# **Supporting Information**

# **S1 Methods**

# **Motor functions**

The ALSFRS-R is a validated, questionnaire-based scale that measures physical function in patients with ALS. All items on the ALSFRS-R are also generally applicable to patients with SBMA [1]. The SBMAFRS is another validated functional rating scale which has been utilized to assess the functional status of patients with SBMA [2–4]. The QMG score is an objective measurement used to detect the fatigue of endurance muscle power [5]. We used a part of the QMG score that measures the muscle endurance of outstretched extremities and the lifted head as the mQMG; the best possible score is 0 and the worst possible score is 15. Grip power was measured using an electronic hand dynamometer (TTM digital dynamometer 110kgYD; Tsutsumi Seisakujyo, Kamagaya, Japan). For measurement, the subjects were instructed to keep their elbows at an angle of 90°, their forearms at a neutral rotation, and their wrists not flexed or pronated. We recorded the maximal power of the dominant hand. Tongue pressure was evaluated using a digital tongue pressure measurement device (JMS Co., Ltd., Hiroshima, Japan). We asked the participants to compress the balloon of a disposable intraoral pressure probe upward onto their palates for 7 s using the maximum voluntary effort of the tongue. We recorded tongue pressure (kPa) 3 times at 1 min intervals, and adopted the maximum pressure recorded as the maximal tongue pressure for each time. We used maximal tongue pressure for our analyses [6]. We assessed all measures of motor function, except for the mQMG, in the healthy controls.

# **Quantitative RT-PCR**

Individual 50 mg frozen samples of muscle were homogenized, and total RNA was extracted using TRIzol (Invitrogen, CA, USA) as described previously [7]. The extracted RNA was then reverse-transcribed into first strand cDNA using a ReverTra Ace® qPCR RT Kit (TOYOBO, Osaka, Japan). Quantitative RT (qRT)-PCR was performed in a total volume of 20 µL that contained 10 µL of 2× KOD SYBR® qPCR Mix (TOYOBO, Osaka, Japan) and 40 µM of each primer (Sigma-Aldrich, St Louis, MO, USA); the amplified products were detected with the CFX96™ Real-Time System (Bio-Rad Laboratories, Hercules, CA, USA). The reaction conditions were 98°C for 2 min and 40 cycles of 10 s at 98°C, 10 s at 60°C, and 30 s at 68°C. The expression level of the internal control β2-microglobulin was quantified simultaneously. Relative gene expression was determined using the ΔΔCT method. The following primers were used: 5′-GACAACTCCTCTCGCTTTGG-3′ and 5′-GGCATAATCGTATGGGTTGG-3′ for *MYH1*; 5′-GATGGCACAGAAGTTGCTGA-3′ and 5′-CTTCTCGTAGACGGCTTTGG-3′ for *MYH2*; 5′-TGTGTCACCGTCAACCCTTA-3′ and 5′-TGGCTGCAATAACAGCAAAG-3′ for *MYH7*; 5′-CCTGCATGAGTGTGTGCTCT-3′ and 5′-Gcaaagaggctggtcttcac-3′ for *PGC-1α*; 5′-gtttgagggggtaacagcaa-3′ and 5′-gctaactgcagagggtgagg-3′ for *PPARα*; 5′-actgagttcgccaagagcat-3′ and 5′-gcgttgaacttgacagcaaa-3′ for *PPARδ*; 5′-gctgtgcaggagatcacaga-3′ and 5′-gggctccataaagtcaccaa-3′ for *PPARγ*; 5′-ggagccttgatgtggtagga-3′ and 5′-tttcatccagccttccattc-3′ for *AMPK*; and 5′-TTTCATCCATCCGACATTGA-3′ and 5′-CCTCCATGATGCTGCTTACA-3′ for β2-microglobulin*.*

# **References**

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