|  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Supplementary Table S1. Trend of alternation of age at onset in successive generations of the reported HSP families.** | | | | | | | | | |
| **Reference** | **Generation** | **# of patients** | **Patient ID** | **AAO (y)** | **Mean of AAO (y)** | **Present age (y)** | **Gender** | **Variant** | **Zygosity** |
| 40 | 3 | 7 | III9 | 50 | 45.87 ± 17.13 | Deceased | F | N/A | NA |
| III23 | 50 |
| III25 | 60 | M |
| III27 | 10 |
| III29 | 41 | F |
| III31 | 50 |
| III33 | 60 |
| 4 | 12 | IV19 | 35 | 26.66 ± 13.28 | Deceased | F |
| IV21 | 35 | M |
| IV63 | 35 | F |
| IV67 | 30 |
| IV69 | 35 | M |
| IV113 | 37 | F |
| IV117 | 12 | M |
| IV119 | 10 | F |
| IV129 | 6 | M |
| IV133 | 45 | F |
| IV143 | 30 | M |
| IV145 | 10 | F |
| 5 | 3 | V37 | 25 | 9.66 ± 13.27 | Deceased | F |
| V137 | 2 |
| V141 | 2 |
| 41 | 1 | 2 | I2 | 50 | 45 ± 7.07 | 82 | F | N/A | NA |
| I3 | 40 | 75 |
| 2 | 5 | II3 | 43 | 29.4 ± 13.93 | 60 |
| II5 | 40 | 57 | M |
| II8 | 16 | 50 | F |
| II13 | 35 | 41 |
| II15 | 13 | 40 | M |
| 3 | 2 | III5 | 23 | 18 ± 7.07 | 36 |
| III7 | 13 | 33 | F |
| 44 | 4 | 10 | IV3 | 70 | 50.8 ± 12.36 | Deceased | M | N/A | NA |
| IV9 | 45 |
| IV13 | 60 | F |
| IV14 | 60 |
| IV15 | 55 |
| IV17 | 43 |
| IV19 | 35 | M |
| IV20 | 57 | F |
| IV23 | 30 | M |
| IV29 | 53 |
| 5 | 5 | V9 | N/A | 35 ± 15.53 | M |
| V10 | 54 |
| V12 | 28 |
| V114 | 18 |
| V21 | 40 | F |
| 6 | 1 | VI6 | 17 | 17 | M |
| 30 | 3 | 2 | III5 | 47 | 41 ± 9.89 | 62 | M | c.1579C>T:p.Pro527Ser | Het |
| III7 | 34 | 53 | F |
| 4 | 1 | IV12 | 15 | 15 | 30 | M |
| 29 | 3 | 2 | III5 | 20 | 22.5 ± 3.53 | N/A | M | c.1130-1G>A; mis-splicing | Het |
| III8 | 25 | F |
| 4 | 6 | IV1 | 25 | 39.2 ± 16 | M |
| IV4 | 55 | F |
| IV7 | 43 | M |
| IV9 | 53 | F |
| IV11 | 20 | M |
| IV14 | N/A | F |
| 5 | 6 | V2 | 35 | 15.75 ± 14.24 | M |
| V5 | N/A |
| V6 | 18 |
| V7 | 4 |
| V9 | N/A | F |
| V12 | 6 | M |
| 31 | 1 | 1 | I1 | 45 | 45 | 75 | N/A | c.1841C>T:p.Thr614Ile | Het |
| 2 | 4 | II1 | 44 | 41.5 ± 2.08 | 76 |
| II3 | 42 | 58 |
| II5 | 41 | 56 |
| II13 | 39 | 55 |
| 3 | 5 | III1 | 38 | 35 ± 3.46 | 41 |
