|  |  |  |  |
| --- | --- | --- | --- |
| **SCA Disorder** | **Position** | **Gene** | **Mutation Screened** |
| SCA1 | 6p22.3 | *ATXN1* | (CAG)n |
| SCA2 | 12q24.12 | *ATXN2* | (CAG)n |
| SCA3 | 14q32.12 | *ATXN3* | (CAG)n |
| SCA4 | 16q22.1 | *PLEKHG4* | Point Mutation |
| SCA5 | 11q13.2 | *SPTBN2* | Point Mutation |
| SCA6 | 19p13.13 | *CACNA1A* | (CAG)n |
| SCA7 | 3p14.1 | *ATXN7* | (CAG)n |
| SCA8 | 13q21.33 | *ATXN8OS/ATXN8* | (CTG)n/(CAG)n |
| SCA10 | 22q13.31 | *ATXN10* | (ATTCT)n |
| SCA11 | 15q15.2 | *TTBK2* | Point Mutation |
| SCA12 | 5q32 | *PPP2R2B* | (CAG)n |
| SCA13 | 19q13.33 | *KCNC3* | Point Mutation |
| SCA14 | 19q13.42 | *PRKCG* | Point Mutation |
| SCA15/16 | 3p26.1 | *ITPR1* | Point Mutation |
| SCA17 | 6q27 | *TBP* | (CAG)n |
| SCA18 | 7q22-q32 | *IRFD1\** | Point Mutation |
| SCA19/22 | 1p13.2 | *KCND3* | Point Mutation |
| SCA21 | 1p36.33 | *TMEM240* | Point Mutation |
| SCA23 | 20p13 | *PDYN* | Point Mutation |
| SCA26 | 19p13.3 | *EEF2* | Point Mutation |
| SCA27 | 13q33.1 | *FGF14* | Point Mutation |
| SCA28 | 18p11.21 | *AFG3L2* | Point Mutation |
| SCA29 | 3p26.1 | *ITPR1* | Point Mutation |
| SCA34 | 6q14.1 | *ELOVL4* | Point Mutation |
| SCA35 | 20p13 | *TGM6* | Point Mutation |
| SCA38 | 6p12.1 | *ELOVL5* | Point Mutation |
| SCA40 | 14q32.11-q32.12 | *CCDC88C* | Point Mutation |
| SCA41 | 4q27 | *TRPC3* | Point Mutation |
| SCA42 | 17q21.33 | *CACNA1G* | Point Mutation |
| SCA43 | 3q25.2 | *MME* | Point Mutation |
| SCA44 | 6q24.3 | *GRM1* | Point Mutation |

**Supplemental Table 1.** Genes screened for SCA mutations

\* Gene is likely responsible for SCA, but requires further confirmation.Gene was screened for known pathogenic point mutations, and novel point mutations, via exome sequencing or PCR repeat analysis

Proband (III-2) Sequential Sagittal T1 Flair Cranial Sections Using Magnetic Resonance Imaging Captured 4 Years Apart. T1 sagittal flair images (A1, B1, and C1) are from 1999; images A2, B2, and C2 are from 2003. Loss of cerebellar volume is evident on neuroradiologic imaging of the patient. The size of the pons does not differ between the imaging obtained in 1999 and 2003