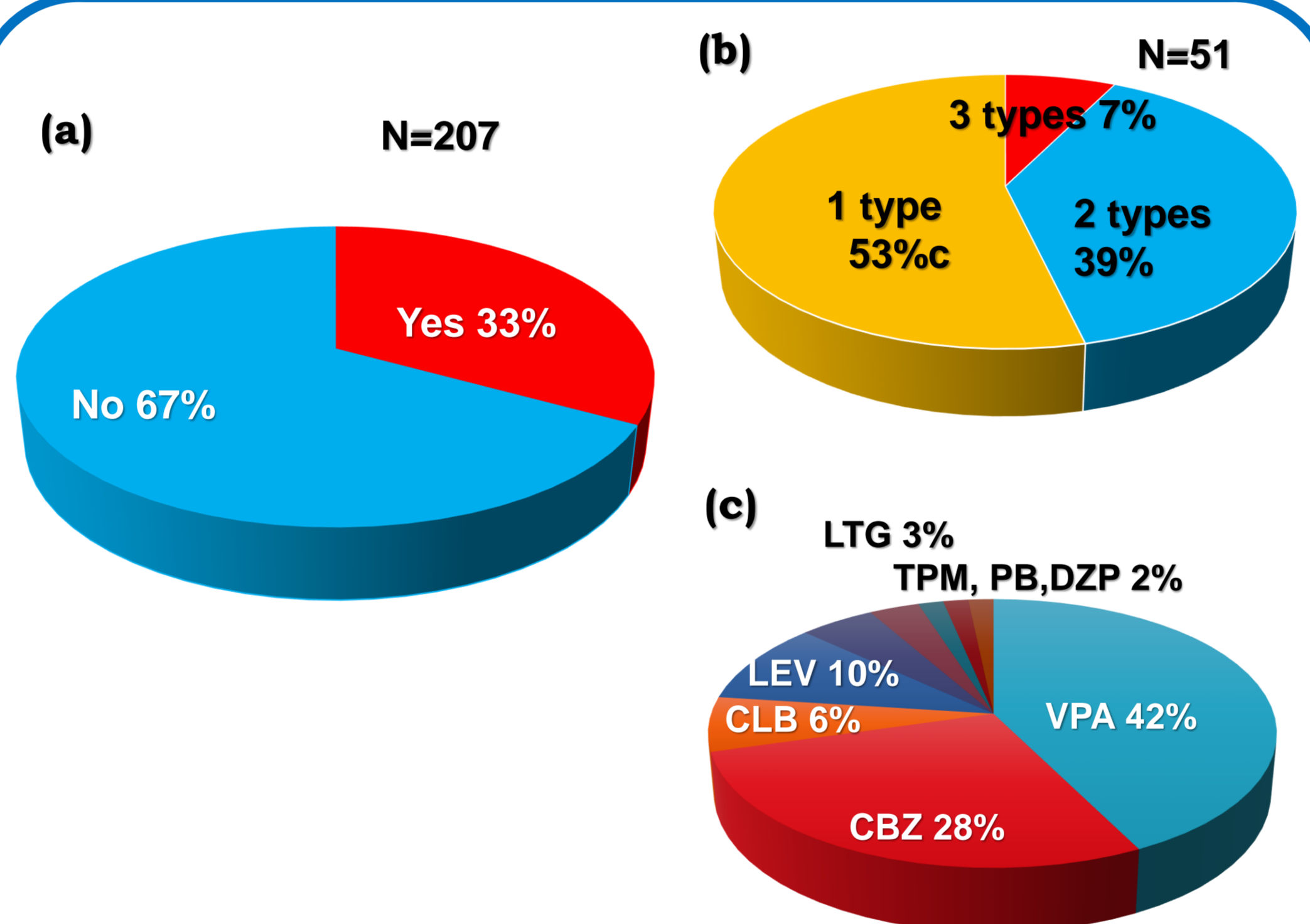


Figure 7(a): Seizures at present
Sixty-nine patients (33%) suffered febrile seizures and/or epilepsy.
(b) The number of anti-epileptic agents
(c) The agents frequently used

