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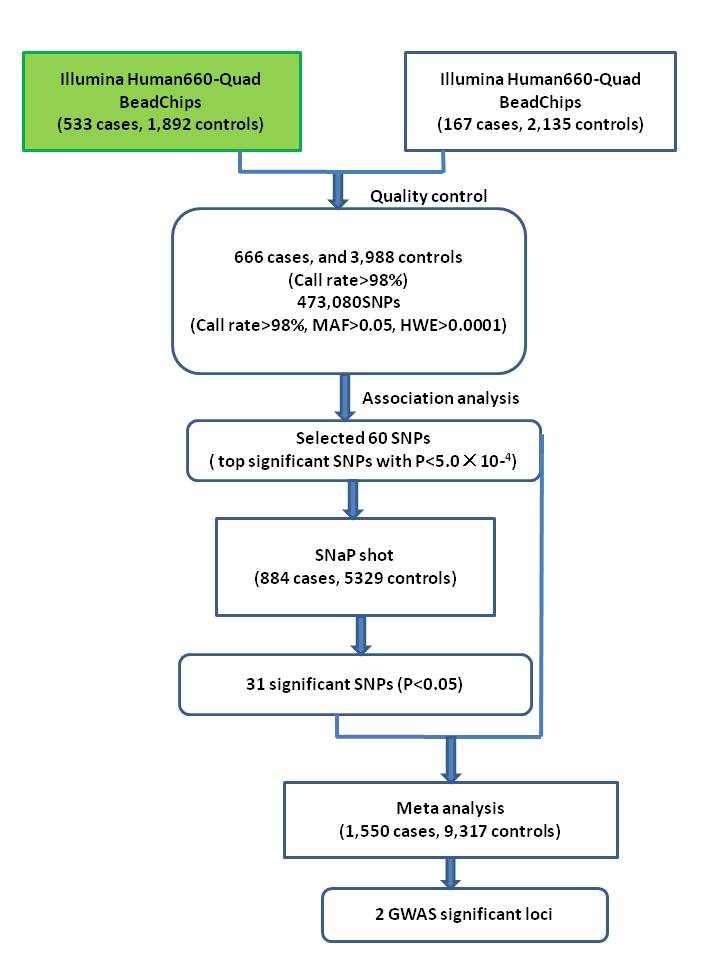
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# Figure e-1 Overview of the study



The boxes in green were accomplished in our previous work and had been published (Deng M. et al. Genome-wide association analyses in Han Chinese identify two new susceptibility loci for amyotrophic lateral sclerosis. Nature Genetics 45, 697-700 (2013).)and the boxes in white were conducted in current study.

# Figure e-2 The principal components analysis (PCA) plot performed in 4,654 samples (666 cases, 3,988 controls) and 206 reference HapMap samples.

ALS-PCA-hapmap-plot20160616.tiff

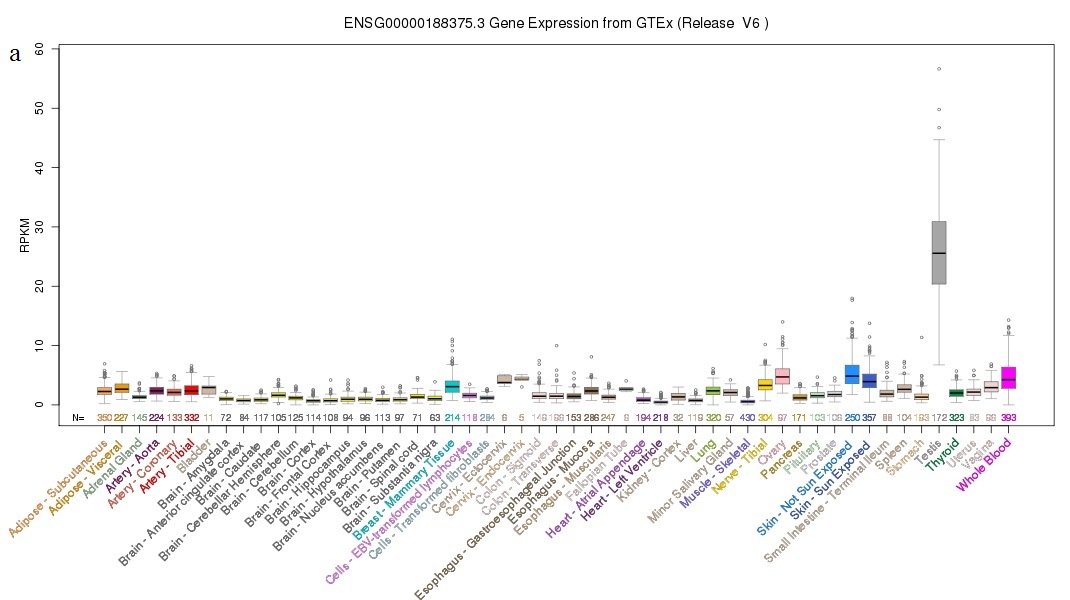
(a) plot of the first and second principle components; (b) plot of the first and third principle components; (c) plot of the first and fourth principle components; (d) plot of the first and fifth principle components.

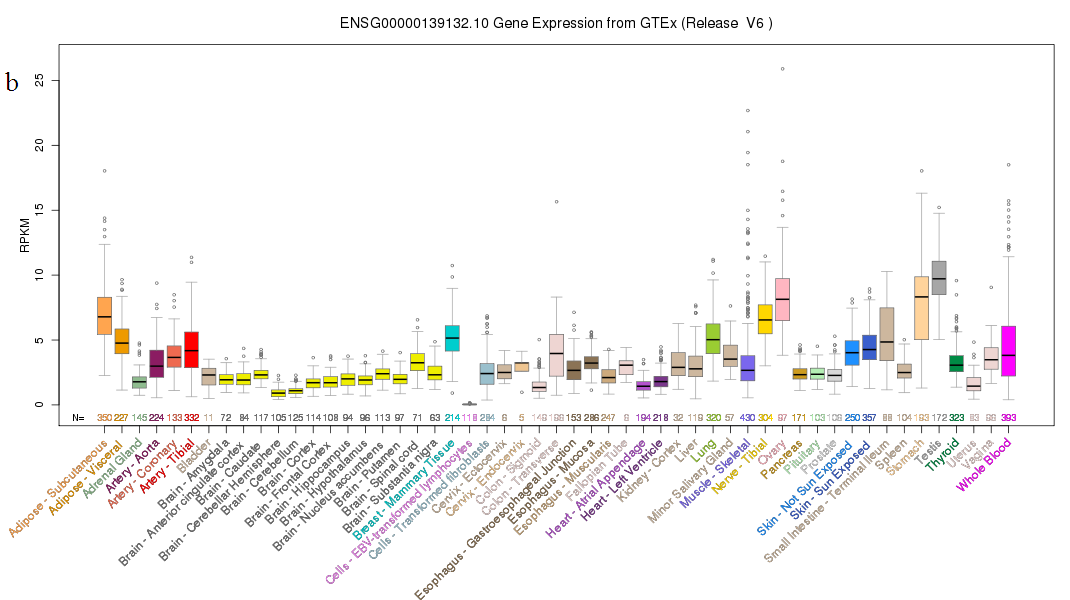
# Figure e-3 The principal components analysis (PCA) plot performed in 4,654 samples(666 cases, 3,988 controls).

**ALS-PCA-plot20160616.tiff**

(a) plot of the first and second principle components; (b) plot of the first and third principle components; (c) plot of the first and fourth principle components; (d) plot of the first and fifth principle components.

# Figure e-4 Gene expression level from GTEx

****

****

The epression level of gene in 53 tissues of 8,555samples. X axis is presented with different tissues. N is the number of samples in each tissue. Y axis is eprssion level quantified by RPKM (reads per kilobase of exon per million mapped reads). (a) *H3F3C*; (b) *FGD4*.

# Figure e-5 STRING analysis of genes from ALSOD

figure 4.tif

The intensity of edges reflects the strengthen of evidence.

# Table e-1The demographic data of cases and controls

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
|  | **Discovery stage** | | |  | **Validation stage** |
|  | **GWAS1** | | **GWAS2** |  |
| **Cases** | | | | | |
| ***Total*** | 533 | 167 | |  | 884 |
| ***Male (%)*** | 61% | 59% | |  | 58% |
| ***Onset age***  ***(year)*** | 48 (16-77) | 50 (18-75) | |  | 52 (20-86) |
| **Controls** | | | | | |
| ***Total*** | 1,892 | 2,135 | |  | 5,329 |
| ***Male (%)*** | 58% | 57% | |  | 58% |

Note: GWAS1, the genotyping data was accomplished in our previous work and had been published; GWAS2, the genotyping data was conducted in current study; the two part of genotyping data was collected for analysis in the discovery stage.Onestage were represented by mean oneset age, minimum and maximum onset age in each cohort.

# Table e-2 The association results of rs6703183 and rs8141797 indenfied in our previous study

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Chr.** | **SNP** | **BP** | **A1** | **MAF\_CA** | **MAF\_CL** | **OR** | **Pgwas** | **MAF\_CA** | **MAF\_CL** | **OR(95%CI)** | **Pvalidation** | **Pcombined** | **Phet** |
| 1 | rs6703183\* | 207779512 | A/G | 0.40 | 0.34 | 1.30 | 2.05 E-05 | 0.41 | 0.34 | 1.32 | 1.98 E-05 | 9.62E-09 | 0.807 |
| 22 | rs8141797\* | 22912041 | G/A | 0.16 | 0.11 | 1.57 | 6.82 E-08 | 0.15 | 0.10 | 1.52 | 1.36 E-05 | 1.18E-12 | 0.778 |

Note: Chr., chromosome; Allele, minor/major allele according to controls; MAF-CA, minor allele frequency in cases; MAF-CL, minor allele frequency in controls;OR, odds ratio; Phet,P value from the heterogeneity test based on GWAS and the validation study samples.

SNP were taken as significant association result with P value passed threshold (Pcombined<5×10−8) combined all samples and with no heterogeneity between GWAS and validation.samples (Phet>0.05).

The p value of GWAS was calculated in current discovery stage with 700 cases and 4,027 controls. The p value of validation was calculated in previous validation stage with 706 cases and 1,777 controls. The combining p value was calculated by combining the two cohorts.

