Measuring Quality of like in patients with SBMA (Kennedy's disease)

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Objective

Spinal and bulbar muscular atrophy (SBMA) is a rare, late onset, X-linked neuromuscular disease characterized by slowly progressive limbs and bulbar muscle weakness and atrophy associated with wide systemic involvement. Nowadays, there is no effective treatment for SBMA and health-related Quality of life (QoL) is an important issue in this chronically progressing disease.

Aim of this study was to assess QoL in a large cohort of SBMA patients using both generic and disease specific questionnaires.

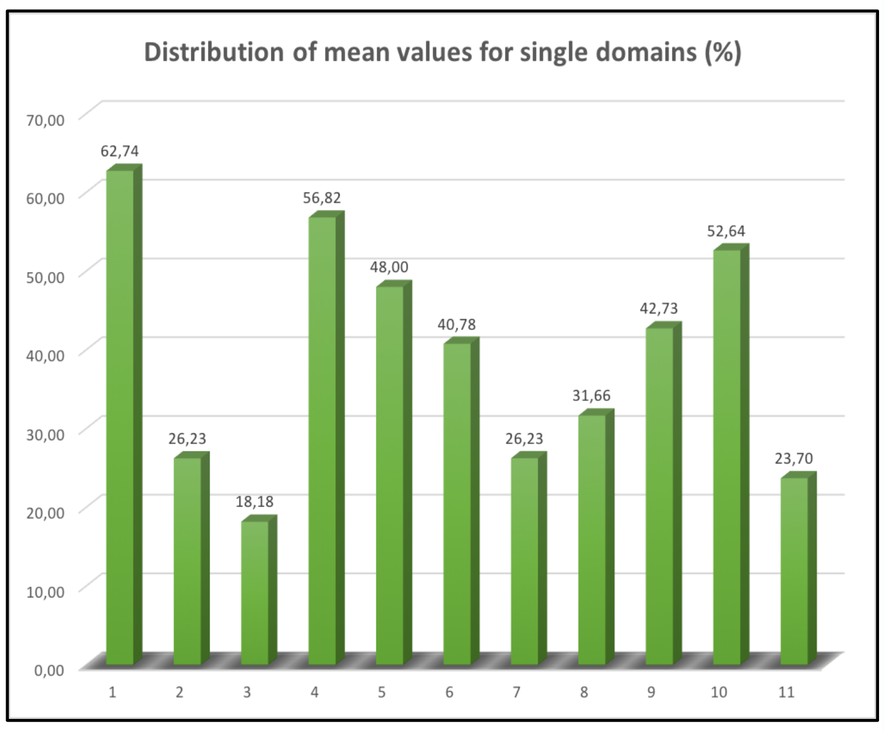
Methods

- **55 genetically confirmed SBMA patients were considered.**
- They underwent medical history collection and functional status evaluation through SBMAFRS, 6MWT and AMAT scales.
- All the patients underwent neuropsychological examination to exclude the presence of cognitive impairment.
- To describe QoL, individualized neuromuscular QoL questionnaire **(INQoL)** was administered. The scale is made by a total score composed by 10 subscores (weakness, locking, pain, fatigue, activities, independence, social relationship, emotions, body image and treatment).
- The presence of anxiety and depression was test using the **HADS scale.**
- Results of InQol and HADS were compared and correlated with clinical status of the patients by using FDR corrected Spearman's correlation coefficient.

