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BACKGROUND

- Spinal muscular atrophy (SMA) is an ultra rare genetic disorder characterized by the progressive systemic degeneration of muscle tissues, leading to **gradually worsening muscle atrophy symptoms**^{1,2}.
- The purpose of this study is to create an HRQoL (Health-Related Quality of Life) for SMA, based on patients who have a thorough understanding of the condition, prior to developing vignettes related to health status that will be administered to the general population in future research.

METHODS

Vignettes

| | | | | |
|--|---|--------------------------|---------------------------|---|
| | | | | |
| Vignette 1 | Vignette 2 | Vignette 3 | Vignette 4 | Vignette 5 |
| Inability to sit and requires permanent respirator | Inability to sit and requires breathing support | Able to sit with support | Able to walk with support | Able to stand and walk with-out support |

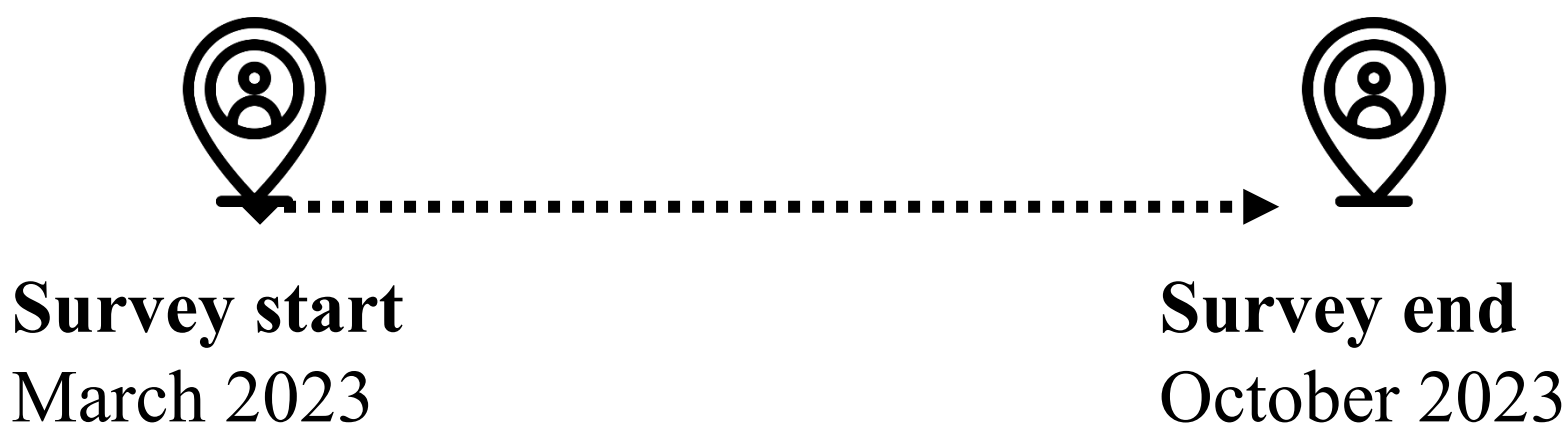
- Five vignettes, developed through research and clinical consultations, were categorized based on increasing severity of the disease and considering the use of respiratory assistance and the level of mobility.

Study Design

- The final vignette was employed to generate weights for HRQoL using European Quality of Life Group's 5-Domain Questionnaires 5 Levels (EQ-5D-5L) self and/or proxy-reported version³.
- The ultimate vignette was conducted in face-to-face sessions with all types of SMA patients who visited Gangnam Severance Hospital in Seoul, South Korea from March 21 to November 20, 2023.**

Ethnics

- All study participants provided informed consent and the study documents and protocols have been approved by an Institute of Review Board of Gangnam Severance Hospital (IRB No. 3-2022-0480).



RESULTS

Patient demographics

- A total of 36 participants responded to the survey. 27 patients responded through self-reported form, while nine caregivers of SMA assumed the patient's perspective and responded through proxy-reported form (**Table 1**).
- The patients for whom caregivers responded are either under eight years of age or unable to self-report due to factors such as permanent ventilator use.