# Table e-3 The association results of 60 validated SNPs

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **CHR** | **SNP** | **BP** | **A1** | **MAF\_CA** | **MAF\_CL** | **OR** | **Pgwas** | **MAF\_CA** | **MAF\_CL** | **OR(95%CI)** | **Pvalidation** | **Pcombined** | **Phet** |
| 1 | rs12145183 | 75045068 | A/G | 0.11 | 0.08 | 1.55 | 3.06E-05 | 0.12 | 0.08 | 1.65 | 8.04E-11 | 2.10E-14 | 0.619 |
| 1 | rs6703930 | 237991493 | G/A | 0.49 | 0.42 | 1.29 | 8.83E-05 | 0.44 | 0.42 | 1.08 | 1.02E-01 | 6.15E-02 | 0.033 |
| 1 | rs16833585 | 30576949 | A/G | 0.14 | 0.10 | 1.41 | 3.30E-04 | 0.11 | 0.11 | 1.05 | 4.89E-01 | 1.84E-01 | 0.019 |
| 1 | rs561686 | 180790126 | G/A | 0.14 | 0.09 | 1.61 | 9.15E-06 | 0.01 | 0.10 | 0.08 | 6.12E-40 | 5.00E-01 | 0.000 |
| 1 | rs6677984 | 4230147 | G/A | 0.17 | 0.24 | 0.64 | 1.91E-06 | 0.53 | 0.27 | 3.01 | 2.16E-97 | 6.71E-01 | 0.000 |
| 1 | rs841853 | 43174025 | A/C | 0.18 | 0.23 | 0.75 | 4.72E-04 | 0.28 | 0.22 | 1.37 | 6.91E-09 | 9.59E-01 | 0.000 |
| 2 | rs1549782 | 67745578 | A/G | 0.08 | 0.05 | 1.66 | 3.39E-05 | 0.05 | 0.05 | 1.01 | 8.99E-01 | 2.97E-01 | 0.003 |
| 2 | rs7558720 | 111313762 | A/G | 0.05 | 0.09 | 0.53 | 1.34E-05 | 0.08 | 0.08 | 0.98 | 8.54E-01 | 3.09E-01 | 0.000 |
| 2 | rs4667857 | 166459759 | C/A | 0.03 | 0.07 | 0.43 | 7.68E-06 | failed | failed | failed | failed | failed | failed |
| 2 | rs13019538 | 12790163 | G/A | 0.24 | 0.19 | 1.31 | 4.83E-04 | 0.11 | 0.19 | 0.55 | 1.08E-15 | 7.05E-01 | 0.000 |
| 3 | rs17015082 | 77333982 | G/A | 0.21 | 0.27 | 0.74 | 1.20E-04 | 0.27 | 0.27 | 0.97 | 6.21E-01 | 2.43E-01 | 0.005 |
| 3 | rs1472476 | 7027542 | A/G | 0.22 | 0.17 | 1.38 | 4.97E-05 | 0.13 | 0.17 | 0.73 | 3.00E-05 | 9.96E-01 | 0 |
| 3 | rs12330894 | 152854479 | A/C | 0.10 | 0.14 | 0.64 | 3.61E-05 | 0.17 | 0.14 | 1.27 | 2.97E-04 | 7.72E-01 | 0.000 |
| 4 | rs4447863 | 9548067 | A/G | 0.39 | 0.46 | 0.75 | 1.90E-05 | 0.43 | 0.46 | 0.91 | 5.20E-02 | 5.17E-02 | 0.022 |
| 4 | rs6813125 | 124489366 | C/A | 0.46 | 0.40 | 1.28 | 1.86E-04 | 0.42 | 0.41 | 1.04 | 4.08E-01 | 1.77E-01 | 0.013 |
| 4 | rs2634945 | 169592042 | G/A | 0.31 | 0.26 | 1.29 | 3.37E-04 | 0.05 | 0.27 | 0.16 | 7.79E-95 | 4.49E-01 | 0.000 |
| 5 | rs890708 | 75719916 | G/A | 0.14 | 0.19 | 0.68 | 2.41E-05 | 0.18 | 0.19 | 0.99 | 8.18E-01 | 2.97E-01 | 0.001 |
| 5 | rs727923 | 110590503 | G/A | 0.05 | 0.09 | 0.53 | 1.01E-05 | 0.13 | 0.09 | 1.59 | 1.54E-10 | 8.88E-01 | 0.000 |
| 6 | rs10214694 | 21321533 | A/C | 0.12 | 0.08 | 1.44 | 4.10E-04 | 0.09 | 0.08 | 1.04 | 6.16E-01 | 2.17E-01 | 0.017 |
| 7 | rs1722923 | 152615014 | A/G | 0.34 | 0.39 | 0.78 | 2.72E-04 | 0.34 | 0.38 | 0.86 | 2.08E-03 | 3.56E-06 | 0.276 |
| 7 | rs940464 | 54189033 | A/G | 0.18 | 0.23 | 0.73 | 1.26E-04 | 0.21 | 0.23 | 0.86 | 9.34E-03 | 1.52E-05 | 0.105 |
| 7 | rs1558475 | 7883885 | G/A | 0.10 | 0.15 | 0.65 | 4.72E-05 | 0.16 | 0.14 | 1.14 | 4.42E-02 | 6.17E-01 | 0.000 |
| 8 | rs4401867 | 123270106 | A/C | 0.18 | 0.14 | 1.39 | 9.76E-05 | 0.17 | 0.15 | 1.18 | 1.03E-02 | 1.04E-05 | 0.134 |
| 8 | rs1808634 | 62133694 | G/A | 0.25 | 0.31 | 0.76 | 1.91E-04 | 0.27 | 0.30 | 0.88 | 2.06E-02 | 4.76E-05 | 0.103 |
| 8 | rs7464428 | 54735548 | A/G | 0.51 | 0.45 | 1.27 | 3.04E-04 | 0.49 | 0.46 | 1.11 | 3.10E-02 | 1.02E-04 | 0.107 |
| 8 | rs4236818 | 94997384 | A/C | 0.26 | 0.31 | 0.76 | 1.90E-04 | 0.59 | 0.31 | 3.25 | 1.20E-132 | 5.33E-01 | 0.000 |
| 9 | rs1483023 | 81685588 | G/A | 0.43 | 0.49 | 0.78 | 1.73E-04 | 0.44 | 0.48 | 0.85 | 1.08E-03 | 1.18E-06 | 0.293 |
| 9 | rs10986088 | 125483780 | A/G | 0.17 | 0.22 | 0.74 | 3.39E-04 | 0.21 | 0.22 | 0.95 | 3.56E-01 | 1.68E-01 | 0.016 |
| 9 | rs4962214 | 134641277 | G/A | 0.10 | 0.07 | 1.48 | 3.53E-04 | 0.08 | 0.07 | 1.03 | 7.63E-01 | 2.60E-01 | 0.012 |
| 9 | rs1111782 | 27202966 | A/G | 0.47 | 0.41 | 1.26 | 4.32E-04 | 0.42 | 0.43 | 0.97 | 4.91E-01 | 4.73E-01 | 0.001 |
| 9 | rs7875184 | 86690918 | A/G | 0.26 | 0.21 | 1.31 | 2.86E-04 | 0.20 | 0.22 | 0.93 | 2.94E-01 | 5.56E-01 | 0.001 |
| 9 | rs12349477 | 27251384 | A/G | 0.39 | 0.33 | 1.31 | 5.89E-05 | 0.33 | 0.35 | 0.93 | 1.47E-01 | 5.83E-01 | 0.000 |
| 9 | rs4240447 | 132018298 | A/G | 0.07 | 0.11 | 0.63 | 1.71E-04 | 0.31 | 0.11 | 3.69 | 7.11E-116 | 6.28E-01 | 0.000 |
| 10 | rs787060 | 26853319 | A/C | 0.43 | 0.37 | 1.27 | 2.74E-04 | 0.16 | 0.37 | 0.31 | 1.01E-78 | 5.11E-01 | 0.000 |
| 10 | rs1703655 | 132494029 | A/C | 0.28 | 0.23 | 1.32 | 1.32E-04 | 0.15 | 0.23 | 0.60 | 2.16E-14 | 7.66E-01 | 0.000 |
| 11 | rs4331088 | 20536042 | G/A | 0.30 | 0.36 | 0.76 | 1.39E-04 | 0.35 | 0.36 | 0.95 | 3.26E-01 | 1.59E-01 | 0.012 |
| 12 | rs1419311 | 31915523 | A/G | 0.40 | 0.34 | 1.28 | 1.83E-04 | 0.39 | 0.33 | 1.25 | 6.54E-06 | 5.19E-09 | 0.777 |
| 12 | rs1666230 | 31921073 | A/G | 0.40 | 0.34 | 1.30 | 8.96E-05 | 0.34 | 0.34 | 1.00 | 9.65E-01 | 3.39E-01 | 0.002 |
| 12 | rs7311906 | 114498749 | A/C | 0.28 | 0.34 | 0.77 | 3.15E-04 | 0.39 | 0.34 | 1.26 | 2.51E-06 | 9.70E-01 | 0.000 |
| 13 | rs1334954 | 18482477 | C/A | 0.17 | 0.13 | 1.38 | 1.82E-04 | failed | failed | failed | failed | failed | failed |
| 13 | rs9568123 | 48373781 | G/A | 0.41 | 0.35 | 1.30 | 9.34E-05 | 0.02 | 0.35 | 0.03 | 5.17E-201 | 3.88E-01 | 0.000 |
| 13 | rs9549299 | 31321045 | A/C | 0.06 | 0.09 | 0.61 | 2.54E-04 | 0.09 | 0.08 | 1.04 | 6.70E-01 | 4.10E-01 | 0.001 |
| 13 | rs7328290 | 71394581 | A/G | 0.19 | 0.24 | 0.75 | 3.86E-04 | 0.47 | 0.23 | 2.87 | 3.61E-100 | 5.68E-01 | 0.000 |
| 14 | rs2193597 | 74304082 | G/A | 0.30 | 0.25 | 1.30 | 2.05E-04 | 0.27 | 0.25 | 1.10 | 7.80E-02 | 2.49E-04 | 0.064 |
| 15 | rs2925269 | 23405763 | G/A | 0.30 | 0.24 | 1.33 | 8.47E-05 | 0.26 | 0.25 | 1.06 | 2.77E-01 | 1.33E-01 | 0.014 |
| 15 | rs2068143 | 61375715 | A/G | 0.15 | 0.10 | 1.58 | 6.80E-07 | 0.05 | 0.11 | 0.44 | 2.31E-15 | 7.74E-01 | 0.000 |
| 16 | rs1420591 | 47607079 | A/G | 0.28 | 0.35 | 0.73 | 1.38E-05 | 0.31 | 0.33 | 0.92 | 1.25E-01 | 9.48E-02 | 0.010 |
| 16 | rs4887970 | 77214539 | A/C | 0.04 | 0.07 | 0.56 | 1.94E-04 | 0.07 | 0.07 | 0.98 | 8.43E-01 | 3.10E-01 | 0.002 |
| 16 | rs152029 | 16079743 | C/A | 0.21 | 0.27 | 0.74 | 1.49E-04 | 0.28 | 0.26 | 1.09 | 1.27E-01 | 5.86E-01 | 0.000 |
| 16 | rs9934342 | 1572966 | A/G | 0.27 | 0.22 | 1.35 | 5.23E-05 | 0.18 | 0.22 | 0.78 | 4.98E-05 | 9.42E-01 | 0.000 |
| 17 | rs8076790 | 38408126 | G/A | 0.22 | 0.17 | 1.35 | 1.86E-04 | 0.17 | 0.18 | 0.99 | 8.65E-01 | 3.65E-01 | 0.003 |
| 17 | rs1948000 | 50502715 | A/C | 0.49 | 0.43 | 1.29 | 9.58E-05 | 0.37 | 0.43 | 0.75 | 6.70E-09 | 9.44E-01 | 0.000 |
| 18 | rs9956771 | 67483382 | G/A | 0.43 | 0.49 | 0.76 | 3.48E-05 | 0.48 | 0.49 | 0.94 | 2.33E-01 | 1.34E-01 | 0.009 |
| 19 | rs577866 | 7513023 | A/G | 0.54 | 0.48 | 1.27 | 2.74E-04 | 0.40 | 0.51 | 0.64 | 3.00E-19 | 7.15E-04 | 0.013 |
| 19 | rs8103733 | 38296371 | A/G | 0.10 | 0.14 | 0.66 | 1.28E-04 | 0.13 | 0.14 | 0.91 | 2.01E-01 | 1.35E-01 | 0.013 |
| 19 | rs12460349 | 38276331 | A/G | 0.10 | 0.14 | 0.67 | 1.67E-04 | 0.15 | 0.14 | 1.07 | 3.53E-01 | 4.86E-01 | 0.000 |
| 20 | rs12625239 | 41076561 | G/A | 0.11 | 0.08 | 1.52 | 8.06E-05 | 0.09 | 0.07 | 1.23 | 1.54E-02 | 1.42E-05 | 0.124 |
| 20 | rs3843781 | 4300277 | A/G | 0.33 | 0.27 | 1.32 | 6.76E-05 | 0.29 | 0.28 | 1.04 | 4.28E-01 | 1.82E-01 | 0.008 |
| 21 | rs2826649 | 21284387 | A/C | 0.17 | 0.12 | 1.47 | 1.18E-05 | failed | failed | failed | failed | failed | failed |
| 22 | rs9610216 | 20428687 | G/A | 0.14 | 0.09 | 1.54 | 6.09E-06 | 0.11 | 0.09 | 1.22 | 9.71E-03 | 1.36E-06 | 0.061 |
| 22 | rs9610417 | 20529970 | A/G | 0.14 | 0.09 | 1.55 | 4.57E-06 | 0.05 | 0.09 | 0.52 | 7.43E-10 | 8.45E-01 | 0.000 |

Note: Chr., chromosome; Allele, minor/major allele according to controls; MAF-CA, minor allele frequency in cases; MAF-CL, minor allele frequency in controls;OR, odds ratio; Phet,P value from the heterogeneity test based on GWAS and the validation study samples.

SNP were taken as significant association result with P value passed threshold (Pcombined<5×10−8) combined all samples and with no heterogeneity between GWAS and validation.samples (Phet>0.05).

# Table e-4 Annotation of rs12145183 and rs1419311

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Chr** | **SNP** | **ref** | **alt** | **AFR** | **AMR** | **ASN** | **EUR** | **Gene** | **Distance** | **dbSNP\_functional\_annotation** | **Score** | **Phylop** | **Phastcons** | **GERPP+** | **Snp effect impact** | **Regulator-id** | **Regulator tissue** | **Factor** |
| 1 | rs12145183 | G | A | 0.11 | 0.43 | 0.08 | 0.48 | TYW3 | 40119 | .intergenetic | 6 | 0.023 | 0.463 | 1.84 | modifier | NA | NA | NA |
| 12 | rs1419311 | G | A | 0.03 | 0.3 | 0.33 | 0.09 | H3F3C | 79080 | . intergenetic | 7 | -0.275 | 0.008 | -0.0842 | modifier | NA | NA | NA |  |

Note:Information by searchingHaploReg (<http://compbio.mit.edu/HaploReg>), Regulome DB (<http://regulome.stanford.edu/>) and SNiPA(<http://snipa.helmholtz-muenchen.de/snipa/> ).Chr., chromosome; ref and alt, refrence allele and alternative allele; AFR, AMR, ASN and EUR, minor allele frequency in African, American, Asian and European population; Gene, nearby gene; Distance, distance from the SNP and the nearby gene; score, Regulome DB score; Phylop, conservation score represented as –log(p) of a test for neutral evolution of a nucleotide, positive score predicates rather conserved; Phastcons, conservation score represented by the probility from 1to 0, 1 predicates rather conserved; GERPP+, conservation score quantified in terms of “rejected substitutions” per nucleotide, positive score is conserved; SNP impact, is catergoried into high/moderate/low/modifider, modifider means there is no evidence of impact; Regulator-id, regulator tissue and factor, regulator effect of SNP in tissues.

