**Supplementary File**

**Copy number variant analysis of spinocerebellar ataxia genes in a cohort of Dutch cerebellar ataxia patients**

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**eTable 1. Description of SCA genes with known SCA symbols. These genes are used in this study for copy number variant analysis.**

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| --- | --- | --- | --- | --- | --- |
| **SCA type** | **Gene name** | **Ref-seq** | **Type of variant** | **Genome region** | **Reference** |
| SCA1 | *ATXN1* | NM\_000332.3 | CAG repeat expansion | Coding | Orr et al., 1993 |
| SCA2 | *ATXN2* | NM\_002973.3 | CAG repeat expansion | Coding | Pulst et al., 1996; Sanpei et al., 1996 |
| SCA3 | *ATXN3* | NM\_004993.5 | CAG repeat expansion | Coding | Kawaguchi et al., 1994 |
| SCA5 | *SPTBN2* | NM\_006946.3 | Missense, in-frame deletion | Coding | Ikeda et al., 2006 |
| SCA6 | *CACNA1A* | NM\_023035.2 | CAG repeat expansion | Coding | Zhuchenko et al., 1997 |
| SCA7 | *ATXN7* | NM\_000333.3 | CAG repeat expansion | Coding | David et al., 1997 |
| SCA8 | *ATXN8* | NR\_002717.2 | CTG repeat expansion | Non-coding | Koob et al., 1999 |
| SCA10 | *ATXN10* | NM\_013236.3 | ATTCT repeat expansion | Non-coding | Matsuura et al., 2000 |
| SCA11 | *TTBK2* | NM\_173500.3 | Frameshift | Coding | Houlden et al., 2007 |
| SCA12 | *PPP2R2B* | NM\_001271899.1 | CAG repeat expansion | Non-coding | Holmes et al., 1999 |
| SCA13 | *KCNC3* | NM\_004977.2 | Missense | Coding | Waters et al., 2006 |
| SCA14 | *PRKCG* | NM\_001316329.1 | Missense | Coding | Chen et al., 2003 |
| SCA15/16/29 | *ITPR1* | NM\_001168272.1 | Missense, deletion | Coding | van de Leemput et al., 2007; Iwaki et al., 2008; Huang et al., 2012 |
| SCA17 | *TBP* | NM\_003194.4 | CAG repeat expansion | Coding | Nakamura et al., 2001 |
| SCA19/22 | *KCND3* | NM\_004980.4 | Missense | Coding | Duarri et al., 2012; Lee et al., 2012 |
| SCA21 | *TMEM240* | NM\_001114748 | Missense | Coding | Delplanque et al., 2014 |
| SCA23 | *PDYN* | NM\_001190892.1 | Missense | Coding | Bakalkin et al., 2010 |
| SCA26 | *eEF2* | NM\_001961.3 | Frameshift | Coding | Hekman et al., 2012 |
| SCA27 | *FGF14* | NM\_001321939.1 | Missense, frameshift | Coding | van Swieten et al., 2003 |
| SCA28 | AFG3L2 | NM\_006796.2 | Missense | Coding | Di Bella et al., 2010 |
| SCA31 | BEAN-TK2 | NM\_001178020.2, NM\_001271934.1 | TGGAA repeat | Non-coding | Sato et al., 2009 |
| SCA34 | *ELOVL4* | NM\_022726 | Missense | Coding | Cadieux-Dion et al., 2014 |
| SCA35 | *TGM6* | NM\_198994.2 | Missense | Coding | Wang et al., 2010 |
| SCA36 | *NOP56* | NM\_006392.3 | GGCCTG repeat expansion | Non-coding | Kobayashi et al., 2011 |
| SCA37 | *DAB1* | NM\_021080.4 | ATTTC repeat expansion | Non-coding | Seixas et al., 2017 |
| SCA38 | *ELOVL5* | NM\_001301856.1 | Missense | Coding | Di Gregorio et al., 2014 |
| SCA40 | *CCDC88C* | NM\_001080414.3 | Missense | Coding | Tsoi et al., 2014 |
| SCA41 | *TRPC3* | NM\_001130698.1 | Missense | Coding | Fogel et al., 2015 |
| SCA42 | *CACNA1G* | NM\_018896.4 | Missense | Coding | Coutelier et al., 2015 |
| SCA43 | *MME* | NM\_000902.3 | Missense | Coding | Depondt et al., 2016 |
| SCA44 | *GRM1* | NM\_001278064.1 | Missense | Coding | Watson et al., 2017 |
| SCA45 | *FAT2* | NM\_001447.2 | Missense | Coding | Nibbeling et al., 2017 |
| SCA46 | *PLD3* | NM\_012268.3 | Missense | Coding | Nibbeling et al., 2017 |
| SCA47 | *PUM1* | NM\_001020658.1 | Missense | Coding | Gennarino et al., 2018 |
| SCA48 | *STUB1* | NM\_005861 | Missense | Coding | Chen et al., 2021 |
| SCA49 | *SAM9DL* | NM\_001303496.3 | Missense | Coding | Corral-Juan et al., 2022 |

Diagram

Description automatically generatedDiagram

Description automatically generated

1. **(B)**

**eFigure 1. Family pedigrees for patient 1 and 2 carrying the *ITPR1* deletion (A), and patient 3 carrying the *PPP2R2B* deletion (B).**

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