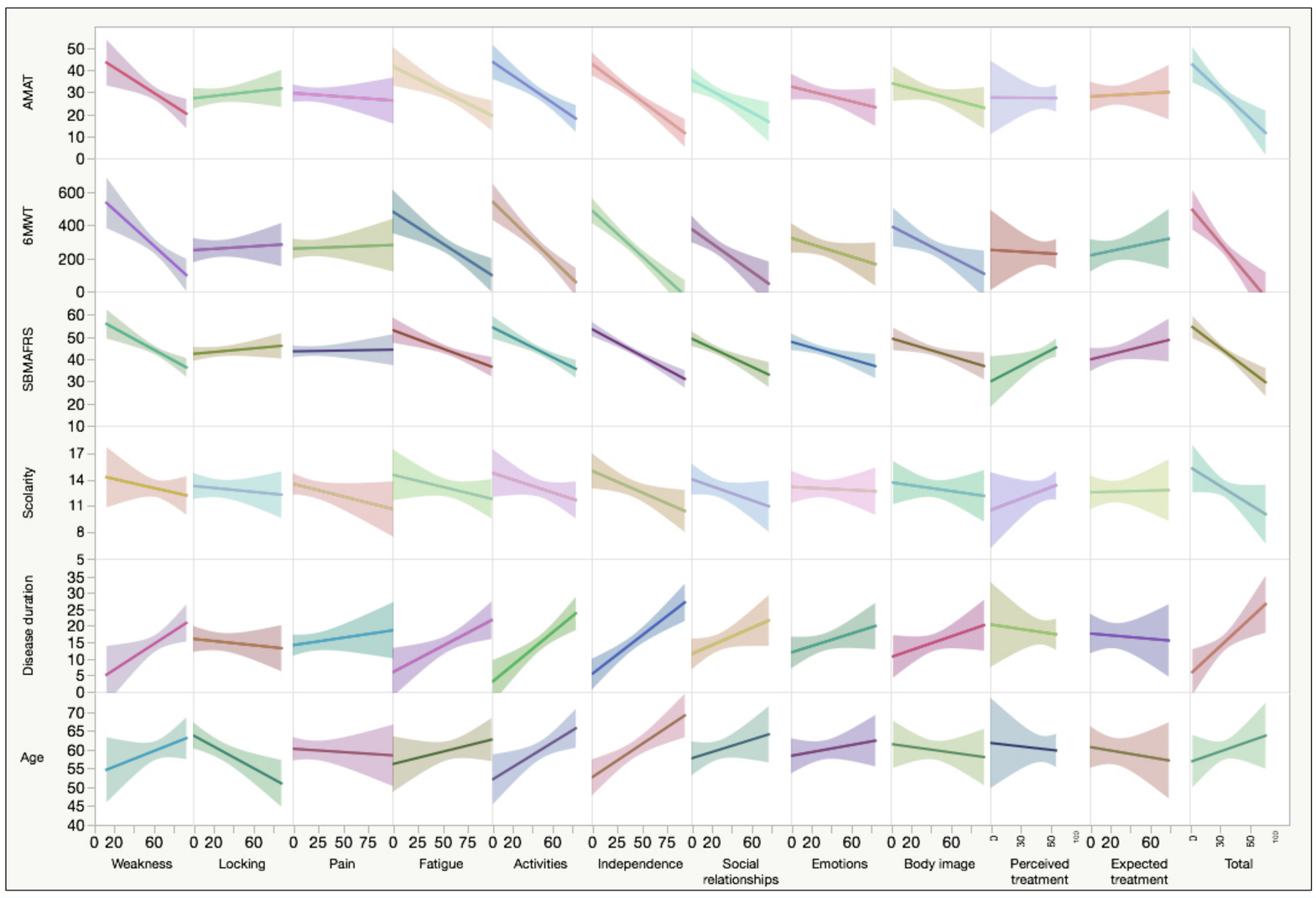
Results

- Mean age of the patients was 60 years +/- 9 (range 40-80). Mean disease duration was 15 years +/- 10 (range 0-45).
- Mean **SBMAFRS score** (indicating functional status and disability) was **44/56 +/- 8**, while mean distance walked at the **6MWT was 263 meters +/- 180** and **mean AMAT score was 29 +/-12**.
- All the patients had a normal cognitive profile.
- Mean InQol Index was 34.9 +/-13 out of 100. Distribution for single domains is shown in the figure.
- At the HADS scale, mean value for anxiety score was 7,6 +/- 4 while the mean score for depression was 6 +/- 3 (diagnostic cut off = 8).
- InQol total score was significantly correlated with disease duration ($p = 0.004$) as well as with SBMA and AMAT score and with the 6WMT ($p < 0.005$).
- Weakness, fatigue, independence and social relations domains significantly correlated with disease duration, 6MWT, AMAT and SBMAFRS ($p<0.05$, R between -0.49 and 0.73).

1 = Weakness
2= Locking
3= Pain
4= Fatigue
5= Activities
6 = Independence
7 = Social relationships
8 = Emotions
9 = Body image
10 = Perceived treatment
11 = Expected treatment



Weakness and fatigue were the domains mostly influencing QoL.



Discussion and Conclusions

This is the first study considering QoL in SBMA patients. Our results evidence a moderate QoL impairment similar to that reported for other neuromuscular diseases (like myotonic dystrophies). Weakness and fatigue are described as the most invalidating symptoms in SBMA.

Wider studies considering bigger populations are warranted to better characterized QoL and to evaluate efficacy of InQol as evaluation tool in SBMA also in the perspective of possible upcoming clinical trials.