# Table e-5 The cis-eQTL for the two SNPS by searhing BRAINEAC

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **Chr** | **SNP** | **GeneSymbol** | **Start** | **Stop** | **P** |
| chr1 | rs12145183 | TYW3 | 75182501 | 75232358 | 3.10E-05 |
| CRYZ | 75153083 | 75203968 | 4.30E-05 |
| CRYZ | 75153083 | 75203968 | 4.80E-05 |
| CRYZ | 75153083 | 75203968 | 7.50E-05 |
| TYW3 | 75182501 | 75232358 | 3.30E-04 |
| TYW3 | 75182501 | 75232358 | 3.80E-04 |
| CRYZ | 75153083 | 75203968 | 4.80E-04 |
| TYW3 | 75182501 | 75232358 | 8.30E-04 |
| CRYZ | 75153083 | 75203968 | 1.70E-03 |
| TYW3 | 75182501 | 75232358 | 2.30E-03 |
| TNNI3K,FPGT,LOC100133219 | 74699504 | 75010108 | 2.50E-03 |
| CRYZ | 75153083 | 75203968 | 5.70E-03 |
| RABGGTB,SNORD45A,SNORD45B,SNORD45C | 76251886 | 76260745 | 1.30E-02 |
| CRYZ | 75153083 | 75203968 | 1.70E-02 |
| TNNI3K,FPGT,LOC100133219 | 74699504 | 75010108 | 2.00E-02 |
| TNNI3K,FPGT,LOC100133219 | 74699504 | 75010108 | 2.70E-02 |
| TYW3 | 75182501 | 75232358 | 3.90E-02 |
| TNNI3K,FPGT,LOC100133219 | 74699504 | 75010108 | 4.10E-02 |
| TYW3 | 75182501 | 75232358 | 4.40E-02 |
| chr12 | rs1419311 | FGD4 | 32654964 | 32798984 | 2.30E-04 |
| FGD4 | 32654964 | 32798984 | 7.50E-04 |
| FGD4 | 32654964 | 32798984 | 8.50E-04 |
| FGD4 | 32654964 | 32798984 | 1.50E-03 |
| FGD4 | 32654964 | 32798984 | 2.30E-03 |
| FGD4 | 32654964 | 32798984 | 3.00E-03 |
| FGD4 | 32654964 | 32798984 | 4.80E-03 |
| DENND5B | 31535162 | 31743975 | 5.40E-03 |
| FGD4 | 32654964 | 32798984 | 5.90E-03 |
| FGD4 | 32654964 | 32798984 | 6.90E-03 |
| C12orf35 | 32101268 | 32218439 | 7.10E-03 |
| FGD4 | 32654964 | 32798984 | 7.30E-03 |
| FGD4 | 32654964 | 32798984 | 9.90E-03 |
| C12orf35 | 32101268 | 32218439 | 1.00E-02 |
| C12orf35 | 32101268 | 32218439 | 1.00E-02 |
| BICD1 | 32259778 | 32533514 | 1.70E-02 |
| YARS2,NAP1L1 | 32878054 | 32908854 | 1.80E-02 |
| YARS2,NAP1L1 | 32878054 | 32908854 | 1.80E-02 |
| AMN1,C12orf72 | 31823443 | 31941741 | 2.00E-02 |
| FGD4 | 32654964 | 32798984 | 2.10E-02 |
| AMN1,C12orf72 | 31823443 | 31941741 | 2.20E-02 |
| OVOS2,OVOS,LOC728715,LOC732435,  LOC100133592,LOC100134413,  LOC100293377,LOC728735,LOC732434 | 31258037 | 31370613 | 2.40E-02 |
| DNM1L | 32832139 | 32900223 | 2.60E-02 |
| DNM1L | 32832139 | 32900223 | 2.90E-02 |
| C12orf35 | 32101268 | 32218439 | 3.00E-02 |
| DNM1L | 32832139 | 32900223 | 3.10E-02 |
| DNM1L | 32832139 | 32900223 | 3.10E-02 |
| FGD4 | 32654964 | 32798984 | 3.10E-02 |
| DNM1L | 32832139 | 32900223 | 3.40E-02 |
| FGD4 | 32654964 | 32798984 | 3.70E-02 |
| YARS2,NAP1L1 | 32878054 | 32908854 | 3.80E-02 |
| FGD4 | 32654964 | 32798984 | 3.90E-02 |
| DNM1L | 32832139 | 32900223 | 4.20E-02 |
| DNM1L | 32832139 | 32900223 | 4.60E-02 |
| FLJ13224 | 31477233 | 31479713 | 4.60E-02 |
| BICD1 | 32259778 | 32533514 | 4.80E-02 |
| DNM1L | 32832139 | 32900223 | 4.90E-02 |

Note:eQTL data by searching BRAINEAC (<http://www.braineac.org/> ). Chr., chromosome; SNP, significant SNPs showed in table 1; Genesymbol, the official gene symbol; Start, the base-pair position of first allele of the gene; Stop, the base-pair position of last allele of the gene.

