**Multi-gene testing in neurological disorders showed an improved diagnostic yield: data from over 1000 Indian patients**

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**Table S1: List of ‘pathogenic’/’likely pathogenic’ single nucleotide and indel variants identified in the study along with NGS (next generation sequencing) quality parameters. All the variants were assessed for supporting reads (SR), total reads (TR), average base quality (ABQ), average mapping quality (AMQ) and strand bias (SB). Variants with the following quality parameters were classified as high confidence variants; SR% ≥30 (for a Het call) and ≥90 (for a Hom/Hemi call), TR ≥40 (for a Het call ‘OR’ if TR < 40, there should be at least 12 (30% of 40) SRs) and ≥20 (for a Hom/Hemi call), ABQ ≥30, AMQ ≥240 and SB% ≤30, and these variants were excluded from Sanger confirmation.**

|  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Sl.No.** | **Sample Id** | **Gene** | **Nucleotide** | **Zygosity** | **%SR** | **TR** | **ABQ** | **AMQ** | **%SB** | **Decision for Sanger test** | **Sanger done** | **Sanger result** |
| 1 | R-4266 | *AAAS* | c.43C>A | Hom | 97.01 | 67 | 35.69 | 254.00 | 0.41 | Accept | No |  |
| 2 | S-3752 | *AAAS* | c.855\_856delinsTT | Hom | 96.75 | 123 | 35.14 | 254.00 | 1.97 | Accept | No |  |
| 3 | S-2407 | *ABCD1* | c.443A>G | Hemi | 98.44 | 64 | 35.11 | 254.00 | 1.79 | Accept | No |  |
| 4 | S-2545 | *ABCD1* | c.459C>A | Hemi | 94.79 | 96 | 34.70 | 254.00 | 1.56 | Accept | No |  |
| 5 | S-5919 | *ABCD1* | c.712delG | Hemi | 94.90 | 98 | 31.26 | 254.03 | 0.70 | Accept | No |  |
| 6 | S-6559 | *ABCD1* | c.796G>A | Hemi | 96.77 | 31 | 33.00 | 254.00 | 0.65 | **\*Accept** | **Yes** | Concordant |
| 7 | S-3648 | *ABCD1* | c.901-1G>A | Hemi | 100.00 | 82 | 34.91 | 254.00 | 0.00 | **\*Accept** | **Yes** | Concordant |
| 8 | S-80 | *ACTC1* | c.107\_120delinsC | Het | 50.00 | 80 | 33.00 | 254.00 | 5.00 | Accept | No |  |
| 9 | S-2936 | *ADAR* | c.788\_791delACAG | Het | 69.19 | 198 | 31.64 | 254.02 | 0.87 | Accept | No |  |
| 10 | S-5239 | *ADSL* | c.1339T>C | Het | 42.86 | 140 | 34.62 | 254.00 | 7.14 | Accept | No |  |
| 11 | S-5981 | *AHI1* | c.988delinsTAAATAATA | Hom | 93.00 | 162 | 34.00 | 254.00 | 3.00 | **\*Accept** | **Yes** | Concordant |
| 12 | S-1948 | *AHI1* | c.1152-1G>C | Hom | 100.00 | 21 | 34.43 | 254.00 | 0.00 | Accept | No |  |
| 13 | S-4566 | *AHI1* | c.2168G>A | Het | 48.46 | 227 | 35.05 | 252.85 | 4.79 | **\*Accept** | **Yes** | Concordant |
| 14 | S-4566 | *AHI1* | c.2212C>T | Het | 54.84 | 217 | 35.33 | 254.00 | 6.51 | **\*Accept** | **Yes** | Concordant |
| 15 | S-4350 | *ALDH3A2* | c.126delG | Hom | 100.00 | 54 | 34.15 | 254.00 | 0.00 | Accept | No |  |
