EE406

DEVELOPMENT OF SCENARIOS BY HEALTH STATE TO MEASURE THE QUALITY OF LIFE OF SPINAL MUSCULAR ATROPHY IN THE GENERAL POPULATION

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OBJECTIVES

Measuring quality of life (QoL) in the general population (GP) without a specific disease is essential for economic evaluation because it can objectify the disease burden. is increasing to estimate the burden of disease and objectify the value of innovative treatment. Since **ultra-rare diseases like spinal muscular atrophy (SMA)** are very unfamiliar to GP, it is difficult to fully understand the condition of the disease and accurately measure the QoL. Therefore, **we aimed to develop scenarios** by reflecting the natural history of disease **to help GP understand patients' condition and measure QoL** by assuming they are in the states.

METHODS

We investigated the **characteristics and prognosis of patients with SMA** and reviewed the literature and disease-specific measures of QoL. A draft scenario describing the condition of SMA patients was developed, and the draft was modified by requesting the clinical advisory group (CAG) for the first review. The second draft was reviewed by the survey expert, and then the final scenario was completed with a second review by CAG.

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RESULTS

Health states of SMA were classified into five states according to the movement ability and respiratory function: **(a) Inability to sit and requires permanent respirator (b) Inability to sit and requires breathing support (c) Able to sit with support (d) Able to walk with support (e) Able to stand and walk without support.** In the worst health scenario (a) includes a description of general weakness, risk of morbidity and mortality from pneumonia, ways of feeding, and minimal requirements of ventilator. A slightly improved scenario (b) includes general weakness in head control, swallowing and eating, 2-year mortality, and breathing related to respiratory muscle weakness. State (c), which represents more improved state, is composed of the possibility of self-moving without a wheelchair, issue related to scoliosis, general weakness, and life expectancy. State (d) includes a description such as daily life availability, progressive muscle weakness, and rates of obesity and osteoporosis. For best condition (e), the degree of interference with daily life, disease progression, life expectancy are described. Every scenario includes images to help understand.

CONCLUSIONS

Scenarios developed to measure the QoL of SMA patients can be applicable when measuring the disease burden of SMA patients in the general population.

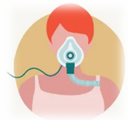
KEYWORLDS

Spinal muscular atrophy, Ultra-rare disease, Quality of life, Scenarios

SCENARIOS

- Images from Flaticon(https://www.flaticon.com/kr/)

(a) Inability to sit and requires permanent respirator



- Requiring artificial ventilation for more than 16 hours a day.
- High risk of morbidity and death from aspiration pneumonia.
- Nutrition is supplied through a feeding tube.
- Unable to move without assistance.

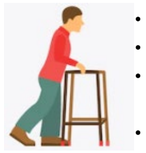
(b)Inability to sit and requires breathing support

- Requiring artificial ventilation for less than 8 hours a day.
- Difficult to hold head, swallow, and breathe due to severe muscle weakness
- Difficulty swallowing saliva or eating milk.
- Without treatment, 90% die within 2 years

(c)Able to sit with support

- Able to hold head, swallow, and breathe independently.
- Able to sit or stand without assistance but require a wheelchair for movement.
- Limited joint movement including spasms in their fingertips
- Average lifespan is until early childhood, with a survival rate of 70% at 25 years old.

(d)Able to walk with support



- Can survive until adulthood and lead a normal daily life.
- Assistance from others may be needed for movement.
- Progressive muscle weakness may occur and loss of strength in the limbs.
- High risk of obesity and osteoporosis who have muscle weakness.

(e)Able to stand and walk without support

- Complications such as scoliosis, metabolic acidosis, and stiff joints may occur.
- No interference in daily life, slow disease progression and same life expectancy with general population.