# 

# Table e-6 The mutation information of genes from ALSOD

|  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| [Gene](javascript:__doPostBack('_ctl0:ContentPlaceHolder1:GridView1','Sort$gene_id')) | [Mutation name](javascript:__doPostBack('_ctl0:ContentPlaceHolder1:GridView1','Sort$mutation_name')) | [Mutation code](javascript:__doPostBack('_ctl0:ContentPlaceHolder1:GridView1','Sort$mutation_mnemonic')) | [Type](javascript:__doPostBack('_ctl0:ContentPlaceHolder1:GridView1','Sort$mutation_type')) | [Seq. Original](javascript:__doPostBack('_ctl0:ContentPlaceHolder1:GridView1','Sort$sequence_original')) | [Seq. Mutated](javascript:__doPostBack('_ctl0:ContentPlaceHolder1:GridView1','Sort$sequence_mutated')) | [AA. Original](javascript:__doPostBack('_ctl0:ContentPlaceHolder1:GridView1','Sort$aa_original')) | [AA. Mutated](javascript:__doPostBack('_ctl0:ContentPlaceHolder1:GridView1','Sort$aa_mutated')) | [Codon Number](javascript:__doPostBack('_ctl0:ContentPlaceHolder1:GridView1','Sort$codon_id')) | [Exon/Intron](javascript:__doPostBack('_ctl0:ContentPlaceHolder1:GridView1','Sort$sequence_location_type')) |
| ALS2 | Ala46delA | 261delA | Deletion | GCA | A | Ala |  | 46 | exon |
| ALS2 | Ala46delA | A47fsX4 | Deletion | GCA at position +3 |  | Ala |  | 47 | exon |
| ALS2 | Ile94Val | I94V | Substitution | ATT | GTT | Ile | Val | 94 | exon |
| ALS2 | Ser100Ile | S100I | Substitution | G | T | Ser | Ile | 100 | exon |
| ALS2 | His102Arg | H102R | Substitution | CAT | CGT | His | Arg | 102 | exon |
| ALS2 | Thr185delA | T185fsX5 | Deletion | ACT at position +1 |  | Thr |  | 185 | exon |
| ALS2 | Val368Met | V368M | Substitution | GTG | ATG | Val | Met | 368 | exon |
| ALS2 | 2660delAT | N846fsX13 | Deletion | GCA | AT | Ala |  | 377 | exon |
| ALS2 | Ala377delAT | 1130delAT | Deletion | GCA | AT | Ala |  | 377 | exon |
| ALS2 | Gln435Leu | Q435L | Substitution | CAG | CTG | Gln | Leu | 435 | exon |
| ALS2 | Glu476delAG | 1548delAG | Deletion | GAG at position +2 |  | Glu |  | 476 | exon |
| ALS2 | Gln490delGTTTCCCCCA | NT1471 | Deletion | CAA | GTTTCCCCCA | Gln |  | 490 | exon |
| ALS2 | Gly540Glu | G540E | Substitution | GGG | GAG | Gly | Glu | 540 | exon |
| ALS2 | Leu623delCT | L623fsX24 | Deletion | CTG at position +1 |  | Leu |  | 623 | exon |
| ALS2 | Gly668STOP | G668X | Substitution | G | T | Gly | STOP | 668 | exon |
| ALS2 | Gln715STOP | Q715X | Substitution | CAG | TAG | Gln | STOP | 715 | exon |
| ALS2 | Val822Met | V822M | Substitution | GTG | ATG | Val | Met | 822 | exon |
| ALS2 | c.2580-2 A>G | c.2580-2 A>G | Substitution | A | G |  |  | 890 | exon |
| ALS2 | Arg998STOP | R998X | Substitution | CGA | TGA | Arg | STOP | 998 | exon |
| ALS2 | Pro1016Thr | P1016T | Substitution | CCT | ACT | Pro | Thr | 1016 | exon |
| ALS2 | Gly1172GlufsX29 | G1172EfsX29 | Deletion | G |  | Gly | Glu | 1172 | exon |
| ALS2 | Lys1174STOP | K1174X | Substitution | AAG | TAG | Lys | STOP | 1174 | exon |
| ALS2 | Val1189TrpfsX19 | V1189WfsX19 | Deletion | G |  | Val | Trp | 1189 | exon |
| ALS2 | Tyr1248delA | M1207X | Deletion | TAT | A | Tyr |  | 1248 | exon |
| ALS2 | Leu1339Leu | L1339L | Substitution | CTG | TTG | Leu | Leu | 1339 | exon |
| ALS2 | Arg1406Lys | R1406K | Substitution | AGG | AAG | Arg | Lys | 1406 | exon |
| ALS2 | Val1525fs | V1525fs | Frameshift |  |  |  |  | 1525 | exon |
| ALS2 | Ile1614delT | V1574fsX44 | Deletion | ATT | T | Ile |  | 1614 | exon |
| ANG | Met-24Ile | M-24I | Substitution | ATG | ATA | Met | Ile | -24 | exon |
| ANG | Phe-13Ser | F-13S | Substitution | TTC | TCC | Phe | Ser | -13 | exon |
| ANG | Phe-13Leu | F-13L | Substitution | C | T | Phe | Leu | -13 | exon |
| ANG | Gly-10Asp | G-10D | Substitution |  |  | Gly | Asp | -10 | exon |
| ANG | Pro-4Ser | P-4S | Substitution | CCG | TCG | Pro | Ser | -4 | exon |
| ANG | Gln12Leu | Q12L | Substitution | CAG | CTG | Gln | Leu | 12 | exon |
| ANG | Tyr14His | Y14H | Substitution | TAT | CAT | Tyr | His | 14 | exon |
| ANG | Lys17Ile | K17I | Substitution | AAA | ATA | Lys | Ile | 17 | exon |
| ANG | Lys17Glu | K17E | Substitution | cAAA | GAA | Lys | Glu | 17 | exon |
| ANG | Gly20Gly | G20G | Polymorphism | GGC | GGG | Gly | Gly | 20 | exon |
| ANG | Asp22Gly | D22G | Substitution | GAT | GGT | Asp | Gly | 22 | exon |
| ANG | Ser28Asn | S28N | Substitution | AGC | AAC | Ser | Asn | 28 | exon |
| ANG | Arg31Lys | R31K | Substitution | AGG | AAG | Arg | Lys | 31 | exon |
| ANG | Cys39Trp | C39W | Substitution | TGCa | TGG | Cys | Trp | 39 | exon |
| ANG | Lys40Ile | K40I | Substitution | AAA | ATA | Lys | Ile | 40 | exon |
| ANG | Ile46Val | I46V | Substitution | tATT | GTT | Ile | Val | 46 | exon |
| ANG | Asn49Ser | N49S | Substitution | A | G | Asn | Ser | 49 | exon |
| ANG | Lys54Glu | K54E | Substitution | AAG | GAG | Lys | Glu | 54 | exon |
| ANG | Leu59Pro | L59P | Substitution | T | C | Leu | Pro | 59 | exon |
| ANG | Thr80Ser | T80S | Substitution | C | G | Thr | Ser | 80 | exon |
| ANG | Lys84Glu | K84E | Substitution | A | G | Lys | Glu | 84 | exon |
| ANG | His84Arg | H84R | Substitution | CAT | CGT | His | Arg | 84 | exon |
| ANG | Phe100Ile | F100I | Substitution |  |  | Phe | Ile | 100 | exon |
| ANG | Val103Ile | V103I | Substitution | GTT | ATT | Val | Ile | 103 | exon |
| ANG | Pro112Leu | P112L | Substitution | CCT | CTT | Pro | Leu | 112 | exon |
| ANG | Val113Ile | V113I | Substitution | GTC | ATC | Val | Ile | 113 | exon |
| ANG | His114Arg | H114R | Substitution | CAC | CGC | His | Arg | 114 | exon |
| ANG | Arg121His | R121H | Substitution | CGT | CAT | Arg | His | 121 | exon |
| ANG | Arg145Cys | R145C | Substitution | C | T | Arg | Cys | 145 | exon |
| ARHGEF28 | intron 6, + 1 delG (GT> TT) | intron 6, + 1 delG (GT> TT) | splicing |  |  |  |  |  | intron |
| ARHGEF28 | K280M> fs40X | K280M> fs40X | frameshift |  |  |  |  | 280 | exon |
| ATXN2 | CAG repeat size 33 | CAG repeat size 33 | RepeatExp | 0 | 0 |  |  | 0 | exon |
| ATXN2 | CAG repeat size 34 | CAG repeat size 34 | RepeatExp | 0 | 0 |  |  | 0 | exon |
| ATXN2 | CAG repeat size 36 | CAG repeat size 36 | RepeatExp | 0 | 0 |  |  | 0 | exon |
| ATXN2 | CAG repeat size 39 | CAG repeat size 39 | RepeatExp | 0 | 0 |  |  | 0 | exon |
| ATXN2 | CAG repeat size 37 | CAG repeat size 37 | RepeatExp | 0 | 0 |  |  | 0 | exon |
| ATXN2 | CAG repeat size 35 | CAG repeat size 35 | RepeatExp | 0 | 0 |  |  | 0 | exon |
| ATXN2 | CAG repeat size 31 | CAG repeat size 31 | RepeatExp | 0 | 0 |  |  | 0 | exon |
| ATXN2 | CAG repeat size 27 | CAG repeat size 27 | RepeatExp | 0 | 0 |  |  | 0 | exon |
| ATXN2 | CAG repeat size 32 | CAG repeat size 32 | RepeatExp | 0 | 0 |  |  | 0 | exon |
| BCL11B | Glu32Val | E32V | Substitution | A | T | Glu | Val | 32 | exon |
| BCL11B | Pro229Ser | P229S | Substitution | C | T | Pro | Ser | 229 | exon |
| BCL6 | Asn73Ser | N73S | Substitution | A | G | Asn | Ser | 73 | exon |
| C9orf72 | HREM | HREM | Repeat |  | GGGGCC |  |  | 0 | intron |
| CDH13 | Arg65Cys | R65C | Substitution | C | T | Arg | Cys | 65 | exon |
| CDH13 | Ala103Val | A103V | Substitution | C | T | Ala | Val | 103 | exon |
| CDH13 | Gly113Arg | G113R | Substitution | G | C | Gly | Arg | 113 | exon |
| CDH13 | Arg246Trp | R246W | Substitution | C | T | Arg | Trp | 246 | exon |
| CDH13 | Glu367Gln | E367Q | Substitution | G | C | Glu | Gln | 367 | exon |
| CDH22 | Glu92Lys | E92K | Substitution | G | A | Glu | Lys | 92 | exon |
| CDH22 | Thr134Met | T134M | Substitution | C | T | Thr | Met | 134 | exon |
| CDH22 | Arg533His | R533H | Substitution | G | A | Arg | His | 533 | exon |
| CHCHD10 | Arg15Leu | R15L | Substitution | C | A | Arg | Leu | 15 | exon |
| CHCHD10 | Pro34Ser | P34S | Substitution | C | T | Pro | Ser | 34 | exon |
| CHCHD10 | Ser59Leu | S59L | Substitution | C | T | Ser | Leu | 59 | exon |
| CHCHD10 | Gly66Val | G66V | Substitution | C | A | Gly | Val | 66 | exon |
| CHCHD10 | Pro80Leu | P80L | Substitution |  |  | Pro | Leu | 80 | exon |
| CHGB | Pro413Leu | P413L | Substitution | C | T | Pro | Leu | 413 | exon |
| CHMP2B | Arg22Gln | R22Q | Substitution | G | A | Arg | Gln | 22 | exon |
| CHMP2B | Ile29Val | I29V | Substitution | A | G | Ile | Val | 29 | exon |
| CHMP2B | Ser103Cys | S103C | Substitution | C | G | Ser | Cys | 103 | exon |
| CHMP2B | Glu201Gln | E201Q | Substitution | G | A | Glu | Gln | 201 | exon |
| CHMP2B | Gln206His | Q206H | Substitution | A | C | Gln | His | 206 | exon |
| CHMP2B | Thr104Asn | T104N | Substitution | C | A | Thr | Asn | 311 | exon |
| CNTN6 | Phe314Val | F314V | Substitution | T | G | Phe | Val | 314 | exon |
| CNTN6 | Glu954Val | E954V | Substitution | A | T | Glu | Val | 954 | exon |
| CRIM1 | Asn406Ser | N406S | Substitution | A | G | Asn | Ser | 406 | exon |
| CRYM | Arg169Cys | R169C | Substitution | C | T | Arg | Cys | 169 | exon |
| CX3CR1 | Val249Ile | V249I | Substitution | 0 | 0 | Val | Ile | 249 | exon |
| CX3CR1 | Thr280Met | T280M | Substitution | 0 | 0 | Thr | Met | 280 | exon |
| DAO | Arg38His | R38H | Substitution | G | A | Arg | His | 38 | exon |
| DAO | Arg199Trp | R199W | Substitution | C | T | Arg | Trp | 199 | exon |
| DCTN1 | Gly59Ser | G59S | Substitution | C | T | Gly | Ser | 59 | exon |
| DCTN1 | Gly59Arg | G59R | Substitution |  |  | Gly | Arg | 59 | exon |
| DCTN1 | Met571Thr | M571T | Substitution | T | C | Met | Thr | 571 | exon |
| DCTN1 | Arg785Trp | R785W | Substitution | C | T | Arg | Trp | 785 | exon |
| DCTN1 | R997W | R997W | Substitution | C | T | Arg | Trp | 997 | exon |
| DCTN1 | Arg1101Lys | R1101K | Substitution | G | A | Arg | Lys | 1101 | exon |
| DCTN1 | Thr1249Ile | T1249I | Substitution | C | T | Thr | Ile | 1249 | exon |
| DIAPH3 | Ile234Thr | I234T | Substitution | T | C | Ile | Thr | 234 | exon |
| DIAPH3 | Pro578Leu | P578L | Substitution | C | T | Pro | Leu | 578 | exon |
| DIAPH3 | Pro588Leu | P588L | Substitution | C | T | Pro | Leu | 588 | exon |
| DIAPH3 | Pro596Leu | P596L | Substitution | C | T | Pro | Leu | 596 | exon |
| DIAPH3 | Arg1042His | R1042H | Substitution | G | A | Arg | His | 1042 | exon |
| DIAPH3 | Arg1191X | R1191X | Substitution | C | T | Arg | X | 1191 | exon |
| DOC2B | Arg209Leu | R209L | Substitution | G | T | Arg | Leu | 209 | exon |
| ERBB4 | Arg927Gln | R927Q | Substitution | G | A | Arg | Gln | 927 | exon |
| ERBB4 | Arg1275Trp | R1275W | Substitution | C | T | Arg | Trp | 1275 | exon |
| Error | CAG repeat size 25 | CAG repeat size 25 | RepeatExp | 0 | 0 |  |  | 0 | exon |
| EWSR1 | Gln235Gln | Q235Q | Subtitution | G | A | Gln | Gln | 235 | exon |
| FEZF2 | Gly188Asp | G188D | Substitution | G | A | Gly | Asp | 188 | exon |
| FIG4 | Arg23del23-55 | R23del(23-55) | Substitution | G | T | Arg | STOP | 23 | exon |