| 16 | [S-2952](file:///C:\..\..\aparna\AppData\Roaming\Microsoft\Excel\Reports\Hyperlinking_Aug2017_May2018\RESCH-000001302.pdf) | *ALDH7A1* | c.1279G>C | Het | 37.80 | 127 | 34.38 | 254.00 | 11.42 | Accept | No |  |
| 17 | S-4199 | *ALDH7A1* | c.1411\_1412insG | Het | 49.00 | 100 | 32.25 | 254.00 | 0.29 | Accept | No |  |
| 18 | S-4679 | *ALG13* | c.320A>G | Het | 45.00 | 179 | 34.67 | 254.00 | 4.73 | Accept | No |  |
| 19 | [S-2593](file:///C:\..\..\aparna\AppData\Roaming\Microsoft\Excel\Reports\Hyperlinking_Aug2017_May2018\STRAN-2018-4749.pdf) | *ALS2* | c.880\_881delCT | Hom | 96.66 | 419 | 34.71 | 254.00 | 0.26 | **\*Accept** | **Yes** | Concordant |
| 20 | S-1091 | *ALS2* | c.3415C>T | Hom | 97.73 | 88 | 35.17 | 254.01 | 0.32 | **\*Accept** | **Yes** | Concordant |
| 21 | S-2989 | *ANG* | c.186\_190dupCTGCA | Het | 36.69 | 616 | 34.21 | 254.00 | 3.79 | Accept | No |  |
| 22 | S-3198 | *ANO5* | c.1733T>C | Hom | 97.70 | 87 | 35.14 | 254.00 | 0.57 | Accept | No |  |
| 23 | S-3469 | *AP4M1* | c.59-1G>C | Het | 43.22 | 118 | 35.35 | 254.12 | 7.91 | **\*Accept** | **Yes** | Concordant |
| 24 | S-3469 | *AP4M1* | c.901delT | Het | 44.57 | 92 | 34.51 | 254.05 | 11.77 | **\*Accept** | **Yes** | Concordant |
| 25 | S-3687 | *APTX* | c.596delG | Hom | 100.00 | 81 | 34.11 | 254.00 | 0.00 | Accept | No |  |
| 26 | S-5910 | *APTX* | c.596delG | Hom | 93.59 | 78 | 33.16 | 254.01 | 1.65 | Accept | No |  |
| 27 | S-3919 | *APTX* | c.596delG | Hom | 98.78 | 82 | 33.65 | 254.00 | 0.81 | Accept | No |  |
| 28 | S-753 | *APTX* | c.875-2A>G | Hom | 96.26 | 107 | 34.37 | 254.04 | 0.56 | **\*Accept** | **Yes** | Concordant |
| 29 | S-1347 | *ARID1B* | c.5547delC | Het | 45.85 | 205 | 32.89 | 254.01 | 7.68 | Accept | No |  |
| 30 | S-1763 | *ARSA* | c.346C>T | Hom | 96.49 | 57 | 33.84 | 254.00 | 2.93 | Accept | No |  |
| 31 | S-2325 | *ARSA* | c.877C>T | Hom | 100.00 | 96 | 34.48 | 254.01 | 0.00 | Accept | No |  |
| 32 | S-4598 | *ARSA* | c.917C>T | Het | 41.10 | 163 | 35.15 | 254.01 | 12.00 | **\*Accept** | **Yes** | Concordant |
| 33 | S-2212 | *ARSA* | c.917C>T | Hom | 97.40 | 77 | 35.35 | 254.00 | 0.80 | **\*Accept** | **Yes** | Concordant |
| 34 | S-3364 | *ARSA* | c.917C>T | Hom | 99.05 | 105 | 35.13 | 254.02 | 1.21 | **\*Accept** | **Yes** | Concordant |
| 35 | S-3145 | *ARSA* | c.931G>A | Hom | 100.00 | 146 | 35.40 | 254.01 | 0.00 | Accept | No |  |
| 36 | S-3827 | *ARX* | c.1425\_1446del | Het | 48.10 | 79 | 33.88 | 254.08 | 3.20 | Accept | No |  |
| 37 | S-6032 | *ASNS* | c.1321-2A>G | Hom | 97.71 | 218 | 34.22 | 247.14 | 0.92 | **\*Accept** | **Yes** | Concordant |