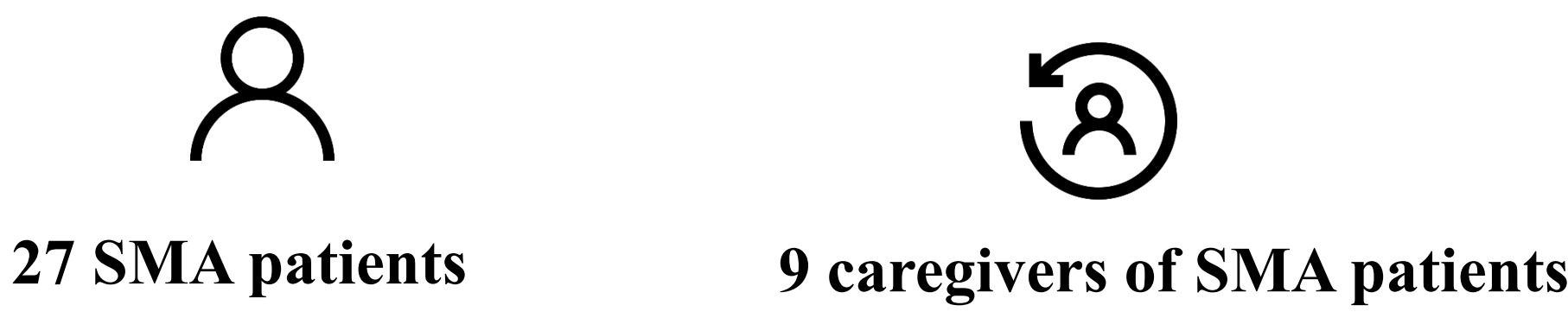
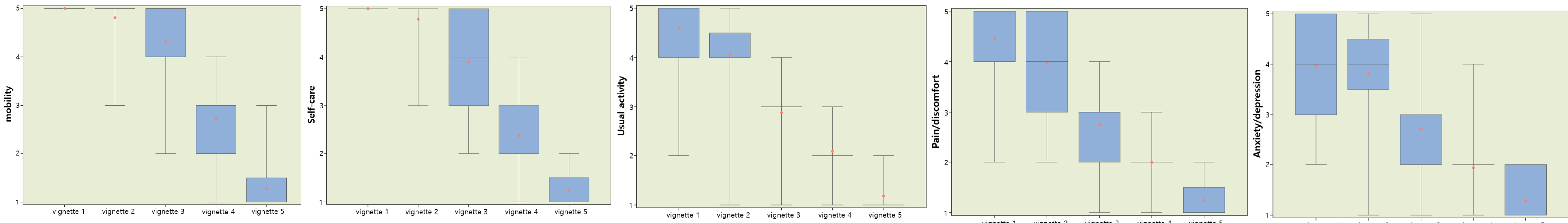


Figure 1. Health-related quality of life by domain across vignettes.



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Table 1. Sociodemographic information related to patients with SMA represented by study participants.

| | | SMA patients (n=36) | | |
|-----------------------|---------------------------------------|------------------------|----------|-----|
| Age | Current age (mean, std) | 22.2±9.9 | | |
| | Under 10 years old | 4 | 11% | |
| | 10-19 years old | 11 | 31% | |
| | 20-29 years old | 12 | 33% | |
| | Those aged 30 and above | 9 | 25% | |
| Sex | Monthage at SMA diagnosis (mean, std) | | | |
| | Male | 16 | 44% | |
| | Female | 20 | 56% | |
| Insurance | National Health Insurance | 30 | 90% | |
| | Medicaid | 4 | 10% | |
| Current disease state | Ventilated, permanent | 5 | 14% | |
| | Ventilated, intermittent | 7 | 19% | |
| | Not ventilated | 24 | 67% | |
| Current best function | Not able to sit | 10 | 28% | |
| | Able to sit with help | 23 | 64% | |
| | Able to stand with help | 1 | 3% | |
| Caregiver | Primary caregiver's age (mean, std) | | 52.7±7.9 | |
| | Primary caregiver's sex | Male | 4 | 11% |
| | | Female | 32 | 89% |
| | Primary caregiver's education level | High school | 10 | 28% |
| | | University | 26 | 72% |
| Family history | Yes | 8 | 22% | |
| | No | 28 | 78% | |

Health-related Quality of Life by domains

- The HRQoL was defined in terms of five domains: mobility, self-care, usual activity, anxiety/depression, and pain/discomfort. The five domains were scored with 5 representing the most severe state and 1 denoting the mildest stage (**Figure 1**)⁴.
- Sensitivity to disease progression was observed in all dimension.