| FIG4 | Asp48Gly | D48G | Substitution | A | G | Asp | Gly | 48 | exon |
| FIG4 | Asp53Tyr | D53Y | Substitution | G | T | Asp | Tyr | 53 | exon |
| FIG4 | Arg183STOP | R183X | Substitution | C | T | Arg |  | 183 | exon |
| FIG4 | Arg388Gly | R388G | Substitution | A | G | Arg | Gly | 388 | exon |
| FIG4 | Gln403STOP | Q403X | Substitution | C | T | Gln |  | 403 | exon |
| FIG4 | Ile411Val | I411V | Substitution | A | G | Ile | Val | 411 | exon |
| FIG4 | Ser424\_K462 | S424del insR | Substitution | G | T | Ser | STOP | 424 | exon |
| FIG4 | Tyr647Cys | Y647C | Substitution | T | G | Tyr | Cys | 647 | exon |
| FIG4 | Ile902Thr | I902T | Substitution | T | C | Ile | Thr | 902 | exon |
| FUS | Ala2Ala | A2A | Substitution | C | T | Ala | Ala | 2 | exon |
| FUS | Pro18Ser | P18S | Substitution | CCC | TCC | Pro | Ser | 18 | exon |
| FUS | Gly49Gly | G49G | Substitution | C | A | Gly | Gly | 49 | exon |
| FUS | Gly51Gly | G51G | Substitution | C | T | Gly | Gly | 51 | exon |
| FUS | Ser57delTCT | S57delTCT | Deletion | TCT |  | Ser |  | 57 | exon |
| FUS | Asn63Ser | N63S | Substitution | AAC | AGC | Asn | Ser | 63 | exon |
| FUS | Gly74Gly | G74G | Substitution | A | G | Gly | Gly | 74 | exon |
| FUS | Tyr91Tyr | Y91Y | Substitution | C | T | Tyr | Tyr | 91 | exon |
| FUS | Ser96delCCTACinsAT | 287291delCCTACinsAT | Deletion |  |  |  |  | 96 | exon |
| FUS | Tyr97Tyr | Y97Y | Substitution | C | T | Tyr | Tyr | 97 | exon |
| FUS | Ser115Asn | S115N | Substitution | AGC | AAC | Ser | Asn | 115 | exon |
| FUS | Ser142Asn | S142N | Substitution | AGC | AAC | Ser | Asn | 142 | exon |
| FUS | c.491\_495 +1delGAGGTgc | G144\_Y149del | Deletion |  | delGGACAG | Gly |  | 144 | exon |
| FUS | Gly156Glu | G156E | Substitution | GGA | GAA | Gly | Glu | 156 | exon |
| FUS | Asn159Tyr | N159Y | Substitution | AAC | TAC | Asn | Tyr | 159 | exon |
| FUS | Gly168Gly | G168G | Substitution | A | T | Gly | Gly | 168 | exon |
| FUS | Gly170Gly | G170G | Substitution | A | T | Gly | Gly | 170 | exon |
| FUS | c.521\_523+3delGAGGTG | G173\_G174del | Deletion | GAGGTG |  | GAGGTG |  | 173 | exon |
| FUS | c.430\_447delGGACAGCAGCAAAGCTAT | G174\_G175del | Deletion |  | delGAG | Gly |  | 174 | exon |
| FUS | Gly175 | G175 | Insertion | GGT | GAGGTG | Gly |  | 175 | exon |
| FUS | Gly187Ser | G187S | Substitution | GGT | AGT | Gly | Ser | 187 | exon |
| FUS | Gly191Ser | G191S | Substitution | GGT | AGT | Gly | Ser | 191 | exon |
| FUS | Gln210His | Q210H | Substitution | G | C | Gln | His | 210 | exon |
| FUS | Arg216Cys | R216C | Substitution | CGC | TGC | Arg | Cys | 216 | exon |
| FUS | c.666\_667insGGC | G222\_G223insG | Insertion |  | insGGC | Gly |  | 222 | exon |
| FUS | c.667\_669delGGC | G223del | Deletion |  | delGGC | Gly |  | 223 | exon |
| FUS | G223-G226del | c.667-678delGGCGGCGGCGGC | Deletion |  |  |  |  | 223 | exon |
| FUS | Gly225Val | G225V | Substitution | GGC | GTC | Gly | Val | 225 | exon |
| FUS | Gly226Ser | G226S | Substitution | G | A | Gly | Ser | 226 | exon |
| FUS | c.676\_684del | 228\_230delGGG | Deletion | GGG |  | GGG |  | 228 | exon |
| FUS | c.684\_685insGGC | Gly228\_Gly229insGly | Insertion |  | GGC |  | GGC | 228 | exon |
| FUS | Gly228Gly | G228G | Substitution | C | T | Gly | Gly | 228 | exon |
| FUS | c.681\_684delGGC | 230delG | Deletion | G |  | G |  | 230 | exon |
| FUS | Gly230Cys | G230C | Substitution | GGT | TGT | Gly | Cys | 230 | exon |
| FUS | Arg234Cys | R234C | Substitution | CGC | TGC | Arg | Cys | 234 | exon |
| FUS | Arg234Leu | R234L | Substitution | CGC | CTC | Arg | Leu | 234 | exon |
| FUS | Arg244Cys | R244C | Substitution | CGT | TGT | Arg | Cys | 244 | exon |
| FUS | Met254Val | M254V | Substitution | ATG | GTG | Met | Val | 254 | exon |
| FUS | Arg383Cys | R383C | Substitution | C | T | Arg | Cys | 383 | exon |
| FUS | Arg386Arg | R386R | Substitution | C | A | Arg | Arg | 386 | exon |
| FUS | Pro391Pro | P391P | Substitution | C | A | Pro | Pro | 391 | exon |
| FUS | Gly399Val | G399V | Substitution | GGT | GTT | Gly | Val | 399 | exon |
| FUS | Ser439Ser | S439S | Silent | TCT | TCC | Ser | Ser | 439 | exon |
| FUS | Ser462Phe | S462F | Substitution | C | T | Ser | Phe | 462 | exon |
| FUS | Met464Ile | M464I | Substitution | ATG | ATT | Met | Ile | 464 | exon |
| FUS | Y485AfsX514 | c.1449-1488delCTACCGGGGCCGCGGCGGGGACCGTGGAGGCTTCCGAGGG | frameshift |  |  |  |  | 485 | exon |
| FUS | R495EfsX527 | c.1483delC | frameshift |  |  |  |  | 485 | exon |
| FUS | G497AfsX527 | c.1485delA | frameshift |  |  |  |  | 485 | exon |
| FUS | K510WfsX517 | c.1527insTGGC | frameshift |  |  |  |  | 485 | exon |
| FUS | Arg487Cys | R487C | Substitution | CGC | TGC | Arg | Cys | 487 | exon |
| FUS | Gly488Gly | G488G | Substitution | C | T | Gly | Gly | 488 | exon |
| FUS | R495QfsX527 | c.1484delG | frameshift |  |  |  |  | 495 | exon |
| FUS | Arg495STOP | R495X | Substitution | CGA | TGA | Arg | STOP | 495 | exon |
| FUS | Gly496Glyfs\*31 | c.1486delG | Deletion | G |  | Gly |  | 496 | exon |
| FUS | c.1507\_1508delAG | G503WfsX12 | Deletion | AG |  | AG |  | 503 | exon |
| FUS | Gly507Asp | G507D | Substitution | GGC | GAC | Gly | Asp | 507 | exon |
| FUS | Lys510Glu | K510E | Substitution | AAG | GAG | Lys | Glu | 510 | exon |
| FUS | Lys510Arg | K510R | Substitution | AAG | AGG | Lys | Arg | 510 | exon |
| FUS | Arg514Ser | R514S | Substitution | AGG | AGT | Arg | Ser | 514 | exon |
| FUS | Arg514Gly | R514G | Substitution | AGG | GGG | Arg | Gly | 514 | exon |
| FUS | Arg514Arg | R514R | Substitution | AGG | CGG | Arg | Arg | 514 | exon |
| FUS | Gly515Cys | G515C | Substitution | GGT | TGT | Gly | Cys | 515 | exon |
| FUS | His517Gln | H517Q | Substitution | CAC | CAG | His | Gln | 517 | exon |
| FUS | His517Asp | H517D | Substitution | CAC | GAC | His | Asp | 517 | exon |
| FUS | Arg518Lys | R518K | Substitution | AGA | AAA | Arg | Lys | 518 | exon |
| FUS | Gln519STOP | Q519X | Substitution | C | T | Gln | STOP | 519 | exon |
| FUS | c.1506dupA | R502fsX15 | Substitution |  |  | Arg |  | 520 | exon |
| FUS | Arg521Gly | R521G | Substitution | CGC | GGC | Arg | Gly | 521 | exon |
| FUS | Arg521His | R521H | Substitution | CGC | CAC | Arg | His | 521 | exon |
| FUS | Arg521Cys | R521C | Substitution | CGC | TGC | Arg | Cys | 521 | exon |
| FUS | Arg521Ser | R521S | Substitution | CGC | AGC | Arg | Ser | 521 | exon |
| FUS | Arg521Leu | R521L | Substitution | CGC | CTC | Arg | Leu | 521 | exon |
| FUS | Arg522Gly | R522G | Substitution | AGG | GGG | Arg | Gly | 522 | exon |
| FUS | Arg522Arg | R522R | Substitution | G | A | Arg | Arg | 522 | exon |
| FUS | Arg524Trp | R524W | Substitution | A | T | Arg | Trp | 524 | exon |
| FUS | Arg524Thr | R524T | Substitution | AGG | ACG | Arg | Thr | 524 | exon |
| FUS | Arg524Ser | R524S | Substitution | AGG | AGC | Arg | Ser | 524 | exon |
| FUS | Arg524Met | R524M | Substitution | G | T | Arg | Met | 524 | exon |
| FUS | Pro525Leu | P525L | Substitution | CCG | CTG | Pro | Leu | 525 | exon |
| FUS | c.1581delA | X527YextX | Deletion | A |  | A |  | 527 | exon |
| GLE1 | c.1965-2A>C | c.1965-2A>C | Splice-site | A | C |  |  |  | intron |
| GLE1 | Ser70STOP | S70X | Deletion | C | A | Ser | STOP | 70 | exon |
| GLE1 | Arg697Cys | R697C | Substitution | C | T | Arg | Cys | 697 | exon |
| GRB14 | His507Tyr | H507Y | Substitution | C | T | His | Tyr | 507 | exon |
| HNRNPA1 | Asp314Asn | D314N | Substitution |  |  | Asp | Asn | 314 | exon |
| HNRNPA1 | Asn319Ser | N319S | Substitution |  |  | Asn | Ser | 319 | exon |
| LMNB1 | Ala436Val | A436V | Substitution | C | T | Ala | Val | 436 | exon |
| LUM | Leu199Pro | L199P | Substitution | T | C | Leu | Pro | 199 | exon |
| MATR3 | Ser85Cys | S85C | Substitution | C | G | Ser | Cys | 85 | exon |
| MATR3 | Phe115Cys | F115C | Substitution | T | G | Phe | Cys | 115 | exon |
| MATR3 | Pro154Ser | P154S | Substitution | C | T | Pro | Ser | 154 | exon |
| MATR3 | Thr622Ala | T622A | Substitution | A | G | Thr | Ala | 622 | exon |
| NEFH | 2368-2370del | 2368-2370del | Deletion |  |  |  |  |  | exon |
| NEFH | Ala40Val | A152V | Substitution | C | T | Ala | Val | 40 | exon |
| NEFH | Arg346His | R346H | Substitution | G | A | Arg | His | 346 | exon |
| NEFH | 1582-1683del | A528delGCT..stop 987 | Deletion | gct at position +1 |  | Ala |  | 528 | exon |
| NEFH | 1965-1988del | P655delAGA..stop at 662 | Deletion | cca at position +3 |  | Pro |  | 655 | exon |
| NEFH | 1989-2006del | P663delTGAGAAGGCCAAGTCCCC | Deletion | cct at position +3 |  | Pro |  | 663 | exon |
| NEFH | 1989-2030del | P663delTGA..stop at 1007 | Deletion | cct at position +3 |  | Pro |  | 663 | exon |
| NEFH | 2080-2163ins | Ala708ins84bp | Insertion | gcc |  | Ala |  | 708 | exon |
| NEFH | 2230-2247del | A744delGCC..stop at 1015 | Deletion | gcc at position +1 |  | Ala |  | 744 | exon |
| NEFH | Lys867Asn | K867N | Substitution | G | T | Lys | Asn | 867 | exon |
| NEFH | Glu918Gly | E918G | Substitution | A | G | Glu | Gly | 918 | exon |
| NETO1 | Gly401Arg | G401R | Substitution | G | A | Gly | Arg | 401 | exon |
| NIPA1 | Ile81Thr | I81T | Substitution | T | C | Ile | Thr | 81 | exon |
| NIPA1 | Ala86Gly | A86G | Substitution |  |  | Ala | Gly | 86 | exon |
| NIPA1 | Ile120Met | I120M | Substitution |  |  | Ile | Met | 120 | exon |
| NIPA1 | Val162Met | V162M | Substitution |  |  | Val | Met | 162 | exon |
| NIPA1 | Met189Ile | M189I | Substitution |  |  | Met | Ile | 189 | exon |
| NIPA1 | Pro221Leu | P221L | Substitution | C | T | Pro | Leu | 221 | exon |
| NIPA1 | Arg281Gln | R281Q | Substitution |  |  | Arg | Gln | 281 | exon |
| OMA1 | His69Tyr | H69Y | Substitution | C | T | His | Tyr | 69 | exon |
| OMA1 | Glu272Gly | E272G | Substitution | A | G | Glu | Gly | 272 | exon |
| OMA1 | Asp365Tyr | D365Y | Substitution | G | T | Asp | Tyr | 365 | exon |
| OPTN | M98K/G291fs | M98K/G291fs | Frameshift |  |  |  |  |  | exon |
| OPTN | c.553-5C>T | c.553-5C>T | Splice-site |  |  |  |  |  | exon |
| OPTN | delExon5 | delExon5 | Substitution |  |  |  |  | 0 | exon |
| OPTN | Gly23STOP | G23X | Substitution | GGA | TGA | Gly | STOP | 23 | exon |
| OPTN | Thr34Thr | T34T | Substitution | G | A | Thr | Thr | 34 | exon |
| OPTN | Leu41Leu | L41L | Substitution | G | A | Leu | Leu | 41 | exon |
| OPTN | Lys59Asn | K59N | Substitution | AAG | AAC | Lys | Asn | 59 | exon |
| OPTN | Ala93Pro | A93P | Subtitution | G | C | Ala | Pro | 93 | exon |
| OPTN | Arg96Leu | R96L | Subtitution | G | T | Arg | Leu | 96 | exon |
| OPTN | Met98Lys | M98K | Substitution | T | A | Met | Lys | 98 | exon |
| OPTN | Leu100Leu | L100L | Silent |  |  | Leu | Leu | 100 | exon |