| 38 | S-887 | *ASPM* | c.277delC | Hom | 98.46 | 65 | 33.70 | 254.02 | 0.34 | **\*Accept** | **Yes** | Concordant |
| 39 | S-1362 | *ASPM* | c.900delC | Het | 41.11 | 253 | 34.01 | 254.01 | 5.50 | Accept | No |  |
| 40 | S-1362 | *ASPM* | c.1729\_1730delAG | Het | 54.65 | 269 | 34.01 | 254.00 | 11.46 | Accept | No |  |
| 41 | S-1339 | *ASPM* | c.7125\_7128dupACTG | Hom | 90.81 | 185 | 34.00 | 254.00 | 2.39 | Accept | No |  |
| 42 | S-5189 | *ATM* | c.387delA | Hom | 100.00 | 54 | 35.04 | 254.02 | 0.00 | Accept | No |  |
| 43 | [S-5948](file:///C:\..\..\aparna\AppData\Roaming\Microsoft\Excel\Reports\Hyperlinking_Aug2017_May2018\STRAN-2018-4850.pdf) | *ATM* | c.4909+1G>A | Hom | 97.18 | 71 | 34.99 | 254.00 | 4.00 | Accept | No |  |
| 44 | S-4309 | *ATM* | c.6096-2A>G | Hom | 99.38 | 160 | 33.77 | 254.00 | 0.63 | Accept | No |  |
| 45 | S-2588 | *ATM* | c.8545C>T | Het | 52.78 | 72 | 35.47 | 254.05 | 1.46 | Accept | No |  |
| 46 | S-2051 | *ATM* | c.8873\_8874delTT | Hom | 100.00 | 51 | 32.39 | 254.00 | 0.00 | Accept | No |  |
| 47 | R-0549 | *ATP1A2* | c.2723G>A | Het | 45.10 | 204 | 33.82 | 254.00 | 2.05 | Accept | No |  |
| 48 | R-4319 | *ATP1A3* | c.2401G>A | Het | 48.09 | 131 | 33.13 | 254.00 | 10.23 | **\*Accept** | **Yes** | Concordant |
| 49 | S-590 | *ATP1A3* | c.2401G>A | Het | 54.41 | 136 | 34.26 | 254.03 | 14.75 | Accept | No |  |
| 50 | S-4078 | *ATP1A3* | c.2401G>A | Het | 51.06 | 94 | 34.46 | 254.02 | 10.28 | **\*Accept** | **Yes** | Concordant |
| 51 | S-2402 | *ATP1A3* | c.2443G>A | Het | 51.35 | 222 | 34.50 | 254.01 | 1.75 | Accept | No |  |
| 52 | S-2223 | *ATP1A3* | c.2839G>A | Het | 40.97 | 227 | 35.21 | 254.01 | 2.79 | Accept | No |  |
| 53 | S-2993 | *ATP1A3* | c.2839G>A | Het | 47.00 | 100 | 34.79 | 254.00 | 12.64 | Accept | No |  |
| 54 | S-4266 | *ATP7B* | c.813C>A | Hom | 99.29 | 567 | 34.99 | 254.00 | 0.41 | Accept | No |  |
| 55 | S-3386 | *ATP8A2* | c.3019-1G>A | Hom | 100.00 | 136 | 34.49 | 254.00 | 0.00 | Accept | No |  |
| 56 | S-2049 | *AUH* | c.505+1G>C | Het | 50.00 | 126 | 35.43 | 254.00 | 1.59 | Accept | No |  |
| 57 | R-0597 | *BRAF* | c.1914T>G | Het | 47.83 | 92 | 33.45 | 254.00 | 14.43 | **\*Accept** | **Yes** | Concordant |
| 58 | S-4320 | *BRCA2* | c.4695\_4698dupGACC | Het | 48.42 | 95 | 34.00 | 254.00 | 3.43 | Accept | No |  |
| 59 | S-4749 | *BTD* | c.98\_104delinsTCC | Het | 47.00 | 87 | 35.00 | 254.00 | 11.90 | Accept | No |  |
| 60 | S-2918 | *BUB1B* | c.933\_951del | Het | 68.75 | 80 | 33.46 | 254.19 | 7.50 | Accept | No |  |
| 61 | S-2832 | *C12orf57* | c.19C>T | Hom | 100.00 | 40 | 35.47 | 254.00 | 0.00 | Accept | No |  |
| 62 | S-3353 | *C12orf65* | c.346delG | Hom | 97.82 | 229 | 34.78 | 254.00 | 0.56 | Accept | No |  |
