**Supplementary Table 1.** Literature review of SCA42 cases/families reported with the missense mutation (c.5144G>A, p.R1715H) in the *CACNA1G* gene

| Number | Total number of affected individuals | Number of individuals with confirmed mutation | Ethnicity | Onset | Clinical manifestation–movement | Clinical manifestation–other associated symptoms and signs | References, year |
|---|---|---|---|---|---|---|---|
| 1 | 10 from family 1 | 8 from family 1 | Japanese (Hiroshima) | Varied from 20s to 70s | Ataxic gait, dysarthria, and gaze-evoked horizontal nystagmus | None | #1, 2015 |
| 2 | 5 from family 2 | 5 from family 2 | Japanese (Hiroshima) | Age from 18 to 57 years | Ataxic gait, dysarthria, and gaze-evoked horizontal nystagmus (One of the affected individuals also presented resting tremor.) | None | #1, 2015 |
| 2 | 7 from family A | 3 from family A | Japanese (Hokkaido) | 30s | Gait disturbance (ataxia), mild-to moderate speech difficulties (dysarthria), and smooth pursuit defects | None | #2, 2017 |
| 3 | 9 from family A, 4 from family B, and 3 from family C | 7 from family A, 3 from family B, and one from family C | French | Between age 9 and 78 | Gait instability, vertigo, dysarthria, and ocular signs | Urinary symptoms, dysphagia, myokymia orbicularis, and depression | #3, 2015 |
| 4 | 12 from family A | 3 from family A | Italian | Between age 22 and 58 | Pure cerebellar ataxia and dysarthria | None | #4, 2018 |
| 5 | 6 from family B | 3 from family B (including asymptomatic woman) | Easter European ancestry | Age 67 and 39 | Ataxia and dysarthria | None | #4, 2018 |
| 6 | One man | One | Yemen | Age 25 | Imbalance, intermittent jerking movements of his neck, tremor with cervical dystonia, and ataxia | None | #4, 2018 |

SCA, spinocerebellar ataxia.