| OPTN | Ala134Ala | A134A | Silent |  |  | Ala | Ala | 134 | exon |
| OPTN | Arg149Arg | R149R | Substitution | G | A | Arg | Arg | 149 | exon |
| OPTN | Gly159Val | G159V | Substitution | GGC | GTC | Gly | Val | 159 | exon |
| OPTN | Glu163Glu | E163E | Substitution | A | G | Glu | Glu | 163 | exon |
| OPTN | Leu164Leu | L164L | Silent |  |  | Leu | Leu | 164 | exon |
| OPTN | Gln165STOP | Q165X | Substitution | C | T | Gln |  | 165 | exon |
| OPTN | Gly172Gly | G172G | Substitution | C | T | Gly | Gly | 172 | exon |
| OPTN | Ser218Ser | S218S | Silent | T | C | Ser | Ser | 218 | exon |
| OPTN | Arg271Cys | R271C | Subtitution | C | T | Arg | Cys | 271 | exon |
| OPTN | Thr282Pro | T282P | Substitution | ACA | CCA | Thr | Pro | 282 | exon |
| OPTN | Pro286Pro | P286P | Substitution | G | A | Pro | Pro | 286 | exon |
| OPTN | Gln314Leu | Q314L | Substitution | CAA | CTA | Gln | Leu | 314 | exon |
| OPTN | Lys316Glu | K316E | Substitution | G | A | Lys | Glu | 316 | exon |
| OPTN | Ser321Ser | S321S | Substitution | C | T | Ser | Ser | 321 | exon |
| OPTN | Glu322Lys | E322K | Substitution | G | A | Glu | Lys | 322 | exon |
| OPTN | del P359 | del P359 | Deletion |  |  |  |  | 359 | exon |
| OPTN | Leu369Leu | L369L | Substitution | A | G | Leu | Leu | 369 | exon |
| OPTN | Gln398STOP | Q398X | Substitution | CAA | TAA | Gln | STOP | 398 | exon |
| OPTN | 1242+1G>A\_insA | 1242+1G>A\_insA | Insertion |  |  |  |  | 414 | exon |
| OPTN | Lys440Asnfs\*8 | K440Nfs\*8 | Substitution |  |  | Lys | Asn | 440 | exon |
| OPTN | Gln454Glu | Q454E | Substitution | C | G | Gln | Glu | 454 | exon |
| OPTN | c.1401+4A/G | c.1401+4A/G | Substitution | A | G | A | G | 467 | exon |
| OPTN | Glu478Gly | E478G | Substitution | GAA | GGA | Glu | Gly | 478 | exon |
| OPTN | Ala481Val | A481V | Substitution | GCG | GTG | Ala | Val | 481 | exon |
| OPTN | Lys557Thr | K557T | Substitution | AAG | ACG | Lys | Thr | 557 | exon |
| OPTN | c.552+1delG | c.552+1delG | Substitution | G |  | Lys | Thr | 557 | exon |
| OPTN | Leu568Leu | L568L | Substitution | A | G | Leu | Leu | 568 | exon |
| OPTN | c.382\_383insAG | 691\_692insAG | Insertion |  | AG |  | AG | 691 | exon |
| PARK7 | Glu45STOP | E45X | Substitution |  |  | Glu | STOP | 45 | exon |
| PCP4 | Ala58Ser | A58S | Substitution | G | T | Ala | Ser | 58 | exon |
| PFN1 | Gly15Gly | G15G | Substitution | G | C | Gly | Gly | 15 | exon |
| PFN1 | Thr16Thr | T16T | Substitution | C | A | Thr | Thr | 16 | exon |
| PFN1 | Ala20Thr | A20T | Substitution | G | A | Ala | Thr | 20 | exon |
| PFN1 | Cys71Gly | C71G | Substitution | T | G | Cys | Gly | 71 | exon |
| PFN1 | Leu88Leu | L88L | Substitution | T | C | Leu | Leu | 88 | exon |
| PFN1 | Thr109Met | T109M | Substitution | C | T | Thr | Met | 109 | exon |
| PFN1 | Leu112Leu | L112L | Substitution | C | T | Leu | Leu | 112 | exon |
| PFN1 | Met114Thr | M114T | Substitution | T | C | Met | Thr | 114 | exon |
| PFN1 | Glu117Gly | E117G | Substitution | AA | GT | Glu | Gly | 117 | exon |
| PFN1 | Gly118Val | G118V | Substitution | G | T | Gly | Val | 118 | exon |
| PFN1 | Arg136Trp | R136W | Substitution | C | T | Arg | Trp | 136 | exon |
| PFN1 | Glu139Leu | E139L | Substitution | A | T | Glu | Leu | 139 | exon |
| PLEKHG5 | Pro630His | P630H | Substitution |  |  | Pro | His | 630 | exon |
| RAMP3 | Leu111Ser | L111S | Substitution | T | C | Leu | Ser | 111 | exon |
| RAMP3 | Glu117Lys | E117K | Substitution | G | A | Glu | Lys | 117 | exon |
| RNASE2 | His100Gln | H100Q | Substitution | C | A | His | Gln | 100 | exon |
| SETX | Thr3Ile | T3I | Substitution | ACA | ATA | Thr | Ile | 3 | exon |
| SETX | Leu389Ser | L389S | Substitution | TTA | TCA | Leu | Ser | 389 | exon |
| SETX | Thr1118Ile | T1118I | Substitution | ACA | ATA | Thr | Ile | 1118 | exon |
| SETX | Cys1554Gly | C1554G | Substitution | T | G | Cys | Gly | 1554 | exon |
| SETX | Lys2018Glu | K2018E | Substitution | AAA | GAA | Lys | Glu | 2018 | exon |
| SETX | Lys2029Glu | K2029E | Substitution | C | G | Lys | Glu | 2029 | exon |
| SETX | Arg2136His | R2136H | Substitution | CGC | CAC | Arg | His | 2136 | exon |
| SETX | Ile2547Thr | I2547T | Substitution | T | C | Ile | Thr | 2547 | exon |
| SIGMAR1 | c.\*58T>C | c.\*58T>C | Substitution | T | C |  |  |  | exon |
| SIGMAR1 | Glu102Gln | E102Q | Substitution | G | C | Glu | Gln | 102 | exon |
| SOD1 | IVS3+62 | IVS3+62 | Insertion |  |  | T | C |  | intron |
| SOD1 | IVS3+34 | IVS3+34 | Insertion |  |  | A | C |  | intron |
| SOD1 | InsAexon2 | Ins-A-exon2 | Insertion | AGTGAA | AGATGAA |  |  |  | exon |
| SOD1 | T-G-intron4-10bp | t - g 10 bp before Exon 5 | Substitution | t | g |  |  |  | intron |
| SOD1 | T-A-intron3-108bp | t - a 108 bp before Exon 2 | Substitution | t | a |  |  |  | intron |
| SOD1 | Lys3Glu | K3E | Substitution | AAG | GAG | Lys | Glu | 3 | exon |
| SOD1 | Ala4Thr | A4T | Substitution | GCC | ACC | Ala | Thr | 4 | exon |
| SOD1 | Ala4Val | A4V | Substitution | GCC | GTC | Ala | Val | 4 | exon |
| SOD1 | Ala4Ser | A4S | Substitution | GCC | TCC | Ala | Ser | 4 | exon |
| SOD1 | Ala4Phe | A4F | Substitution | GCC | TTC | Ala | Phe | 4 | exon |
| SOD1 | Val5Leu | V5L | Substitution | GTG | TTG | Val | Leu | 5 | exon |
| SOD1 | Cys6Gly | C6G | Substitution | TGC | GGC | Cys | Gly | 6 | exon |
| SOD1 | Cys6Phe | C6F | Substitution | TGC | TTT | Cys | Phe | 6 | exon |
| SOD1 | Cys6Ser | C6S | Substitution | TGC | AGC | Cys | Ser | 6 | exon |
| SOD1 | Cys6Trp | C6W | Substitution | TGC | TGG | Cys | Trp | 6 | exon |
| SOD1 | Val7Glu | V7E | Substitution | GTG | GAG | Val | Glu | 7 | exon |
| SOD1 | Leu8Gln | L8Q | Substitution | CTG | CAG | Leu | Gln | 8 | exon |
| SOD1 | Leu8Val | L8V | Substitution | CTG | GTG | Leu | Val | 8 | exon |
| SOD1 | Gly10Gly | G10G | Polymorphism | GGC | GGT | Gly | Gly | 10 | exon |
| SOD1 | Gly10Val | G10V | Substitution | GGC | GTT | Gly | Val | 10 | exon |
| SOD1 | Gly10Arg | G10R | Substitution | GGC | CGC | Gly | Arg | 10 | exon |
| SOD1 | Asp11Tyr | D11Y | Substitution | GAC | TAC | Asp | Tyr | 11 | exon |
| SOD1 | Asp11Ala | D11A | Substitution | GAC | GCC | Asp | Ala | 11 | exon |
| SOD1 | Gly12Arg | G12R | Substitution | GGC | CGC | Gly | Arg | 12 | exon |
| SOD1 | Val14Met | V14M | Substitution | GTG | ATG | Val | Met | 14 | exon |
| SOD1 | Val14Gly | V14G | Substitution | GTG | GGG | Val | Gly | 14 | exon |
| SOD1 | Gly16Ala | G16A | Substitution | GGC | GCC | Gly | Ala | 16 | exon |
| SOD1 | Gly16Ser | G16S | Substitution | GGC | AGC | Gly | Ser | 16 | exon |
| SOD1 | Asn19Ser | N19S | Substitution | AAT | AGT | Asn | Ser | 19 | exon |
| SOD1 | Phe20Cys | F20C | Substitution | TTC | TGC | Phe | Cys | 20 | exon |
| SOD1 | Glu21Lys | E21K | Substitution | GAG | AAG | Glu | Lys | 21 | exon |
| SOD1 | Glu21Gly | E21G | Substitution | GAG | GGG | Glu | Gly | 21 | exon |
| SOD1 | Gln22Leu | Q22L | Substitution | CAG | CTG | Gln | Leu | 22 | exon |
| SOD1 | Gln22Arg | Q22R | Substitution | CAG | CGG | Gln | Arg | 22 | exon |
| SOD1 | Gly27delGGACCA | G27delGGACCA | Deletion | GGA | GACCAGTG | Gly |  | 27 | exon |
| SOD1 | Val29insA | V29insA | Insertion | GTG | A | Val |  | 29 | exon |
| SOD1 | Val29Ala | V29A | Substitution | GTG | GCG | Val | Ala | 29 | exon |
| SOD1 | Val31Ala | V31A | Substitution | GTG | GCG | Val | Ala | 31 | exon |
| SOD1 | Gly37Val | G37V | Substitution | GGA | GTA | Gly | Val | 37 | exon |
| SOD1 | Gly37Arg | G37R | Substitution | GGA | AGA | Gly | Arg | 37 | exon |
| SOD1 | Leu38Arg | L38R | Substitution | CTG | CGG | Leu | Arg | 38 | exon |
| SOD1 | Leu38Val | L38V | Substitution | CTG | GTG | Leu | Val | 38 | exon |
| SOD1 | Glu40Gly | E40G | Substitution | GAA | GGA | Gliu | Gly | 40 | exon |
| SOD1 | Glu41Gly | E41G | Substitution | A | G | Glu | Gly | 41 | exon |
| SOD1 | Gly41Ser | G41S | Substitution | GGC | AGC | Gly | Ser | 41 | exon |
| SOD1 | Gly41Asp | G41D | Substitution | GGC | GAC | Gly | Asp | 41 | exon |
| SOD1 | His43Arg | H43R | Substitution | CAT | CGT | His | Arg | 43 | exon |
| SOD1 | Phe45Cys | F45C | Substitution | TTC | TGC | Phe | Cys | 45 | exon |
| SOD1 | Phe45Ser | F45S | Substitution | T | C | Phe | Ser | 45 | exon |
| SOD1 | His46Arg | H46R | Substitution | CAT | CGT | His | Arg | 46 | exon |
| SOD1 | His46Asp | H46D | Substitution | CAT | GAT | His | Asp | 46 | exon |
| SOD1 | Val47Phe | V47F | Substitution | GTT | TTT | Val | Phe | 47 | exon |
| SOD1 | Val47Ala | V47A | Substitution | GTT | GCT | Val | Ala | 47 | exon |
| SOD1 | His48Arg | H48R | Substitution | CAT | CGT | His | Arg | 48 | exon |
| SOD1 | His48Gln | H48Q | Substitution | CAT | CAG | His | Gln | 48 | exon |
| SOD1 | Glu49Lys | E49K | Substitution | GAG | AAG | Glu | Lys | 49 | exon |
| SOD1 | Thr54Arg | T54R | Substitution | ACA | AGA | Thr | Arg | 54 | exon |
| SOD1 | Cys57Arg | C57R | Substitution | TGT | CGT | Cys | Arg | 57 | exon |
| SOD1 | Ser59Ser | S59S | Polymorphism | AGT | AGC | Ser | Ser | 59 | exon |
| SOD1 | Ser59Ile | S59I | Substitution | AGT | ATT | Ser | Ile | 59 | exon |
| SOD1 | Gly61Arg | G61R | Substitution | GGT | CGT | Gly | Arg | 61 | exon |
| SOD1 | Phe64Leu | F64L | Substitution | TTT | CTT | Phe | Leu | 64 | exon |
| SOD1 | Asn65Ser | N65S | Substitution | AAT | AGT | Asn | Ser | 65 | exon |
| SOD1 | Pro66Ala | P66A | Substitution | CCT | GCT | Pro | Ala | 66 | exon |
| SOD1 | Pro66Ser | P66S | Substitution | CCT | TCT | Pro | Ser | 66 | exon |
| SOD1 | Pro66Arg | P66R | Substitution | CCT | CGT | Pro | Arg | 66 | exon |
| SOD1 | Leu67Pro | L67P | Substitution | CTA | CCA | Leu | Pro | 67 | exon |
| SOD1 | Leu67Arg | L67R | Substitution | CTA | CGA | Leu | Arg | 67 | exon |
| SOD1 | His71Tyr | H71Y | Substitution | C | T | His | Tyr | 71 | exon |
| SOD1 | Gly72Ser | G72S | Substitution | GGT | AGT | Gly | Ser | 72 | exon |
| SOD1 | Gly72Cys | G72C | Substitution | GGT | TGT | Gly | Cys | 72 | exon |
| SOD1 | Asp76Tyr | D76Y | Substitution | GAT | TAT | Asp | Tyr | 76 | exon |
| SOD1 | Asp76Val | D76V | Substitution | GAT | GTT | Asp | Val | 76 | exon |
| SOD1 | His80Arg | H80R | Substitution | CAT | CGT | His | Arg | 80 | exon |
| SOD1 | Asp83Gly | D83G | Substitution | GAC | GGC | Asp | Gly | 83 | exon |
| SOD1 | Leu84Phe | L84F | Substitution | TTG | TTC | Leu | Phe | 84 | exon |
| SOD1 | Leu84Val | L84V | Substitution | TTG | GTG | Leu | Val | 84 | exon |
| SOD1 | Leu84Phe | L84F | Substitution | TTG | TTT | Leu | Phe | 84 | exon |
| SOD1 | Gly85Arg | G85R | Substitution | GGC | CGC | Gly | Arg | 85 | exon |
| SOD1 | Gly85Ser | G85S | Substitution | GGC | AGC | Gly | Ser | 85 | exon |
| SOD1 | Asn86Ile | N86I | Substitution | AAT | ATT | Asn | Ile | 86 | exon |
| SOD1 | Asn86Ser | N86S | Substitution | AAT | AGT | Asn | Ser | 86 | exon |