| 63 | S-3673 | *CAPN3* | c.383delA | Hom | 100.00 | 121 | 31.31 | 254.01 | 0.00 | Accept | No |  |
| 64 | [S-5902](file:///C:\..\..\aparna\AppData\Roaming\Microsoft\Excel\Reports\Hyperlinking_Aug2017_May2018\STRAN-2018-4638.pdf) | *CAPN3* | c.701G>A | Het | 60.90 | 133 | 35.00 | 254.00 | 8.74 | Accept | No |  |
| 65 | S-3642 | *CAPN3* | c.802-9G>A | Hom | 100.00 | 95 | 34.46 | 254.01 | 0.00 | Accept | No |  |
| 66 | S-5902 | *CAPN3* | c.1063C>T | Het | 40.62 | 128 | 34.90 | 254.00 | 20.67 | Accept | No |  |
| 67 | S-4649 | *CAPN3* | c.2051-1G>T | Het | 43.79 | 354 | 34.39 | 254.02 | 5.22 | Accept | No |  |
| 68 | [S-4102](file:///C:\..\..\aparna\AppData\Roaming\Microsoft\Excel\Reports\Hyperlinking_Aug2017_May2018\STRAN-2018-4762.pdf) | *CAPN3* | c.2185-2A>G | Hom | 99.29 | 282 | 32.98 | 254.00 | 0.39 | Accept | No |  |
| 69 | S-3597 | *CAPN3* | c.2243G>A | Hom | 98.46 | 195 | 35.33 | 254.03 | 0.80 | Accept | No |  |
| 70 | S-3859 | *CAPN3* | c.2243G>A | Hom | 98.58 | 141 | 34.45 | 254.00 | 0.74 | Accept | No |  |
| 71 | S-4649 | *CAPN3* | c.2338G>C | Het | 47.28 | 349 | 35.10 | 254.00 | 2.76 | Accept | No |  |
| 72 | S-707 | *CAPN3* | c.2338G>C | Hom | 98.59 | 213 | 34.99 | 251.62 | 0.62 | Accept | No |  |
| 73 | S-2936 | *CASK* | c.79C>T | Het | 48.57 | 140 | 35.19 | 254.00 | 3.61 | Accept | No |  |
| 74 | S-4099 | *CD59* | c.21delT | Het | 60.23 | 88 | 34.51 | 254.00 | 5.83 | Accept | No |  |
| 75 | S-4086 | *CDK5RAP2* | c.1139C>A | Het | 52.63 | 95 | 35.06 | 254.02 | 11.79 | **\*Accept** | **Yes** | Concordant |
| 76 | S-488 | *CDKL5* | c.1842T>A | Het | 53.12 | 401 | 34.97 | 254.01 | 0.07 | Accept | No |  |
| 77 | S-4826 | *CEP135* | c.473-1G>C | Het | 41.45 | 152 | 34.90 | 254.00 | 2.44 | Accept | No |  |
| 78 | S-1775 | *CEP290* | c.2365C>T | Het | 53.54 | 99 | 35.09 | 254.00 | 2.63 | Accept | No |  |
| 79 | S-1400 | *CEP290* | c.4219delT | Hom | 100.00 | 70 | 36.21 | 254.00 | 0.00 | Accept | No |  |
| 80 | S-1848 | *CEP290* | c.4792A>T | Hom | 100.00 | 39 | 37.18 | 254.00 | 0.00 | **\*Accept** | **Yes** | Concordant |
| 81 | S-4631 | *CEP290* | c.5649delA | Hom | 98.05 | 205 | 33.53 | 254.00 | 0.73 | **\*Accept** | **Yes** | Concordant |
| 82 | S-1775 | *CEP290* | c.5668G>T | Het | 43.94 | 198 | 34.91 | 254.00 | 4.88 | Accept | No |  |
| 83 | S-2563 | *CEP290* | c.5766\_5769delAGAA | Hom | 98.73 | 79 | 32.45 | 254.00 | 1.20 | Accept | No |  |
| 84 | S-6496 | *CHD2* | c.4909C>T | Het | 53.85 | 130 | 34.17 | 254.00 | 3.74 | Accept | No |  |
| 85 | S-3110 | *CHD7* | c.1611G>A | Het | 50.22 | 462 | 30.94 | 253.46 | 1.15 | Accept | No |  |
| 86 | S-3773 | *CHD7* | c.4756\_4757delAA | Het | 43.27 | 171 | 34.08 | 254.00 | 9.70 | Accept | No |  |