| III2 | 38 | 39 |
| III8 | 36 | 43 |
| III11 | 33 | 36 |
| III22 | 30 | 38 |
| 4 | 5 | IV1 | 22 | 22.4 ± 0.54 | 22 |
| IV8 | 22 | 21 |
| IV9 | 23 | 23 |
| IV11 | 23 | 23 |
| IV18 | 22 | 22 |
| 35 | 1 | 1 | I2 | 70 | 70 | N/A | F | c.1340\_1344delTATAA\* | Het |
| 2 | 2 | II1 | 50 | 50 ± 0 | M |
| II2 | 50 | F |
| 3 | 2 | III1 | 24 | 24.5 ± 0.70 | 48 |
| III3 | 25 | 51 | M |
| 4 | 2 | IV1 | 3 | 6.5 ± 4.94 | 23 |
| IV6 | 10 | N/A |
| 28 | 1 | 1 | I2 | 42 | 42 | 61 | F | Del exon 17 at the 5'-end | Het |
| 2 | 1 | II2 | 11 | 11 | 38 |
| 3 | 1 | III1 | 1 | 1 | 6 | N/A |
| 28 | 1 | 1 | I2 | >60 | >60 | Deceased | N/A |  |  |
| 2 | 3 | II1 | 40 | 45 ± 7 | 62 | M | c.1382T>C:p.Leu461Pro | Het |
| II2 | 53 | 58 |
| II3 | 42 | 52 |
| 3 | 4 | III4 | -- | -- | 30 |
| III5 | 24 | 21.5 ± 3.53 | 27 | F |
| III6 | 19 | 25 | M |
| III7 | -- | -- | 25 | F |
| 33 | 2 | 1 | II3 | 25 | 25 | 54 | M | c.1783A>C:p.Ser595Arg | Het |
| 3 | 1 | III2 | 15 | 15 | 23 |
| 32 | 1 | 1 | I2 | 43 | 43 | 53 | F | del exons 8-17 | Het |
| 2 | 1 | II1 | 6 | 6 | 28 |
| 27 | 1 | 1 | I2 | 70 | 70 | 90 | F | c.1004+3A>C:p.Gly290Trpfs\*5 | Het |
| 2 | 1 | II2 | 56 | 56 | 66 |
| 3 | 1 | III1 | 30 | 30 | 38 | M |
| 1 | 1 | I1 | 54 | 54 | 56 | M |
| 2 | 2 | II3 | 24 | 22.5 ± 2.12 | 31 | F |
| II5 | 21 | 27 | M |
| 3 | 1 | III3 | 3 | 3 | 8 | F |
| 34 | 2 | 1 | II6 | 30 | 30 | 63 | F | c.1710\_1712delGAA:p.Lys570del | Het |
| 3 | 1 | III9 | 10 | 10 | 32 | F |
| 4 | 1 | IV9 | 3 | 3 | 10 | M |
| 26 | 2 | 2 | 203 | 60 | 57.5 ± 3.53 | 72 | F | c.1299delG:exon9Ter # | Het |
| 206 | 55 | 73 | F |
| 3 | 5 | 302 | 41 | 34.2 ± 6.53 | 52 | M |
| 305 | 37 | 56 | M |
| 308 | 38 | 50 | M |
| 309 | 25 | 48 | M |
| 312 | 30 | 55 | M |
| 4 | 3 | 401 | 4 | 9.5 ± 7.77 | 34 | M |
| 410 | 15 | 15 | M |
| 411 | # | 15 | F |
| 26 | 3 | 2 | 306 | 55 | 55 ± 0 | 73 | F | del exon 13-16 | Het |
| 320 | 55 | 83 | F |
| 4 | 4 | 408 | 40 | 22.5 ± 11.90 | 56 | F |
| 413 | 20 | 48 | M |
| 414 | 15 | 45 | M |
| 430 | 15 | 55 | M |
| 5 | 3 | 511 | 23 | 15.66 ± 10.21 | 28 | M |
| 518 | 4 | 22 | M |
| 519 | 20 | 20 | M |
| # Based on article | |  |  |  |  |  |  |  |  |