| SOD1 | Asn86Asp | N86D | Substitution | AAT | GAT | Asn | Asp | 86 | exon |
| SOD1 | Asn86Lys | N86K | Substitution | AAT | AAA | Asn | Lys | 86 | exon |
| SOD1 | Val87Ala | V87A | Substitution | GTG | GCG | Val | Ala | 87 | exon |
| SOD1 | Val87Met | V87M | Substitution | GTG | ATG | Val | Met | 87 | exon |
| SOD1 | Thr88delTAD | T88delTAD | Deletion | act at position +1 |  | Thr |  | 88 | exon |
| SOD1 | Ala89Thr | A89T | Substitution | GCT | ACT | Ala | Thr | 89 | exon |
| SOD1 | Ala89Val | A89V | Substitution | GCT | GTT | Ala | Val | 89 | exon |
| SOD1 | Asp90Ala,Asp96Asn | D90A,D96N | Compound |  |  |  |  | 90 |  |
| SOD1 | Asp90Val | D90V | Substitution | GAC | GTC | Asp | Val | 90 | exon |
| SOD1 | Asp90Ala | D90A | Substitution | GAC | GCC | Asp | Ala | 90 | exon |
| SOD1 | Gly93Val | G93V | Substitution | GGT | GTT | Gly | Val | 93 | exon |
| SOD1 | Gly93Ala | G93A | Substitution | GGT | GCT | Gly | Ala | 93 | exon |
| SOD1 | Gly93Cys | G93C | Substitution | GGT | TGT | Gly | Cys | 93 | exon |
| SOD1 | Gly93Arg | G93R | Substitution | GGT | CGT | Gly | Arg | 93 | exon |
| SOD1 | Gly93Asp | G93D | Substitution | GGT | GAT | Gly | Asp | 93 | exon |
| SOD1 | Gly93Ser | G93S | Substitution | GGT | AGT | Gly | Ser | 93 | exon |
| SOD1 | Val94Ala | V94A | Substitution | GTG | GCG | Val | Ala | 94 | exon |
| SOD1 | Ala95Val | A95V | Substitution | GCC | GTC | Ala | Val | 95 | exon |
| SOD1 | Ala95Thr | A95T | Substitution | GCC | ACC | Ala | Thr | 95 | exon |
| SOD1 | Ala95Gly | A95G | Substitution | GCC | GGC | Ala | Gly | 95 | exon |
| SOD1 | Asp96Metfs8 | D96Mfs8 | Frameshift |  |  | Asp | Met | 96 | exon |
| SOD1 | Asp96Val | D96V | Substitution | GAT | GTT | Asp | Val | 96 | exon |
| SOD1 | Asp96Asn | D96N | Substitution | GAT | AAT | Asp | Asn | 96 | exon |
| SOD1 | Val97Met | V97M | Substitution | GTG | ATG | Val | Met | 97 | exon |
| SOD1 | Val97Leu | V97L | Substitution | GTG | CTG | Val | Leu | 97 | exon |
| SOD1 | Ile99Val | I99V | Substitution | ATT | GTT | Ile | Val | 99 | exon |
| SOD1 | Glu100Gly | E100G | Substitution | GAA | GGA | Glu | Gly | 100 | exon |
| SOD1 | Glu100Lys | E100K | Substitution | GAA | AAA | Glu | Lys | 100 | exon |
| SOD1 | Asp101Gly | D101G | Substitution | GAT | GGT | Asp | Gly | 101 | exon |
| SOD1 | Asp101Asn | D101N | Substitution | GAT | AAT | Asp | Asn | 101 | exon |
| SOD1 | Asp101Tyr | D101Y | Substitution | GAT | TAT | Asp | Tyr | 101 | exon |
| SOD1 | Asp101His | D101H | Substitution | GAT | CAT | Asp | His | 101 | exon |
| SOD1 | Ile104Phe | I104F | Substitution | ATC | TTC | Ile | Phe | 104 | exon |
| SOD1 | Ser105delTCACTC | S105del SL | Deletion | tca at position +1 |  | Ser |  | 105 | exon |
| SOD1 | Ser105Leu | S105L | Substitution | TCA | TTA | Ser | Leu | 105 | exon |
| SOD1 | Leu106Val | L106V | Substitution | CTC | GTC | Leu | Val | 106 | exon |
| SOD1 | Leu106Phe | L106F | Substitution | CTC | TTC | Leu | Phe | 106 | exon |
| SOD1 | Gly108Val | G108V | Substitution | GGA | GTA | Gly | Val | 108 | exon |
| SOD1 | Asp109Tyr | D109Y | Substitution | GAC | TAC | Asp | Tyr | 109 | exon |
| SOD1 | Cys111Tyr | C111Y | Substitution | TGC | TAC | Cys | Tyr | 111 | exon |
| SOD1 | Ile112Thr | I112T | Substitution | ATC | ACC | Ile | Thr | 112 | exon |
| SOD1 | Ile112Met | I112M | Substitution | ATC | ATG | Ile | Met | 112 | exon |
| SOD1 | Ile113Thr | I113T | Substitution | ATT | ACT | Ile | Thr | 113 | exon |
| SOD1 | Ile113Phe | I113F | Substitution | ATT | TTT | Ile | Phe | 113 | exon |
| SOD1 | Gly114Ala | G114A | Substitution | GGC | GCC | Gly | Ala | 114 | exon |
| SOD1 | Arg115Gly | R115G | Substitution | CGC | GGC | Arg | Gly | 115 | exon |
| SOD1 | Arg115Cys | R115C | Substitution | CGC | TGC | Arg | Cys | 115 | exon |
| SOD1 | Thr116Thr | T116T | Polymorphism | ACA | ACG | Thr | Thr | 116 | exon |
| SOD1 | Thr116Arg | T116R | Substitution | ACA | AGA | Thr | Arg | 116 | exon |
| SOD1 | Leu117Val | L117V | Substitution | CTG | GTG | Leu | Val | 117 | exon |
| SOD1 | Val118insAAAAC | V118insAAAAC | Insertion | GTG | AAAAC | Val |  | 118 | exon |
| SOD1 | Val118Leu | V118L | Substitution | GTG | TTG | Val | Leu | 118 | exon |
| SOD1 | Val118Leu | V118L | Substitution | GTG | CTG | Val | Leu | 118 | exon |
| SOD1 | Val118Met | V118M | Substitution | GTG | ATG | Val | Met | 118 | exon |
| SOD1 | His120Leu | H120L | Substitution | CAT | CTT | His | Leu | 120 | exon |
| SOD1 | Glu121Gly | E121G | Substitution | GAA | GGA | Glu | Gly | 121 | exon |
| SOD1 | Asp124Val | D124V | Substitution | GAT | GTT | Asp | Val | 124 | exon |
| SOD1 | Asp124Gly | D124G | Substitution | GAT | GGT | Asp | Gly | 124 | exon |
| SOD1 | Asp125delTT | D125delTT | Deletion | GAC | TT | Asp |  | 125 | exon |
| SOD1 | Asp125His | D125H | Substitution | GAC | CAC | Asp | His | 125 | exon |
| SOD1 | Leu126delTT | L126delTT stop at 131 | Deletion | TTG | \*\*G | Leu |  | 126 | exon |
| SOD1 | Leu126Ser | L126S | Substitution | TTG | TCG | Leu | Ser | 126 | exon |
| SOD1 | Leu126STOP | L126Z | Substitution | TTG | TAG | Leu | STOP | 126 | exon |
| SOD1 | Gly127insTGGG | G127insTGGG | Insertion | GGC | TGGG | Gly |  | 127 | exon |
| SOD1 | Gly127Arg | G127R | Substitution | GGC | CGC | Gly | Arg | 127 | exon |
| SOD1 | Glu133delGAA | E133delGAA | Deletion | GAA | \*\*\* | Glu |  | 132 | exon |
| SOD1 | Glu132insTT | E132insTT | Insertion | GAA | insTT | Glu |  | 132 | exon |
| SOD1 | Glu132Lys | E132K | Substitution | GAA | AAA | Glu | Lys | 132 | exon |
| SOD1 | Glu133del | E133delGAA | Deletion | GAA | GAA | Glu |  | 133 | exon |
| SOD1 | Glu133insTT | E133insTT | Insertion | GAA | TT | Glu |  | 133 | exon |
| SOD1 | Glu133Val | E133V | Substitution | GAA | GTA | Glu | Val | 133 | exon |
| SOD1 | Ser134Asn | S134N | Substitution | AGT | AAT | Ser | Asn | 134 | exon |
| SOD1 | Ser134Thr | S134T | Substitution | AGT | ACT | Ser | Thr | 134 | exon |
| SOD1 | Thr137Ala | T137A | Substitution | ACA | GCA | Thr | Ala | 137 | exon |
| SOD1 | Thr137Arg | T137R | Substitution | ACA | AGA | Thr | Arg | 137 | exon |
| SOD1 | Gly138Glu | G138E | Substitution | GGA | GAA | Gly | Glu | 138 | exon |
| SOD1 | Asn139Asn | N139N | Polymorphism | AAC | AAT | Asn | Asn | 139 | exon |
| SOD1 | Asn139His | N139H | Substitution | AAC | CAC | Asn | His | 139 | exon |
| SOD1 | Asn139Lys | N139K | Substitution | AAC | AAA | Asn | Lys | 139 | exon |
| SOD1 | Asn139Asp | N139D | Substitution | AAC | GAC | Asn | Asp | 139 | exon |
| SOD1 | Ala140Ala | A140A | Polymorphism | GCT | GCA | Ala | Ala | 140 | exon |
| SOD1 | Ala140Gly | A140G | Substitution | GCT | GGT | Ala | Gly | 140 | exon |
| SOD1 | Gly141STOP | G141X | Substitution | GGA | TGA | Gly | STOP | 141 | exon |
| SOD1 | Gly141Glu | G141E | Substitution | GGA | GAA | Gly | Glu | 141 | exon |
| SOD1 | Gly141Ala | G141A | Substitution | GGA | GCA | Gly | Ala | 141 | exon |
| SOD1 | Leu144Phe | L144F | Substitution | TTG | TTC | Leu | Phe | 144 | exon |
| SOD1 | Leu144Ser | L144S | Substitution | TTG | TCG | Leu | Ser | 144 | exon |
| SOD1 | Ala145Thr | A145T | Substitution | GCT | ACT | Ala | Thr | 145 | exon |
| SOD1 | Ala145Gly | A145G | Substitution | GCT | GGT | Ala | Gly | 145 | exon |
| SOD1 | Cys146Arg | C146R | Substitution | TGT | CGT | Cys | Arg | 146 | exon |
| SOD1 | Cys146STOP | C146X | Substitution | TGT | TGA | Cys | STOP | 146 | exon |
| SOD1 | Gly147Asp | G147D | Substitution | GGT | GAT | Gly | Asp | 147 | exon |
| SOD1 | Gly147Arg | G147R | Substitution | GGT | CGT | Gly | Arg | 147 | exon |
| SOD1 | Gly147Ser | G147S | Substitution | GGT | AGT | Gly | Ser | 147 | exon |
| SOD1 | Val148Gly | V148G | Substitution | GTA | GGA | Val | Gly | 148 | exon |
| SOD1 | Val148Ile | V148I | Substitution | GTA | ATA | Val | Ile | 148 | exon |
| SOD1 | Ile149Thr | I149T | Substitution | ATT | ACT | Ile | Thr | 149 | exon |
| SOD1 | Ile149Thr | I149T | Substitution | T | C | Ile | Thr | 149 | exon |
| SOD1 | Ile151Thr | I151T | Substitution | ATC | ACC | Ile | Thr | 151 | exon |
| SOD1 | Ile151Ser | I151S | Substitution | ATC | AGC | Ile | Ser | 151 | exon |
| SOD1 | Gln153Gln | Q153Q | Polymorphism | CAA | CAG | Gln | Gln | 153 | exon |
| SOX5 | Gln362Pro | Q362P | Substitution | A | C | Gln | Pro | 362 | exon |
| SPG11 | IVS30+1G>A | IVS30+1G>A | Substitution |  |  |  |  |  | intron |
| SPG11 | N1962S/Y2272X | N1962S/Y2272X | Substitution |  |  |  |  |  | exon |
| SPG11 | Phe2265Leu | F2265L | Substitution |  |  |  |  |  | exon |
| SPG11 | Gln40X | Q40X | Substitution | C | T | Gln | STOP | 40 | exon |
| SPG11 | Trp89X | W89X | Substitution | G | A | Trp | STOP | 89 | exon |
| SPG11 | Met245Valfs | 733\_734delAT | Deletion |  |  | Met | Val | 245 | exon |
| SPG11 | Leu733X | L733X | Substitution | T | G | Leu | STOP | 733 | exon |
| SPG11 | Ile870Valfs | I870V | frameshift | A | G | Ile | Val | 870 | exon |
| SPG11 | Arg1026fs | 3076\_3077insA | Insertion |  |  | Arg |  | 1026 | exon |
| SPG11 | Val1468Leufs | 4461\_4462delGT | Deletion |  |  | Val | Leu | 1468 | exon |
| SPG11 | Tyr1990X | Y1990X | Substitution | C | G | Tyr | STOP | 1990 | exon |
| SPG11 | Arg1992X | R1992X | Substitution | C | T | Arg | STOP | 1992 | exon |
| SPG11 | Val2053Met | V2053M | Substitution | G | A | Val | Met | 2053 | exon |
| SPG11 | Val2344Cysfs | 7029\_7030insT | Insertion |  |  | Val | Cys | 2344 | exon |
| SQSTM1 | g.3'+7G>C | g.3'+7G>C | Intronic | G | C |  |  |  | intron |
| SQSTM1 | g.5'-37C>T | g.5'-37C>T | Intronic | C | T |  |  |  | intron |
| SQSTM1 | Ala33Val | A33V | Substitution | C | T | Ala | Val | 33 | exon |
| SQSTM1 | Ala53Thr | A53T | Substitution |  |  | Ala | Thr | 53 | exon |
| SQSTM1 | Val153Ile | V153I | Substitution | G | A | Val | Ile | 153 | exon |
| SQSTM1 | Pro228Leu | P228L | Substitution | C | T | Pro | Leu | 228 | exon |
| SQSTM1 | Val234Val | V234V | Substitution | G | A | Val | Val | 234 | exon |
| SQSTM1 | Lys238delGAA | c.714-716delGAA | Deletion | GAA |  | Lys |  | 238 | exon |
| SQSTM1 | His261His | H261H | Substitution | C | T | His | His | 261 | exon |
| SQSTM1 | Glu274Asp | E274D | Substitution |  |  |  |  | 274 | exon |
| SQSTM1 | Ser318Pro | S318P | Substitution | T | C | Ser | Pro | 318 | exon |
| SQSTM1 | Arg321Cys | R321C | Substitution | C | T | Arg | Cys | 321 | exon |
| SQSTM1 | Ser370Pro | S370P | Substitution | T | C | Ser | Pro | 370 | exon |
| SQSTM1 | Pro392Leu | P392L | Substitution | C | T | Pro | Leu | 392 | exon |
| SQSTM1 | Gly411Ser | G411S | Substitution | G | A | Gly | Ser | 411 | exon |
| SQSTM1 | Gly425Arg | G425R | Substitution | G | A | Gly | Arg | 425 | exon |
| SQSTM1 | Pro439Leu | P439L | Substitution |  |  | Pro | Leu | 439 | exon |
