Supplementary Table S3. Clinical characteristics of SCA40 patients published in the literature.

| | Family 1 | | Family 2 | | | | Family 3 | Family 4 | Family 5 | | | | Family 6 | Family 7 | Family 8 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| Nationality | Chinese | | Polish | | | | Sudanese | Kurdish | Chinese | | | | Chinese | Indian | Hungarian |
| Mutation (DNA) | c.1391G>A | | c.127G>A | | | | c.1993G>A | c.5922T>G | c.590G>A | | | | c.1886G>A | c.3072C>A | c.607C>T |
| Mutation (protein) | p.R464H | | p.D43N | | | | p.E665K | p.S1974R | p.R197Q | | | | p.R629Q | p.F1024L | p.R203W |
| Patient No. | II-4 | II-5 | III-1 | IV-1 | IV-2 | IV-3 | F83-581 | IV-3 | I-2 | II-1 | II-5 | III-1 | II-3 | - | - |
| Sex | F | M | F | F | M | M | F | F | M | F | F | F | M | M | F |
| Age (years) | 65 | 62 | 84 | 63 | 59 | 53 | 48 | 27 | 72* | 62 | 51 | 39 | 61 | 50 | 65 |
| Age at onset (years) | 43 | 42 | NA | 45 | 49 | 33 | childhood | 13 | 48 | 42 | 40 | 35 | 59 | 50 | 62 |
| First symptom(s) | Gait ataxia, dysarthria | Gait ataxia, dysarthria | UL tremor | UL tremor | UL tremor | UL tremor | Gait problem (tip-toeing) | Hand tremor | NA | Unsteady gait, blurred vision | Ocular dysmetria | Hand tremor | Hand tremor | Unsteady gait | Gait ataxia, dizziness |
| Gait ataxia | + | + | NA | + | + | + | - | + | + | + | + | + | + | + | + |
| UL ataxia | + | + | NA | NA | NA | NA | - | + | NA | NA | NA | NA | + | NA | + (mild) |
| LL ataxia | + | + | NA | NA | NA | NA | - | + | NA | NA | NA | NA | + | NA | + (mild) |
| Ocular dysmetria | + | + | NA | - | - | - | - | - | NA | + | + | + | - | + | - |
| Dysarthria | + | + | NA | - | - | - | - | + | + | + | + | + | + | + | - |
| Rest tremor | - | - | + | + | + | + | - | - | + | + | + | + | + | - | - |
| Action tremor | - | - | + | + | + | + | - | + | NA | - | - | - | - | - | - |
| Intention tremor | + | - | + | - | + | + | - | + | NA | + | - | - | + | - | + (mild) |
| Spasticity | - | + | NA | - | - | - | + | - | NA | - | - | - | + | + | + (mild) |
| Brisk tendon reflexes | + | - | NA | - | + | - | + | + | NA | + | + | - | + | + | - |
| Parkinsonism | - | - | NA | + | + | + | - | - | + | + | - | - | + | - | - |
| Dystonia | - | - | NA | - | - | - | - | - | NA | + | + | + | + | + | - |
| Cognitive impairment | - | - | + | + | + | + | - | - | - | + | + | - | + | - | - |
| Other | - | Impaired vertical gaze | Head tremor | - | - | - | Paraparesis, Babinski sign, foot deformities | Voice tremor | - | Paroxysmal limb jitter, Babinski sign | - | Paroxysmal limb jitter | Chorea/ dyskinesia, dysphagia, anxiety, depression | - | Divergent strabism, dizziness |
| SARA (0-40 points) | 24 | 22 | NA | 5 | 10 | 3 | NA | 14 | NA | 17 | 10 | 6 | 18 | NA | 5 |
| MMSE (0-30 points) | NA | NA | NA | NA | NA | NA | NA | 29 | NA | 21 | 20 | 30 | 20 | NA | NA |
| ACE (0-100 points) | NA | NA | NA | 79 | 84 | 82 | NA | NA | NA | NA | NA | NA | NA | NA | NA |
| Brain MRI | Mild pontine and cerebellar atrophy, moderate WM lesions | Mild pontine and cerebellar atrophy, mild WM lesions | NA | Normal | Normal | Normal | Periventricular leukomalacia with ischemic foci (WM, cerebellum, pons) | Normal | Age-related brain atrophy | Severe cerebellar, mild pontine atrophy | Normal | Moderate cerebellar, mild pontine atrophy | Mild cerebellar and pontine atrophy | Cerebellar and brainstem atrophy, bilateral olivary degeneration | Mild cerebellar (mainly vermian), mild frontal atrophy |
| Nerve conduction studies | NA | NA | NA | NA | NA | NA | Normal | NA | NA | Demyelination of motor nerves | LL neuropathy | NA | NA | Normal | Normal |
| Reference | 2 | | 4 | | | | 5 | 6 | 7 | | | | 8 | 9 | current MS |

+: present, -: not present, *: died

ACE: Addenbrooke's Cognitive Examination, F: female, LL: lower limb, M: male, MMSE: Mini-Mental State Examination, MRI: magnetic resonance imaging, NA: not available, SARA: Scale for the Assessment and Rating of Ataxia, UL: upper limb, WM: white matter