Clinical and demographic characteristics of included PSP-ph cases

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| --- | --- | --- | --- | --- | --- | --- | --- |
| **GENE** | **MUTATION** | **N** | **ORIGIN** | **AO** | **ONSET PHENOTYPE** | **OTHER SIGNS** | **REFERENCES** |
| **MAPT** | p. R5L | 1 | American | 41 | prob. PSP-RS | - | 9 |
| p.L284R | 3 | BritishBritishBritish | 404843 | poss. PSP-OMprob. PSP-RS/Fprob. PSP-RS/F | --- | 101010 |
| p.S285R | 3 | DutchDutchJapanese | 404146 | poss. PSP-OMprob. PSP-RSprob. PSP-RS | L-imb dystoniaBell’s phenomenon (grasping?) | 111112 |
| p.delN296 | 2 | ItalianSpanish | 3939 | prob. PSP-RSprob. PSP-F/CBS | -- | 1326 |
| p.N296N | 3 | JapaneseJapaneseJapanese | 424143 | prob. PSP-Pprob. PSP-P/RSprob. PSP-P/RS | OscillopsiaVisual graspingVisual grasping | 151212 |
| p.P301L | 4 | French CanadianAmericanArgentinean | 57634156 | prob. PSP-F-prob. PSP-Pprob. PSP-F/poss. PSP-SL | Fasciculations and brisk reflexes-Fasciculations and brisk reflexes- | 27181716 |
| p.G303V | 7 | French FrenchFrenchFrenchFrenchFrenchFrench | 37413751394232 | prob. PSP-RSs.o. PSP-Pprob. PSP-Ps.o PSP-Pprob. PSP-Pprob. PSP-RSprob. PSP-P | ------- | 20282828282828 |
| p.S305S | 5 | Australian AustralianAustralianFinnishFinnish | 5548544640 | prob. PSP-F/poss. PSP-SLprob. PSP-Pprob. PSP-F/poss. PSP-SLprob. PSP-Fprob. PSP-F | ----- | 2121212222 |
| IVS10+3 | 15 | ItalianItalian-PolishBritish (?)British (?)British (?)British (?)British (?)British (?)British (?)British (?)British (?)British (?)British (?)British (?)British (?) | 506150454254453950475258585248 | poss. PSP-SLprob. PSP-Fprob. PSP-Fprob. PSP-Fprob. PSP-Fprob. PSP-Fprob. PSP-Pprob. PSP-Fprob. PSP-Fs.o. PSP-P prob. PSP-Ps.o. PSP-F/poss. PSP-SLprob. PSP-Fs.o. PSP-Fs.o. PSP-P | ------Hearing lossUp-beating nystagmus--DenervationParesthesiaDenervation ---- | 232425252525252525252525252525 |
| **LRRK2** | R1441H | 1 | Cretan | 61 | s.o. PSP-P | - | 83 |
| p.A1413T | 1 | Filipino | 72 | prob. PSP-RS/P | - | 84 |
| p.G2019S | 1 | Amer. (?) | 73 | prob. PSP-RS | - | 84 |
| **PGRN** | c.813\_816delCACT | 1 | Italian | 68 | s.o PSP-F | - | 48 |
| c.1477CT | 1 | Irish | 63 | pos. PSP-SL/s.o PSP-F | Difficulties in speechOrobuccal dyskinesia | 49 |
| c.102delC | 1 | Czech | 64 | s.o PSP-F/s.o. PSP-P | Possible denervation | 50 |
| del SA470 | 1 | Italian | 63 | s.o PSP-P/prob. PSP-RS | - | 51 |
| c.720CT | 1 | Korean | - | - | - | 52 |
| **DCTN1** | G71E | 2 | FrenchFrench | 5949 | s.o. PSP-Pprob. PSP-F/P | -Limb dystonia | 6161 |
| G71R  | 1 | British(?) | 46 | s.o. PSP-F | - | 64 |
| p.K56R | 2 | ChineseTaiwanese | -- | prob. PSP-Pprob. PSP-P | -- | 6262 |