| SS18L1 | Ileu123Met | I123M | Substitution | T | G | Ile | Met | 123 | exon |
| SS18L1 | Gln222\_Ser224del | Q222\_S224del | Deletion |  |  |  |  | 222 | exon |
| SS18L1 | Ala264Thr | A264T | Substitution | G | A |  |  | 264 | exon |
| SYNE | Glu7644STOP | E7644X | Substitution |  |  | Glu | STOP | 7644 | exon |
| SYNE | Arg7842STOP | R7842X | Substitution |  |  | Arg | STOP | 7842 | exon |
| SYT9 | Val154Met | V154M | Substitution | G | A | Val | Met | 154 | exon |
| SYT9 | Ile238Val | I238V | Substitution | A | G | Ile | Val | 238 | exon |
| SYT9 | Leu353Val | L353V | Substitution | C | G | Leu | Val | 353 | exon |
| TAF15 | Ala31Thr | A31T | Subtitution | G | A | Ala | Thr | 31 | exon |
| TAF15 | Gly239Gly | G239G | Subtitution | G | A | Gly | Gly | 239 | exon |
| TAF15 | Asp386Asn | D386N | Subtitution | G | A | Asp | Asn | 386 | exon |
| TAF15 | Arg388His | R388H | Subtitution | G | A | Arg | His | 388 | exon |
| TAF15 | Arg395Gln | R395Q | Subtitution | G | A | Arg | Gln | 395 | exon |
| TAF15 | c.1392\_1415del | G464\_G471del | Deletion |  |  |  |  | 464 | exon |
| TAF15 | c.1422\_1445del | R474\_D481del | Deletion |  |  |  |  | 474 | exon |
| TARDBP | 5' UTR | c.-69C>T | Polymorphism | C | T |  |  | 0 | exon |
| TARDBP | 5' UTR | c.-66G>T | Polymorphism | G | T |  |  | 0 | exon |
| TARDBP | p.Leu27 | c.81G>A | Polymorphism | G | A |  |  | 0 | intron |
| TARDBP | c.-12-54G>A | c.-12-54G>A | Polymorphism | G | A |  |  | 0 | intron |
| TARDBP | p.Ala66 | c.81G>A | Polymorphism | T | C |  |  | 0 | intron |
| TARDBP | p.Ser104 | c.312C>T | Polymorphism | G | A |  |  | 0 | intron |
| TARDBP | p.Lys137 | c.411A>G | Polymorphism | A | G |  |  | 0 | intron |
| TARDBP | c.403-80G>A | c.403-80G>A | Polymorphism | G | A |  |  | 0 | intron |
| TARDBP | c.543+112C>A | c.543+112C>A | Polymorphism | C | A |  |  | 0 | intron |
| TARDBP | Gly40Gly | G40G | Polymorphism | GGG | GGA | Gly | Gly | 40 | exon |
| TARDBP | Ala66Ala | A66A | Substitution | GCT | GCC | Ala | Ala | 66 | exon |
| TARDBP | Ala90Val | A90V | Substitution | GCT | GTT | Ala | Val | 90 | exon |
| TARDBP | Asp169Gly | D169G | Substitution | GAT | GGT | Asp | Gly | 169 | exon |
| TARDBP | Asn267Ser | N267S | Substitution | AAT | AGT | Asn | Ser | 267 | exon |
| TARDBP | Gly287Ser | G287S | Substitution | GGT | AGT | Gly | Ser | 287 | exon |
| TARDBP | Gly290Ala | G290A | Substitution | GGT | GCT | Gly | Ala | 290 | exon |
| TARDBP | Ser292Asn | S292N | Substitution | AGC | AAC | Ser | Asn | 292 | exon |
| TARDBP | Gly294Ala | G294A | Substitution | GGG | GCG | Gly | Ala | 294 | exon |
| TARDBP | Gly294Val | G294V | Substitution | GGG | GTG | Gly | Val | 294 | exon |
| TARDBP | Gly295Arg | G295R | Substitution | GGT | CGT | Gly | Arg | 295 | exon |
| TARDBP | Gly295Ser | G295S | Substitution | GGT | AGT | Gly | Ser | 295 | exon |
| TARDBP | Gly298Ser | G298S | Substitution | GGT | AGT | Gly | Ser | 298 | exon |
| TARDBP | Met311Val | M311V | Substitution | ATG | GTG | Met | Val | 311 | exon |
| TARDBP | Ala315Thr | A315T | Substitution | GCG | ACG | Ala | Thr | 315 | exon |
| TARDBP | Ala321Gly | A321G | Substitution | GCC | GGC | Ala | Gly | 321 | exon |
| TARDBP | Ala321Val | A321V | Substitution | GCC | GTC | Ala | Val | 321 | exon |
| TARDBP | Gln331Lys | Q331K | Substitution | CAG | AAG | Gln | Lys | 331 | exon |
| TARDBP | Ser332Asn | S332N | Substitution | AGC | AAC | Ser | Asn | 332 | exon |
| TARDBP | Gly335Asp | G335D | Substitution | GGT | GAT | Gly | Asp | 335 | exon |
| TARDBP | Met337Val | M337V | Substitution | ATG | GTG | Met | Val | 337 | exon |
| TARDBP | Gln343Arg | Q343R | Substitution | CAG | CGG | Gln | Arg | 343 | exon |
| TARDBP | Asn345Lys | N345K | Substitution | AAC | AAA | Asn | Lys | 345 | exon |
| TARDBP | Gly348Cys | G348C | Substitution | GGC | TGC | Gly | Cys | 348 | exon |
| TARDBP | Gly348Val | G348V | Substitution | GGC | GTC | Gly | Val | 348 | exon |
| TARDBP | Asn352Ser | N352S | Substitution | AAT | AGT | Asn | Ser | 352 | exon |
| TARDBP | Met359Val | M359V | Substitution | ATG | GTG | Met | Val | 359 | exon |
| TARDBP | Arg361Ser | R361S | Substitution | AGG | AGT | Arg | Ser | 361 | exon |
| TARDBP | Pro363Ala | P363A | Substitution | CCA | GCA | Pro | Ala | 363 | exon |
| TARDBP | Ala366Ala | A366A | Polymorphism | GCC | GCG | Ala | Ala | 366 | exon |
| TARDBP | Tyr374STOP | Y374X | Substitution | TAT | TAA | Tyr | STOP | 374 | exon |
| TARDBP | Gly376Asp | G376D | Substitution | GGC | GAC | Gly | Asp | 376 | exon |
| TARDBP | Asn378Asp | N378D | Substitution | AAT | GAT | Asn | Asp | 378 | exon |
| TARDBP | Ser379Pro | S379P | Substitution | TCT | CCT | Ser | Pro | 379 | exon |
| TARDBP | Ser379Cys | S379C | Substitution | TCT | TGT | Ser | Cys | 379 | exon |
| TARDBP | Ala382Thr | A382T | Substitution | GCA | ACA | Ala | Thr | 382 | exon |
| TARDBP | Ala382Pro | A382P | Substitution | GCA | CCA | Ala | Pro | 382 | exon |
| TARDBP | Ile383Val | I383V | Substitution | ATT | GTT | Ile | Val | 383 | exon |
| TARDBP | Ile383Thr | I383T | Substitution | ATT | ACT | Ile | Thr | 383 | exon |
| TARDBP | Gly384Arg | G384R | Substitution | GGT | CGT | Gly | Arg | 384 | exon |
| TARDBP | Trp385Gly | W385G | Substitution | TGG | GGG | Trp | Gly | 385 | exon |
| TARDBP | Asn390Asp | N390D | Substitution | AAT | GAT | Asn | Asp | 390 | exon |
| TARDBP | Asn390Ser | N390S | Substitution | AAT | AGT | Asn | Ser | 390 | exon |
| TARDBP | Ser393Leu | S393L | Substitution | TCG | TTG | Ser | Leu | 393 | exon |
| TBK1 | Arg47His | R47H | Substitution | G | A | Arg | His | 47 | exon |
| TBK1 | Thr77TrpfsX4 | T77WfsX4 | Frameshift | T | C | Thr | Trp | 77 | exon |
| TBK1 | Tyr105Cys | Y105C | Substitution | A | G | Tyr | Cys | 105 | exon |
| TBK1 | Tyr185X | Y185X | Frameshift | T | A | Tyr | STOP | 185 | exon |
| TBK1 | Ile305Thr | I305T | Substitution | T | C | Ile | Thr | 305 | exon |
| TBK1 | Arg308Gln | R308G | Substitution | G | A | Arg | Gln | 308 | exon |
| TBK1 | Thr320GlnfsX40 | T320QfsX40 | Frameshift | A |  | Thr | Gln | 320 | exon |
| TBK1 | Arg357Gln | R357G | Substitution | G | A | Arg | Gln | 357 | exon |
| TBK1 | Ala417X | A417X | Frameshift | G | A | Ala | STOP | 417 | exon |
| TBK1 | Arg440X | R440X | Frameshift | C | T | Arg | STOP | 440 | exon |
| TBK1 | Ile450LysfsX15 | I450KfsX15 | Frameshift | ATT |  | Ile | Lys | 450 | exon |
| TBK1 | Val479GlufsX4 | V479EfsX4 | Frameshift | TG |  | Val | Glu | 479 | exon |
| TBK1 | Met559Arg | M559R | Substitution | T | G | Met | Arg | 559 | exon |
| TBK1 | Ala571Val | A571V | Substitution | C | T | Ala | Val | 571 | exon |
| TBK1 | Met598Val | M598V | Substitution | A | G | Met | Val | 598 | exon |
| TBK1 | Glu643del | E643del | Frameshift | AAG |  | Glu |  | 643 | exon |
| TBK1 | 690-713del | 690-713del | Frameshift | T | C |  |  | 690 | exon |
| TBK1 | Glu696Lys | E96K | Substitution | G | A | Glu | Lys | 696 | exon |
| TRPM7 | Thr1482Ile | T1482I | Substitution |  |  |  |  | 1482 | exon |
| TUBA4A | c.226+4A>G | IVS2+4A>G | Frameshift | A | G |  |  |  | exon |
| TUBA4A | Val7Ile | V7I | Substitution | C | A | Val | Ile | 7 | exon |
| TUBA4A | Gly43Val | G43V | Substitution | C | A | Gly | Val | 43 | exon |
| TUBA4A | Thr145Pro | T145P | Substitution | T | G | Thr | Pro | 145 | exon |
| TUBA4A | Arg215Cys | R215C | Substitution | G | A | Arg | Cys | 215 | exon |
| TUBA4A | Arg320Cys | R320C | Substitution | G | A | Arg | Cys | 320 | exon |
| TUBA4A | Arg320His | R320H | Substitution | G | A | Arg | His | 320 | exon |
| TUBA4A | Thr349Ser | T349S | Substitution | A | T | Thr | Ser | 347 | exon |
| TUBA4A | Ala383Thr | A383T | Substitution | C | T | Ala | Thr | 383 | exon |
| TUBA4A | Trp407STOP | W407X | Substitution | C | T | Trp | X | 407 | exon |
| TUBA4A | Lys430Asn | K430N | Substitution | C | G | Lys | Asn | 430 | exon |
| TUBA4A | Asp438Asn | D438N | Substitution | G | A | Asp | Asn | 438 | exon |
| UBQLN2 | Ser155Asn | S155N | Substitution | G | A | Ser | Asn | 155 | exon |
| UBQLN2 | Pro189Thr | P189T | Substitution | C | A | Pro | Thr | 189 | exon |
| UBQLN2 | Ala282Val | A282V | Substitution | C | T | Ala | Val | 282 | exon |
| UBQLN2 | Ala283Thr | A283T | Substitution | G | A | Ala | Thr | 283 | exon |
| UBQLN2 | Asp314Glu | D314E | Substitution | T | A | Asp | Glu | 314 | exon |
| UBQLN2 | Ser340Ile | S340I | Substitution | G | T | Ser | Ile | 340 | exon |
| UBQLN2 | Ser346Cys | S346C | Substitution | C | G | Ser | Cys | 346 | exon |
| UBQLN2 | Met392Ile | M392I | Substitution |  |  | Met | Ile | 392 | exon |
| UBQLN2 | Ser400Gly | S400G | Substitution | A | G | Ser | Gly | 400 | exon |
| UBQLN2 | Glu425Arg | Q425R | Substitution | A | G | Gln | Arg | 425 | exon |
| UBQLN2 | Asn439Ile | N439I | Substitution | A | T | Asn | Ile | 439 | exon |
| UBQLN2 | Pro440Leu | P440L | Substitution | C | T | Pro | Leu | 440 | exon |
| UBQLN2 | Met446Arg | M446R | Substitution | T | G | Met | Arg | 446 | exon |
| UBQLN2 | Gly461Gly | G461G | Substitution | G | G | Gly | Gly | 461 | exon |
| UBQLN2 | Thr487Thr | T487T | Substitution | C | A | Thr | Thr | 487 | exon |
| UBQLN2 | Thr487Ile | T487I | Substitution | C | T | Thr | Ile | 487 | exon |
| UBQLN2 | Pro497His | P497H | Substitution | C | A | Pro | His | 497 | exon |
| UBQLN2 | Pro497Ser | P497S | Substitution | C | T | Pro | Ser | 497 | exon |
| UBQLN2 | Pro497Leu | P497L | Substitution | C | T | Pro | Leu | 497 | exon |
| UBQLN2 | Gly502\_Ile504del | G502\_I504del | Deletion | C | delCATAGGCCC | Gly |  | 502 |  |
| UBQLN2 | Pro506Ser | P506S | Substitution | C | T | Pro | Ser | 506 | exon |
| UBQLN2 | Pro506Thr | P506T | Substitution | C | A | Pro | Thr | 506 | exon |
| UBQLN2 | Pro509Ser | P509S | Substitution | C | T | Pro | Ser | 509 | exon |
| UBQLN2 | Pro525Ser | P525S | Substitution | C | T | Pro | Ser | 525 | exon |
| UBQLN2 | Pro533Leu | P533L | Substitution | C | T | Pro | Leu | 533 | exon |
| UBQLN2 | Val538Leu | V538L | Substitution | G | C | Val | Leu | 538 | exon |
| VAPB | Thr46Ile | T46I | Substitution | C | T | Thr | Ile | 46 | exon |
| VAPB | Pro56Ser | P56S | Substitution | CCC | TCC | Pro | Ser | 56 | exon |
| VCP | c.129+47G>A | c.129+47G>A | Intronic |  |  |  |  |  | intron |
| VCP | c.811+3G>A | c.811+3G>A | Intronic |  |  |  |  |  | intron |
| VCP | c.1082 â€“7\_8delinsTTGTGTACTGT | c.1082 â€“7\_8delinsTTGTGTACTGT | Intronic |  |  |  |  |  | intron |
| VCP | Arg155His | R155H | Substitution | G | A | Arg | His | 155 | exon |
| VCP | Arg159Gly | R159G | Substitution | C | G | Arg | Gly | 159 | exon |
| VCP | Arg191Gln | R191Q | Substitution | G | A | Arg | Gln | 191 | exon |
| VCP | Asp592Asn | D592N | Substitution | G | A | Asp | Asn | 592 | exon |