Natural history and clinical characteristics of ALS in Taiwan

Kang-Yang Jih[a,b,d], Yi-Chung Lee[a,b,c]
[a]Department of Neurology, Taipei Veterans General Hospital, Taipei, Taiwan
[b]Department of Neurology, National Yang-Ming Chiao Tung University School of Medicine, Taipei, Taiwan
[c]Brain Research Center, National Yang-Ming University, Taipei, Taiwan
[b]Department of Physiology, National Yang-Ming Chiao Tung University School of Medicine, Taipei, Taiwan

Background

Amyotrophic lateral sclerosis (ALS) is a fatal neurodegenerative disease that affects both upper and lower motor neurons. We conducted the largest Taiwanese cohort to investigate the nature history and prognostic factor of ALS.

Methods

We recruited 525 patients diagnosed with definite or probably ALS. All patients were tested for common disease genes including *C9ORF72, SOD1, FUS* and *TARDBP* and detail clinical characteristics were acquired after informed consent. The patients were followed up for evaluation of ALSFRS-R score. 292 of them have received biannually ALSFRS-R score evaluations.

Results

The male and female ratio is 1.44. 13% of the patients exhibited bulbar onset. Average diagnosis delay since symptoms onset was 13.9 months. On average, the functional outcome, evaluated by ALSFRS-R score, decline from 34.2 to 23.8 over the first year of diagnosis. Age of onset, presence of disease causing genes and gender did not affect the rate of functional decline. Initial ALSFRS-R score, bulbar onset, BMI and older age of onset resulted in worse survival outcome. The clinical characteristics are shown in the following figures.

Figure 1. Clinical characteristics of ALS patients

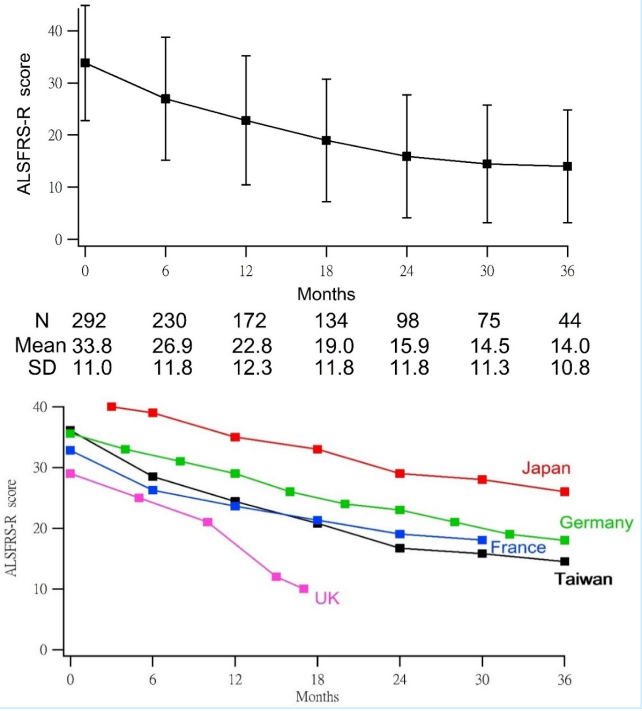
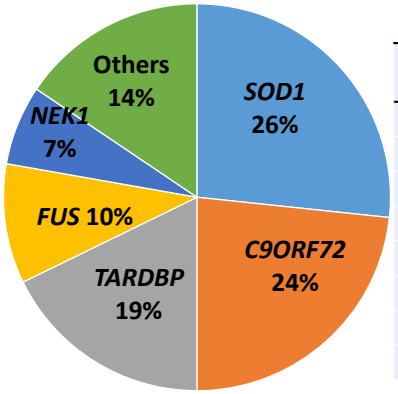


Figure 2. Genetic analysis of ALS patients



Gene	Case number	Gene	Case number
SOD1	24	*HTT*	1
C9ORF72	21	*OPTN*	1
TARDBP	16	*MATR3*	1
FUS	9	*TBK1*	1
NEK1	6	*ANXA11*	1
CCNF	2	*DNAJC7*	1
ATXN2	2	*GLT8D1*	1
TBP	2	*NOTCH2NLC*	1

Figure 3. Factors affecting survival of ALS patients

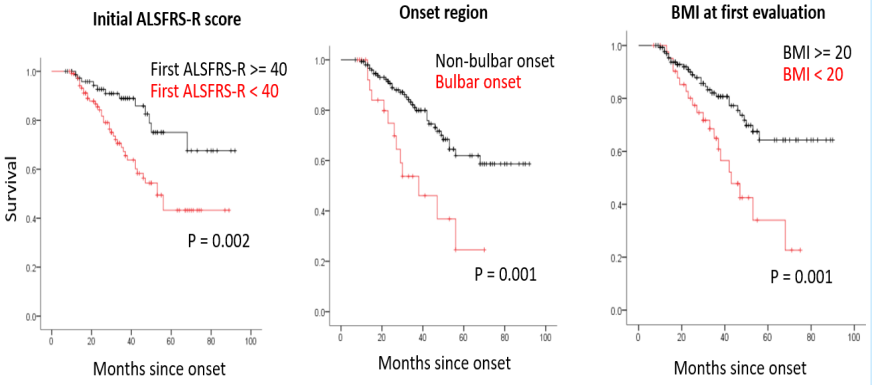
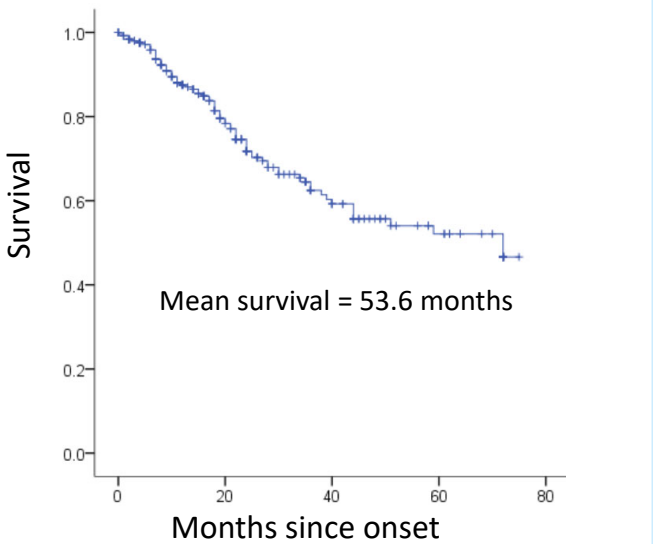


Figure 4. Survival of ALS patients in the cohort



References

[1] Watanabe, Hazuki, et al. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration* 16.3-4 (2015): 230-236.
[2] Jun, Kyo Yeon, et al. *Journal of Neurology, Neurosurgery & Psychiatry* 90.4 (2019): 395-403.
[3] Chen, Lu, et al. *Journal of Neurology, Neurosurgery & Psychiatry* 86.10 (2015): 1075-1081.
[4] Chiò, Adriano, et al. *Neurology* 72.8 (2009): 725-731.
[5] Rooney, James, et al. *PloS one* 8.9 (2013): e74733.

Correspondence: ycli@vghtpe.gov.tw