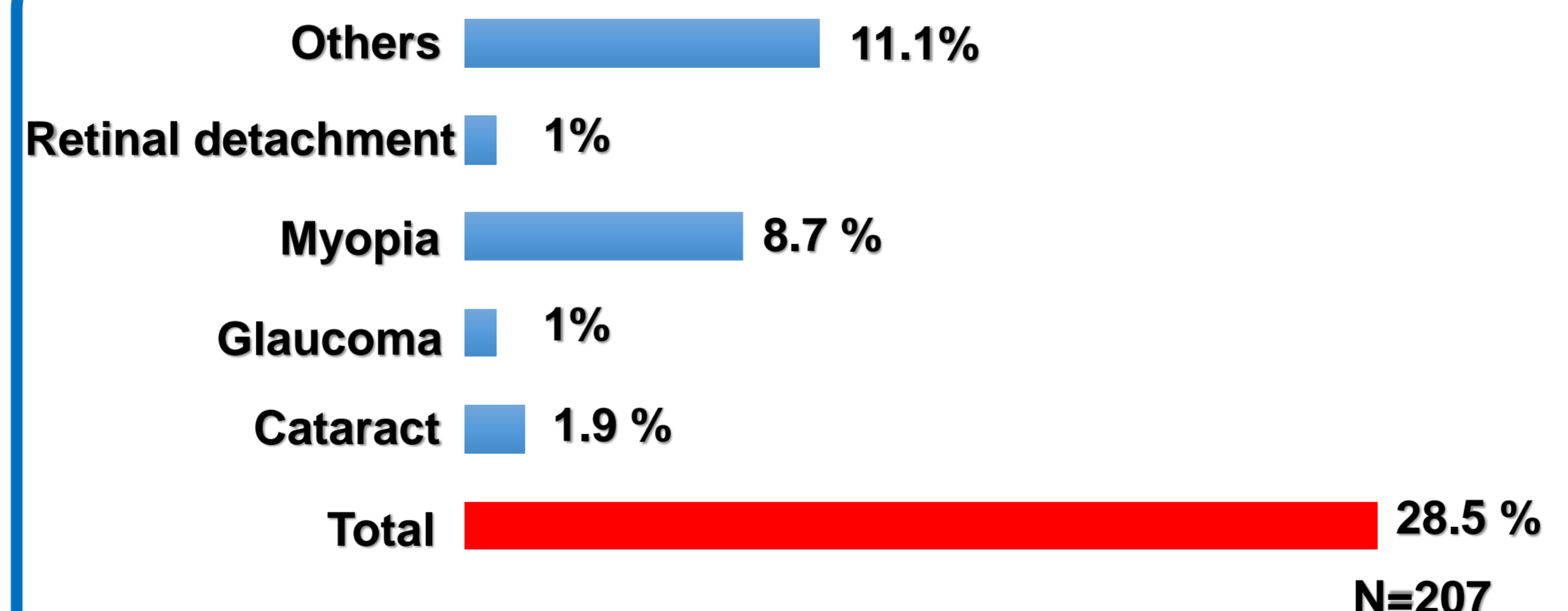


Figure 8: Incidence of ocular abnormalities
Myopia had the highest prevalence (18 patients; 8.7%), followed by strabismus (5.9%).

