

S4 Table. Homozygous variants after filtering in individual MS-II-1

| CHROM | POS | ID | REF | ALT | GeneName | Func | ExonicFunc | AAChange | 1000g2015aug_ | esp6500siv | ExAC_ALL |
|-------|-----------|-------------|-----------|-------------|-----------|---------------|-------------------|-----------------------------|---------------|------------|----------|
| 5 | 7757704 | rs764722596 | G | GCCCCAGAGC | ADCY2 | splicing | . | . | . | . | 8.33E-03 |
| 7 | 150783911 | rs769408537 | A | G | AGAP3 | exonic | missense SNV | c.A83G:p.Q28R | . | . | 0 |
| 7 | 150783917 | rs775200524 | T | G | AGAP3 | exonic | missense SNV | c.T89G:p.L30R | . | . | 0 |
| 7 | 150783922 | rs766984834 | T | TGGGG | AGAP3 | exonic | frameshift INS | c.94_95insGGGG:p.C32fs | . | . | 0 |
| 7 | 150783919 | rs776100575 | GTCT | G | AGAP3 | exonic | nonframeshift DEL | c.92_94del:p.31_32del | . | . | 0 |
| X | 48925533 | . | G | A | CCDC120 | exonic | missense SNV | c.G1883A:p.R628H | 0.000264901 | . | 0.0030 |
| 14 | 39784012 | rs554677567 | A | ATGTGTGTGT | CTAGE5 | splicing | . | . | 0.00479233 | . | . |
| 15 | 45401064 | . | AGCTGGGCA | A | DUOX2 | exonic | nonframeshift DEL | c.1300_1320del:p.434_440del | . | . | . |
| 8 | 11666218 | rs71711801 | GTCCAC | G | FDFT1 | exonic | nonframeshift DEL | c.193_198del:p.65_66del | . | . | . |
| 6 | 32632646 | rs9274387 | G | T | HLA-DQB1 | exonic | missense SNV | c.C308A:p.T103N | . | . | 0.0036 |
| 19 | 55239223 | . | G | A | KIR3DL3 | exonic | missense SNV | c.G502A:p.V168I | . | . | . |
| 15 | 43621201 | rs774895599 | A | C | LCMT2 | exonic | missense SNV | c.T1487G:p.F496C | . | . | 1.65E-02 |
| 21 | 35334666 | rs766424 | C | T | LINC00649 | exonic | missense SNV | c.C377T:p.S126F | . | . | . |
| 3 | 100170600 | rs71132521 | A | ATCCTAGAAG | LNP1 | exonic | nonframeshift INS | ¹ see below | . | . | . |
| 15 | 56736015 | rs185005213 | G | A | MNS1 | exonic | stopgain | c.C724T:p.R242X | 0.000399361 | 7.7e-05 | 0.0004 |
| 1 | 17086085 | rs200532237 | G | GC | MST1L | exonic | frameshift INS | c.811dupG:p.A271fs | . | . | 0.0010 |
| X | 153421854 | . | A | T | OPN1LW | exonic | missense SNV | c.A830T:p.Y277F | . | . | . |
| X | 153421859 | . | G | T | OPN1LW | exonic | missense SNV | c.G835T:p.V279F | . | 0.0002 | 1.15E-02 |
| X | 153421844 | . | A | G | OPN1LW | exonic | missense SNV | c.A820G:p.I274V | . | . | . |
| X | 153421847 | . | T | C | OPN1LW | exonic | missense SNV | c.T823C:p.F275L | . | . | . |
| X | 153421849 | . | T | G | OPN1LW | exonic | missense SNV | c.T825G:p.F275L | . | . | 1.15E-02 |
| X | 153496198 | . | T | A | OPN1MW2 | exonic | missense SNV | c.T926A:p.F309Y | . | . | 0.0001 |
| X | 153496164 | . | C | G | OPN1MW2 | exonic | missense SNV | c.C892G:p.P298A | 0.0015894 | . | 0.0003 |
| X | 153496125 | . | G | A | OPN1MW2 | exonic | missense SNV | c.G853A:p.A285T | . | . | 0.0005 |
| 3 | 16926607 | rs7626448 | T | C | PLCL2 | exonic | unknown | UNKNOWN | . | . | 0 |
| 3 | 16926544 | rs368284573 | T | G | PLCL2 | exonic;splici | unknown | UNKNOWN | . | . | 0 |
| X | 153036027 | . | C | T | PLXNB3 | exonic | missense SNV | c.C1940T:p.P647L | . | . | 6.37E-02 |
| 21 | 14982886 | rs142207431 | G | A | POTED | exonic | missense SNV | c.G337A:p.G113S | . | . | . |
| 21 | 14982952 | rs117112505 | A | G | POTED | exonic | missense SNV | c.A403G:p.I135V | . | . | . |
| 21 | 15013735 | rs78643169 | A | G | POTED | exonic | missense SNV | c.A1603G:p.M535V | . | . | . |
| 1 | 33430102 | rs113840389 | T | G | RNF19B | exonic | missense SNV | c.A185C:p.Q62P | . | . | . |
| 8 | 10467637 | rs369606728 | T | TCCTCTAACTG | RP1L1 | exonic | nonframeshift INS | ² see below | . | . | . |
| 13 | 78272267 | rs201380414 | T | TGG | SLAIN1 | exonic | frameshift INS | c.219_220insGG:p.A73fs | . | . | 0 |
| 3 | 14444329 | rs67559364 | CT | C | SLC6A6 | splicing | . | . | . | . | . |
| 15 | 51041909 | rs145832874 | C | T | SPPL2A | exonic | missense SNV | c.G101A:p.G34E | 0.00259585 | 0.0011 | 0.0019 |
| 3 | 12046256 | rs782385358 | G | CCCCCCCC | SYN2 | exonic | unknown | UNKNOWN | . | . | 0 |
| 3 | 12046268 | rs782162668 | G | GCGCGA | SYN2 | exonic | unknown | UNKNOWN | . | . | 0 |
| 3 | 12046222 | rs71624921 | A | C | SYN2 | exonic | unknown | UNKNOWN | . | . | 0 |
| 3 | 12046215 | rs2623876 | G | C | SYN2 | exonic | unknown | UNKNOWN | . | . | 0 |
| 2 | 46707808 | rs70940616 | C | CAGCGGCAGC | TMEM247 | exonic | nonframeshift INS | ³ see below | . | . | . |
| 3 | 42251577 | rs10634555 | C | CGGA | TRAK1 | exonic | nonframeshift INS | ⁴ see below | . | . | . |
| 14 | 38183928 | rs78042024 | G | A | TTC6 | exonic | missense SNV | c.G1688A:p.G563E | 0.00439297 | . | 0.0028 |
| 4 | 9236859 | rs776377841 | A | T | USP17L15 | exonic | missense SNV | c.A749T:p.N250I | . | . | 0 |

¹c.194_195insTCCTAGAAGGCATTCTCATGAGGACCAGGAATCCGATGCCGATCGTCTGACCGTCT:p.H65delinsHPRRSHEDQEFRCRSDRL

²c.3970_3971insGGACTAAAGTAATAGAAGGGCTGCAAGAAGAGAGGGTGCAGTTAGAGG:p.E1324delinsGTKVIEGLQEERVQLEE

³c.382_383insAGCGGCAGCACGAGGTGGTGTGGAGCAGCTGCAGCGGG:p.Q128delinsQRQHEVVMEQLQRE

⁴c.1841_1842insGGA:p.T614delinsTE