Supplementary Material

**Figure e-1.**

Single-photon emission computed tomography (SPECT) images with technetium Tc $^{99m}$ bicisate (Tc $^{99m}$ ethyl cysteinate dimer; Tc $^{99m}$ ECD) revealed bilateral hypoperfusion in the temporal area, inferior frontal area, hippocampus, and thalamus (red arrows) in Japanese patient (Case 1).

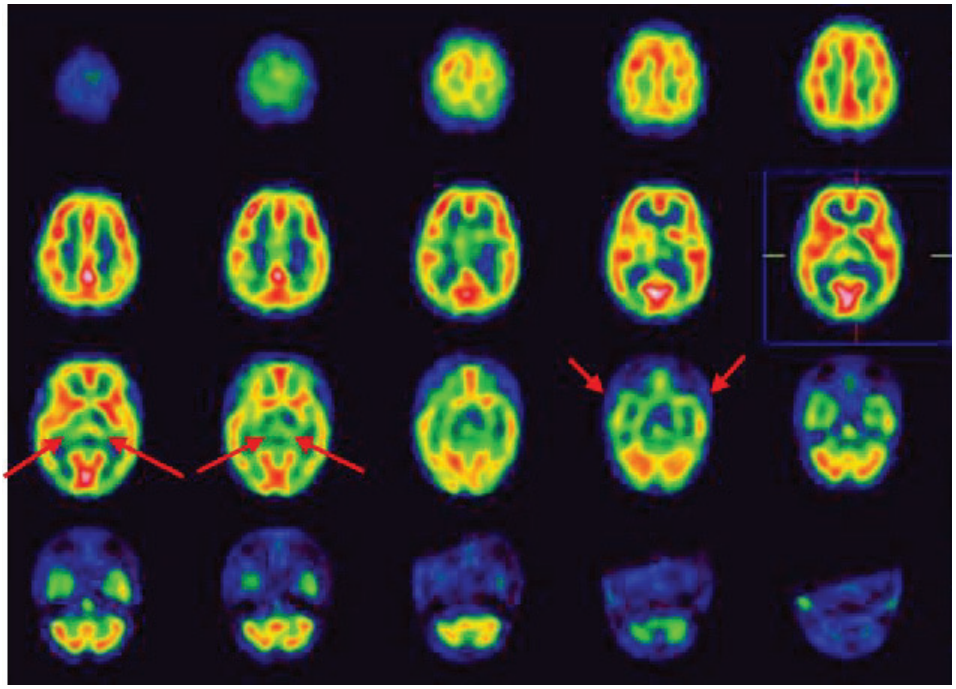


**Table e-1. Measurements of cerebral blood flow in $^{99m}$ Tc ECD SPECT (ml/100g/min)**

| Segment | Right | Left |
|---|---|---|
| Limbic area | 64.34 | 62.88 |
| Parietal area | 57.74 | 60.99 |
| Angular area | 55.55 | 60.99 |
| Temporal area | 57.57 | 57.89 |

| | | |
|---|---|---|
| Occipital area | 61.76 | 60.99 |
| Thalamus | 57.44 | 49.64 |
| Hippocampus | 46.75 | 47.03 |
| Cerebellum | 62.52 | 62.26 |

CBF was 59.1 ml/100 g/min in the right hemisphere and 62.6 ml/100 g/min in the left hemisphere. Hypoperfusion was present in the bilateral temporal area, thalamus, and hippocampus.

**Figure e-2.**

EEG of Japanese case (Case 1) showed bilateral slow wave dysrhythmia with small sharp waves.