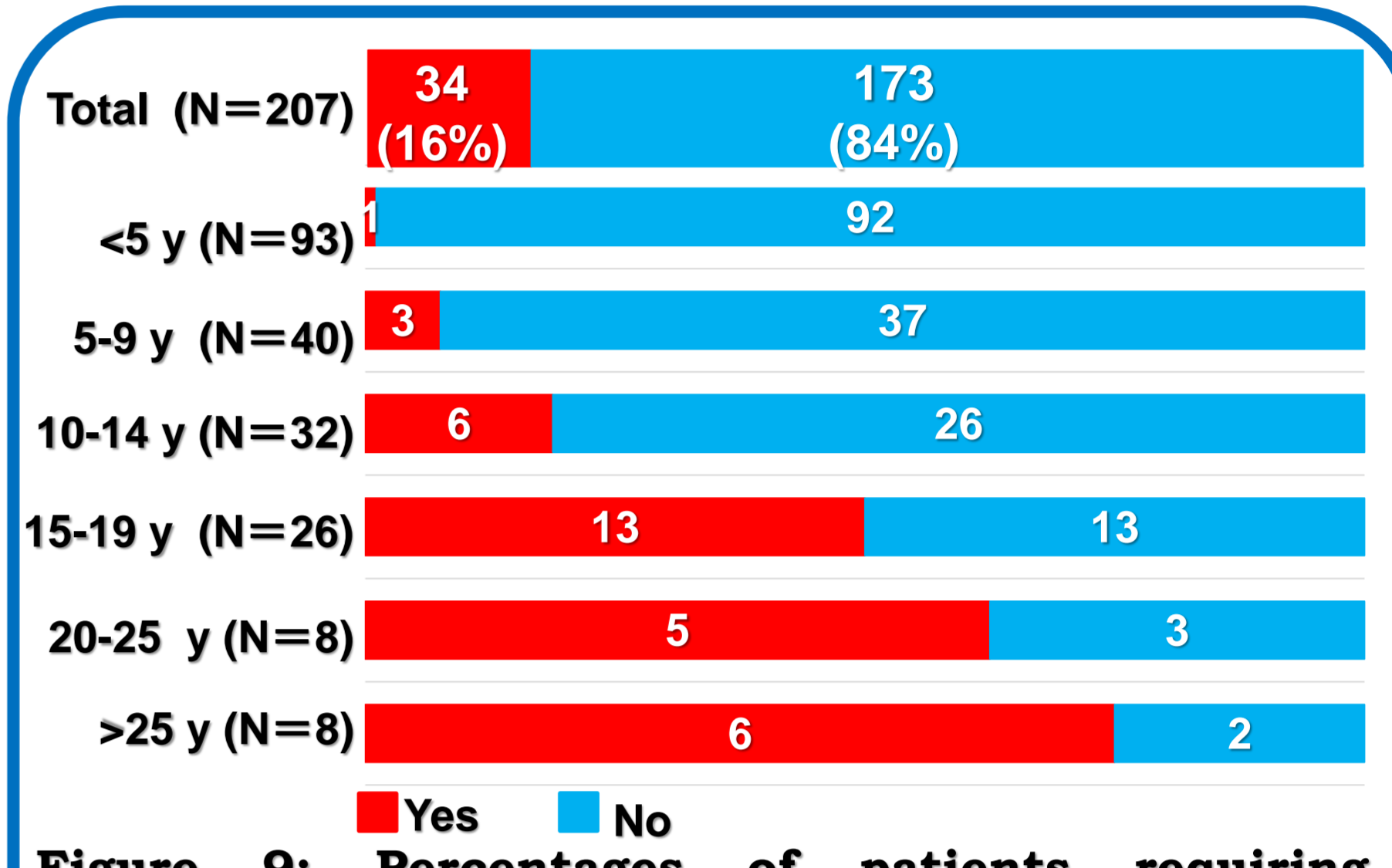


Figure 9: Percentages of patients requiring respiratory support, by age group
Thirty-five patients (17%) required respiratory support. After age 15 years, the percentage of patients requiring respiratory support increased with age.