| c.G36A | 1 | - | 84 |  | - | 63 |
| **C9Orf72** | >30 GGGGCC repeat | 1232 | -FrenchItalian | 645568 | prob. PSP-RSs.o. PSP-Fprob. PSP-P/RS | --- | 383940 |
| **SYNJ1** | Compound c.4217\_4218insC and c.4126A>G | 1 | Tunisian | 21 | prob. PSP-P/RS | - | 55 |
| p.Arg258Gln homozyg. | 3 | ItalianItalianItalian | 282822 | s.o PSP-Ps.o PSP-Ps.o PSP-P | Developmental delayL-dopa induced dystoniaEarly anarthriaLimb dystoniaL-dopa induced oro-mandibular and limb dyskinesia | 565758 |
| **TBK1** | p.Glu643del | 1 | Turkish | 62 | s.o PSP-PI | - | 85 |
| **TARDBP** | p.A382T | 2 | Sardinian | 6770 | s.o. PSP-Pprob. PSP-P/RS | - | 86 |
| **ATP13A2** | c.1632\_1653dup22 | 4 | Jordanian | 13 | prob. PSP-P/RS | FFF mini-myoclonus | 44 |
| c.3176T>Gc.3253delC | 2 | Chinese | 17 | prob. PSP-P/RS | FFF mini-myoclonusOlfactory dysfunction |
| c.2552\_2553delTT | 1 | Afghan | 10 | prob. PSP-P | FFF mini-myoclonus |
| c.1103\_1104insGA | 1 | Pakistani | 17 | prob. PSP-P/RS | FFF mini-myoclonusOlfactory dysfunction |
| c.3057delCc.130615G>A | 5 | Chilean | 13 | prob. PSP-P/RS | FFF mini-myoclonusOlfactory dysfunction |
| c.1510G>C | 1 | Brazilian | 12 | prob. PSP-RS |  |
| c.546C>A | 1 | Japanese | 22 | prob. PSP-P/RS | FFF mini-myoclonus |
| c.2629G>A | 2 | Italian | 7.5 | prob. PSP-P | FFF mini-myoclonus |
| c.2473delCinsAA | 6 | Inuit | 19.5- | prob. PSP-P/RS | FFF mini-myoclonus |
| c.2572C>T | 3 | Iranian | 18 | prob. PSP-P/RS | FFF mini-myoclonus |
| **NPC1/2** | I1061T, V39M/V39M… | - | American, British, French, Canadian, Portuguese, Italian, German | - | prob. PSP-F/poss. PSP-OM | Chorea, limb dystonia, ataxia, myoclonusHearing loss | 46 |
| **GBA** | N370S (SA492) | 2 | ItalianGerman | 4168 | poss. PSP-CBSprob. PSP-RS | Agraphia- | 7778 |
| **PRNP** | A133V M129V | 11 | IndonesianSpanish | 6271 | prob. PSP-RSprob. PSP-F | - | 67 |
| c.198S>F | 4 | AmericanAmericanAmericanAmerican | 51655740 | prob. PSP-P/Fs.o. PSP-Ps.o. PSP-OMprob. PSP-P/F | NystagmusNystagmus, ataxiaNystagmus, ataxiaNystagmus, ataxiaMyoclonus, ataxia | 68686868 |
| c.200E>K | 5 | GermanGermanGermanGermanGerman | 4864594448 | s.o PSP-OMs.o PSP-OMs.o PSP-OMs.o PSP-OMs.o PSP-OM | AtaxiaAtaxiaAtaxiaAtaxiaAtaxia | 6969696969 |
| **ATN1** | >100 CAG  | 31 | JapaneseJapaneseJapaneseChinese | 15133848 | prob. PSP-Ps.o PSP-OMs.o PSP-OMprob. PSP-RS | Ptosis, ataxia, nystagmus, limb dystonia, atetosisAtaxia, nystagmus, limb dystoniaAtaxia, nystagmusAtaxia, dystonia, chorea | 80808049 |
| **ATXN3** | >56 CAG  |  | - | 35 | prob. PSP-P/s.o PSP-OM | Ataxia, nystagmus, limb dystonia | 71, 43 |

AO = Age of onset