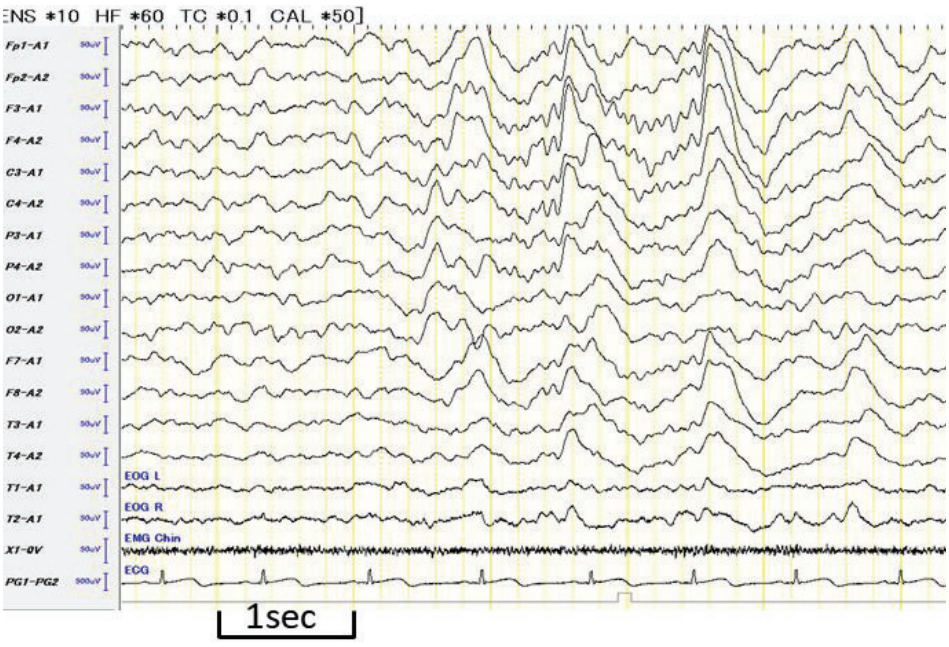



**Figure e-3**

Electrophysiological examinations included saccadic eye movement recorded by a video-based eye-tracking system (EyeLink 1000). Irregular memory-guided saccade (MGS) trace is shown in the Japanese patient.

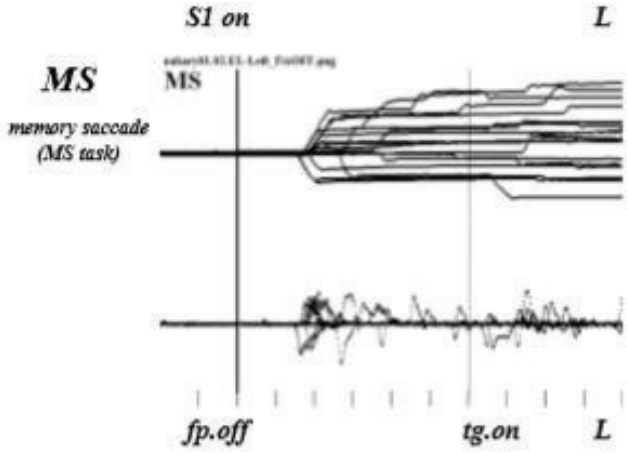


**Figure e-4**

Examination of memory-guided saccade (MGS) showed a decrease to 80.6% of that in age-matched healthy controls, indicating dysfunction of the dopaminergic system in the basal ganglia. Decreased frequency of memory saccade was recorded, as compared with a normal control, at 10 and 20 degrees. The dots show the findings for the present patient.