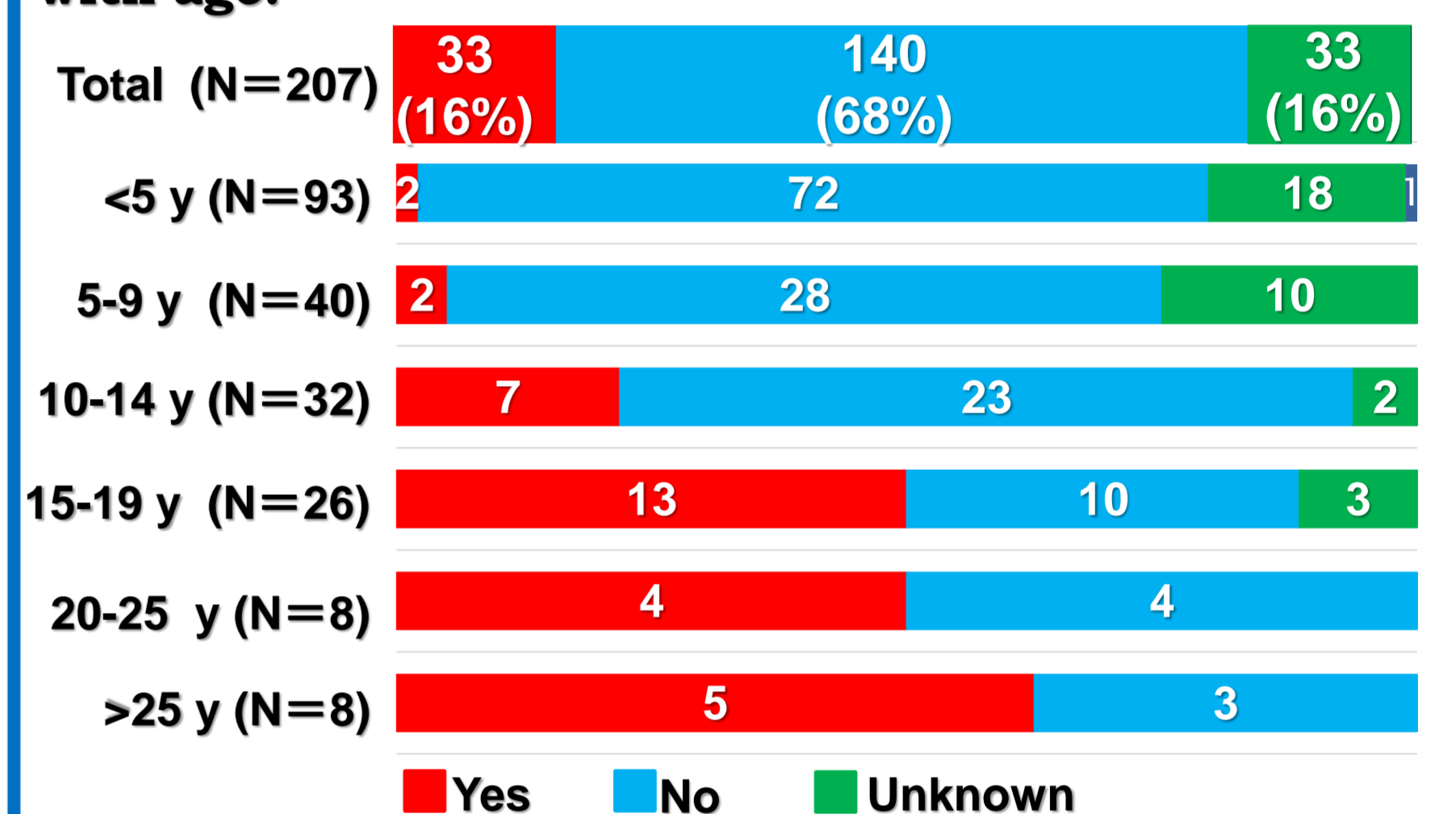


Figure 10: Percentage of patients with cardiac dysfunction by age group
Cardiac dysfunction was detected in 33 patients (16%). Beyond age 15 years, 50% of patients showed cardiac dysfunction but this percentage did not change in the older groups.