Health-related Quality of Life

- The utility weights for EQ-5D-5L data were derived using the Korean population EQ-5D-5L tariff.⁵
- The utility weights for each disease progression state were as follows:
 - Inability to sit and requires permanent respirator (**0.022**)
 - Inability to sit and requires breathing support (**0.132**)
 - Able to sit with support (**0.411**)
 - Able to stand with assistance (**0.685**)
 - Able to stand and walk without assistance (**0.911**)

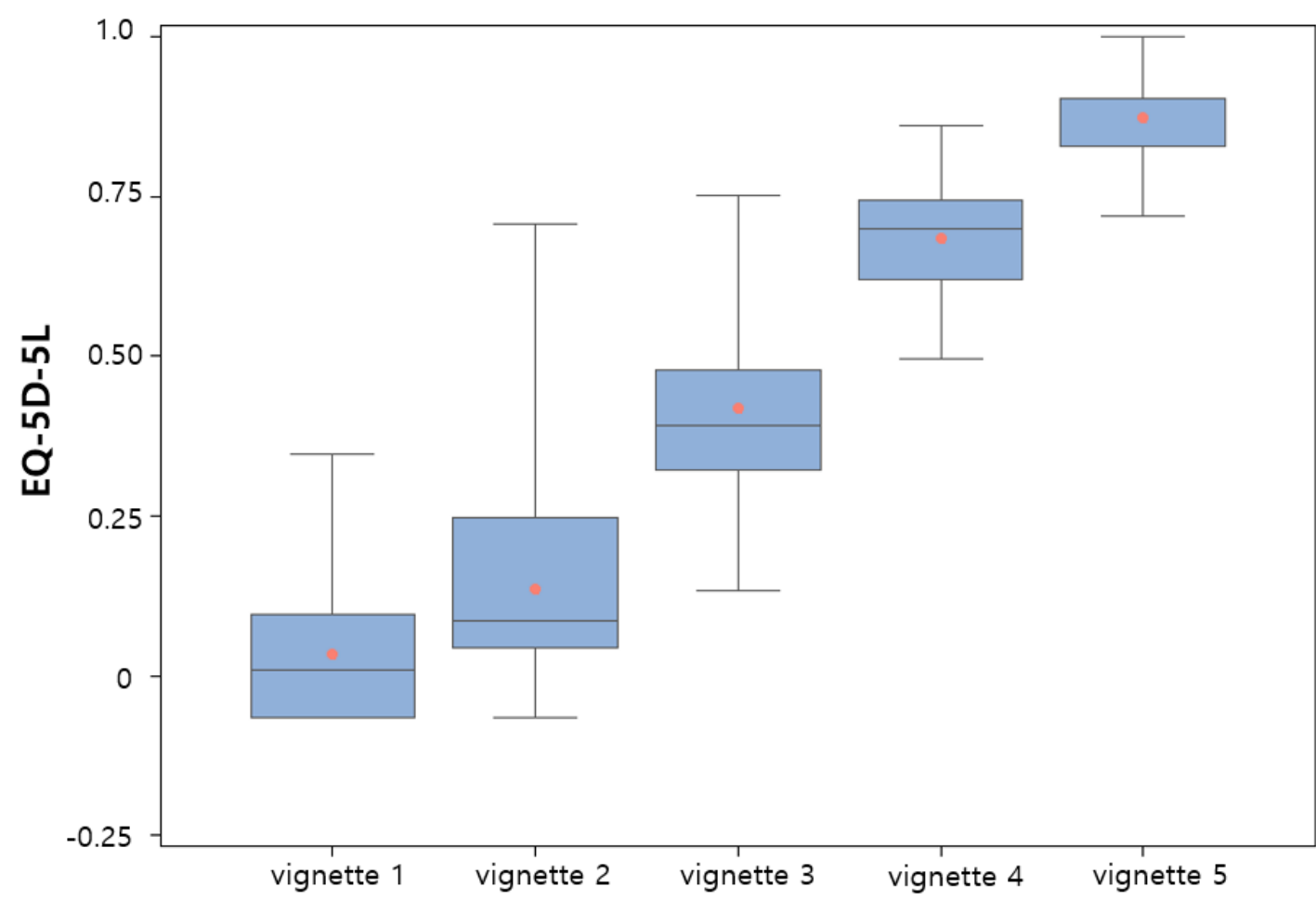


Figure 2. Health-related quality of life of patients with SMA measured by EQ-5D-5L.

CONCLUSION

- This study represents the first demonstration of HRQoL in SMA patients in Korea, through five health status vignettes explaining the disease progression.
- It was found that as the severity of SMA worsened, the quality of life significantly decreased, impacting HRQoL in all domains.

ACKNOWLEDGEMENT

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