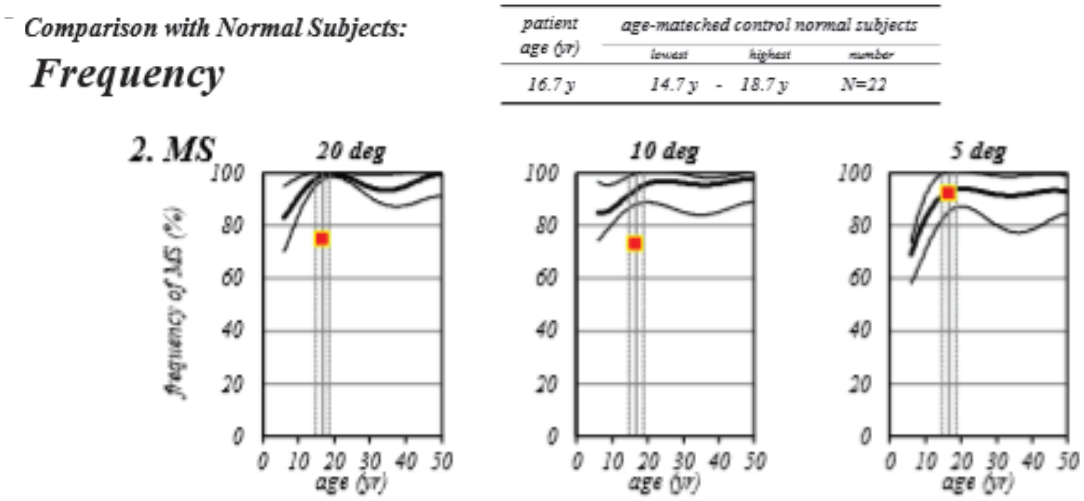
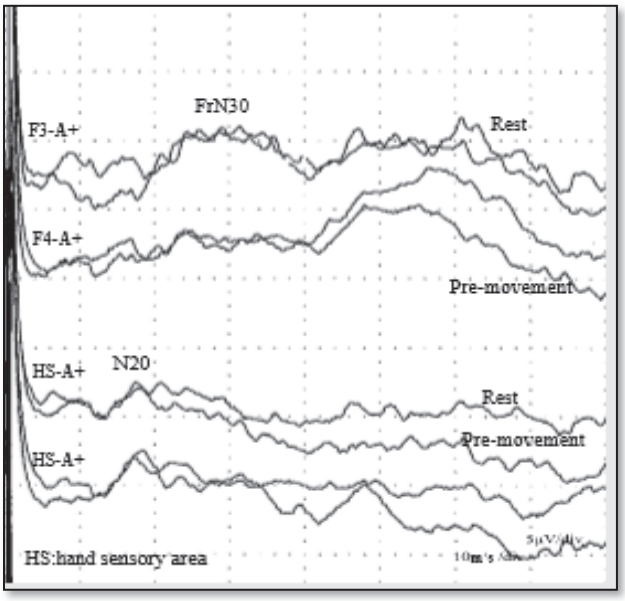



**Fig. e-5. Results of pre-movement gating of somatosensory evoked potentials (gating SEP).**

The amplitude of the contralateral FrN30 (frontal N30) was normally attenuated before movement, suggesting that central gating of SEP was preserved on the left side. However, the affected right side could not be examined because of the severe dystonic posture of the patient. A follow-up examination is necessary in order to evaluate sensorimotor integration in this patient.




**Legends for the supplementary video of case 1**

Segment 1. The right fingers and arm exhibit stiffness and dystonic posture. Rotation of the outstretched forearms from pronation to supination is slow. The patient has limited facial movement, and his jaw is retracted.

Segment 2. Mouth opening and tongue protrusion are restricted because of oromandibular dystonia. His face shows slight hypomimia.

Segment 3. The patient speaks at a low volume with a squeezing voice, probably because of laryngeal dystonia.

Segment 4. The right fifth finger exhibits a dystonic posture, but the patient can write and draw spirals. His writing is micrographic.

Segment 5. Striatal or dystonic toe is noted while he uses his smart phone.

Segment 6. Myerson's sign is present.

Segment 7. The patient has no postural instability.

Segment 8. Arm swing is limited during walking and running. His posture is slightly stooped and stiff. An overall rostro-caudal (face>arm>leg) gradient of involvement is evident.

**Figure e-6. Structure of the transmembrane hairpin site of p.Phe296 and p.Phe297.** Structural consequences of the one-amino acid deletion can be predicted from the crystal structure of Na,K-ATPase in $Na^+$ form (PDB file 3WGU) using Swiss PDB Viewer 4.1. Transmembrane span 3 (M3) is on the left and M4 is on the right; M4 has an interruption of the α-helix (unwound kink) required to accommodate bound ions. The sodium ions (orange spheres) are closer to the viewer than the transmembrane spans, while F296 points toward the viewer and F297 points away. The deletion of 3 bases from the DNA will preserve F296 and twist the residues that follow it, distorting their positions to an extent that is not known. The arrow indicates the direction that the extracellular loop (magenta) will be forced to move. p.Tyr305, over the ion binding site, has hydrophobic interactions with the tops of the adjacent M5-M6 spans that also participate in ion binding. p.Glu309 is a residue that affects the orientation of M4. The top half of the M3-M4 hairpin is known to move with opening and closing of the ion pathway (Ogawa et al., 2015).