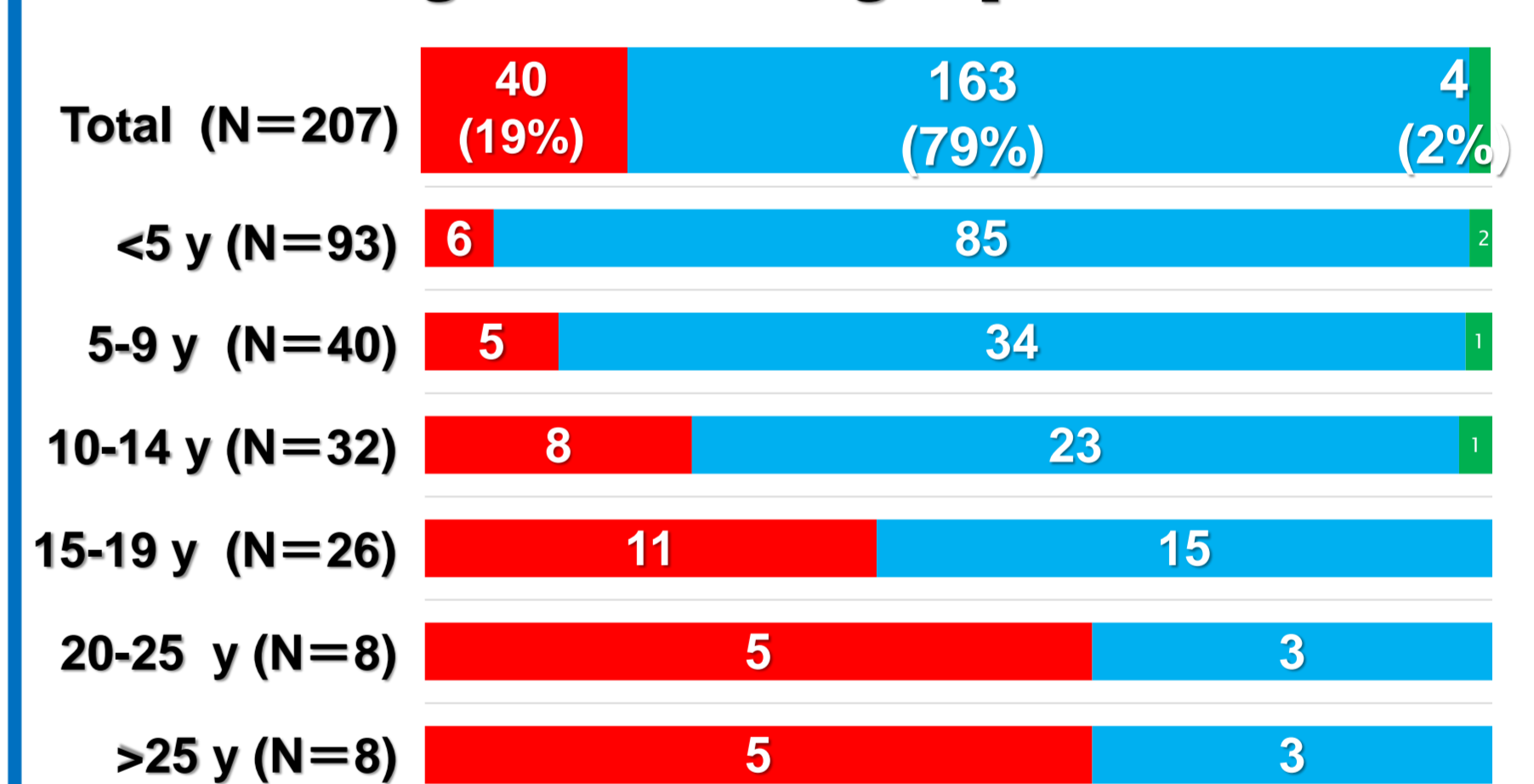


Figure 11(a): Percentage of patients with dysphagia

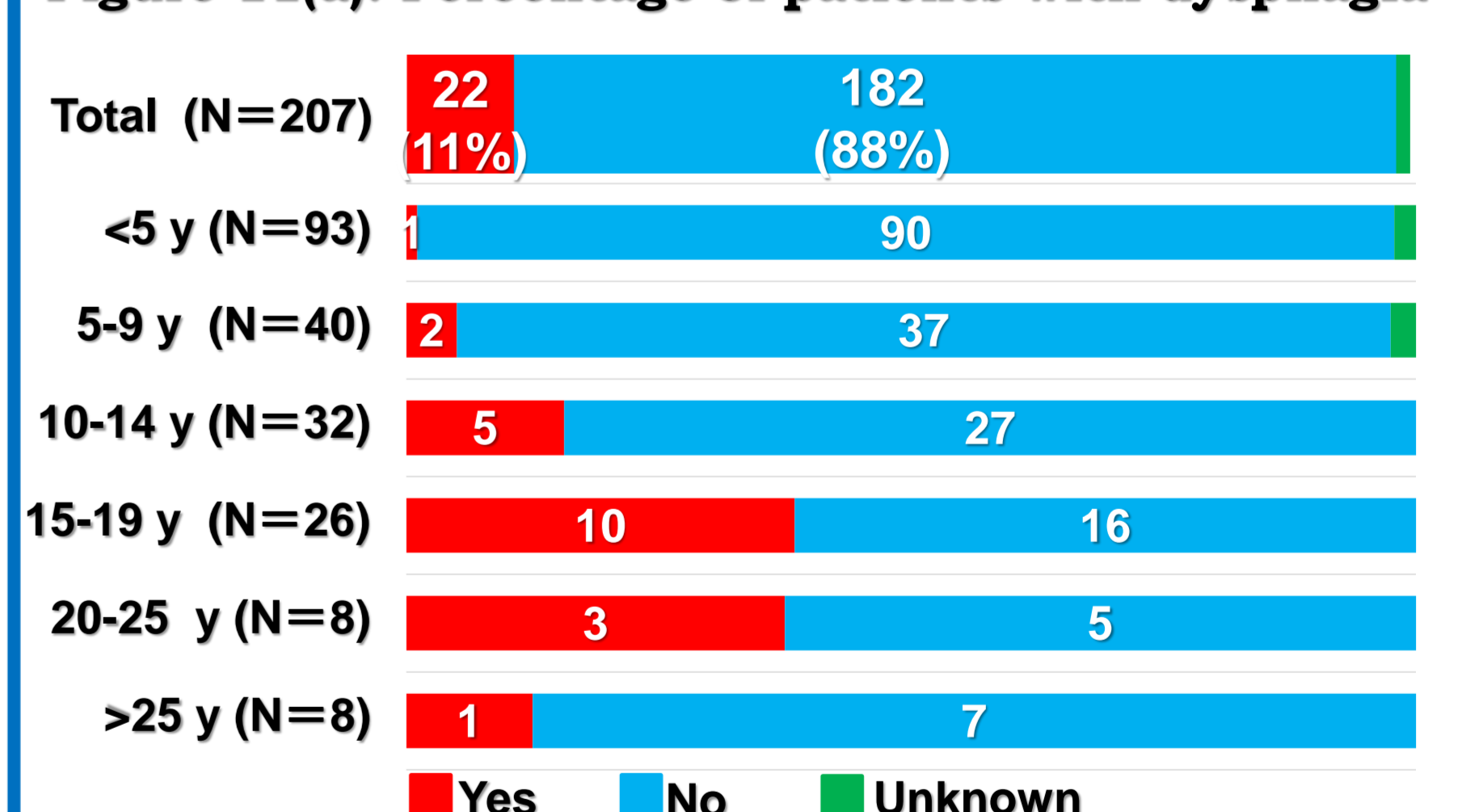


Figure 11(b): Percentage of patients with gastrostomy
By age 15 years, nearly 40% of patients had undergone gastrostomy, though some advanced-stage patients were still able to ingest food orally even beyond 25 years of age.

Discussion

- In FCMD patients, respiratory dysfunction and congestive heart failure are life-threatening complications that generally appear around 20 years of age. We found that after age 15 years, the percentage of patients requiring respiratory support increased with age, while that with cardiac dysfunction did not change in the older.
- In FCMD patients, dysphagia emerged earlier than respiratory dysfunction and necessitated active intervention. However, this study revealed that only 40% of patients underwent gastrostomy, even after age 15 years. Moreover, even some of the advanced-stage patients were able to ingest food orally beyond 25 years of age.
- The FCMD patient registry is useful for clarifying the natural history of this disease and recruiting patients for clinical trials. However, the age profiles of participants were relatively young and an information campaign designed to highlight the importance of the registry for older patients is needed.

Conclusion

The FCMD patient registry is useful for clarifying the natural history of this disease, but we need an information campaign aimed at highlighting the importance of the registry for older patients.

- Conflict of Interest (COI) of the Principal presenter : No potential COI to disclose.
- Conflict of Interest (COI) of the Co-presenters : No potential COI to disclose.