**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), 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:** FRTK gene mutation. A homozygous founder 3-kb insertion mutation in the FRTK 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. Understanding how to read the clock: 19.8%. Counting numbers: 36%. Two words: 45%. One word: 14%. Babbling: 34%. Severe phenotypes: 36%. Moderate phenotypes: 12%. Typical phenotypes: 20%. Mild phenotypes: 16%.

- **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:** Disabilities at present. Sixty-nine patients (33%) suffered febrile seizures and/or epilepsy. The number of anti-epileptic agents The agents frequently used.

- **Figure 8:** Incidence of ocular abnormalities. Myopia had the highest prevalence (18 patients; 8.7%), followed by strabismus (8.8%).

- **Figure 9:** Percentages of patients requiring respiratory support, by age group. Thirty-five patients (17%) required respiratory support. After age 18 years, the percentage of patients requiring respiratory support increased with age.

- **Figure 10:** Percentages of patients with cardiac dysfunction by age group. Cardiac dysfunction was detected in 33 patients (16%). Beyond age 15 years, 80% of patients showed cardiac dysfunction but this percentage did not change in the older groups.

- **Figure 11(a):** Percentages of patients with dysphagia. Over 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 group.

- **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 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.