| 87 | S-3056 | *CHRNE* | c.183\_187dupCTCAC | Hom | 97.00 | 146 | 32.98 | 254.00 | 4.01 | Accept | No |  |
| 88 | S-5167 | *CHRNE* | c.183\_187dupCTCAC | Hom | 92.00 | 85 | 31.76 | 254.02 | 7.02 | Accept | No |  |
| 89 | S-2875 | *CHRNE* | c.799C>T | Hom | 99.32 | 148 | 34.09 | 254.01 | 0.79 | Accept | No |  |
| 90 | S-3586 | *CHRNE* | c.878\_887dup | Hom | 100.00 | 230 | 31.98 | 254.02 | 0.34 | Accept | No |  |
| 91 | S-876 | *CHRNE* | c.1216\_1219+19del | Hom | 96.67 | 60 | 34.18 | 254.11 | 1.49 | Accept | No |  |
| 92 | S-705 | *CHRNE* | c.1327delG | Hom | 93.43 | 137 | 33.05 | 252.08 | 0.32 | Accept | No |  |
| 93 | S-1157 | *CHRNE* | c.1327delG | Hom | 97.92 | 144 | 32.74 | 254.02 | 2.04 | Accept | No |  |
| 94 | S-1632 | *CHRNE* | c.1327delG | Hom | 97.12 | 139 | 32.87 | 254.02 | 1.63 | Accept | No |  |
| 95 | S-168 | *CHRNE* | c.1371delC | Hom | 100.00 | 137 | 34.47 | 254.01 | 0.00 | Accept | No |  |
| 96 | S-1860 | *CHRNE* | c.1371delC | Hom | 96.23 | 159 | 34.21 | 254.01 | 1.92 | Accept | No |  |
| 97 | S-4641 | *CLCN1* | c.47G>A | Het | 47.02 | 285 | 34.49 | 254.04 | 2.18 | Accept | No |  |
| 98 | R-0515 | *CLCN1* | c.803C>T | Hom | 100.00 | 32 | 35.16 | 254.00 | 0.00 | **\*Accept** | **Yes** | Concordant |
| 99 | S-4721 | *CLCN1* | c.908G>A | Hom | 99.29 | 141 | 35.26 | 254.00 | 0.65 | Accept | No |  |
| 100 | S-4681 | *CLCN1* | c.1129C>T | Hom | 100.00 | 91 | 34.90 | 254.00 | 0.00 | Accept | No |  |
| 101 | S-2685 | *CLCN1* | c.1495G>A | Hom | 98.66 | 149 | 35.29 | 254.01 | 1.24 | **\*Accept** | **Yes** | Concordant |
| 102 | S-690 | *CLCN1* | c.1667T>A | Het | 38.76 | 178 | 34.57 | 254.00 | 0.62 | Accept | No |  |
| 103 | S-2058 | *CLCN1* | c.1876C>T | Hom | 97.74 | 177 | 34.72 | 254.02 | 0.14 | Accept | No |  |
| 104 | S-2577 | *CLCN1* | c.2472\_2473dupTC | Hom | 96.69 | 181 | 33.72 | 254.00 | 1.05 | Accept | No |  |
| 105 | S-690 | *CLCN1* | c.2472\_2473dupTC | Het | 43.33 | 150 | 33.78 | 254.00 | 0.31 | Accept | No |  |
| 106 | S-5686 | *CLCN2* | c.2068G>T | Hom | 98.95 | 95 | 33.69 | 254.11 | 1.03 | Accept | No |  |
| 107 | S-4390 | *CLN5* | c.595C>T | Hom | 99.53 | 215 | 35.09 | 254.00 | 0.50 | Accept | No |  |
| 108 | S-3224 | *CLN5* | c.746\_747insCA | Hom | 100.00 | 48 | 34.00 | 254.00 | 14.58 | Accept | No |  |
| 109 | S-3555 | *CLN6* | c.476C>T | Hom | 100.00 | 34 | 32.12 | 254.00 | 0.00 | Accept | No |  |
| 110 | S-3731 | *CLN6* | c.775G>C | Hom | 96.67 | 300 | 34.57 | 254.00 | 0.05 | Accept | No |  |
| 111 | S-4672 | *CLN8* | c.1A>C | Hom | 100.00 | 15 | 36.00 | 254.20 | 0.00 | Check | Yes | Concordant |
| 112 | [S-5622](file:///C:\..\..\aparna\AppData\Roaming\Microsoft\Excel\Reports\Hyperlinking_Aug2017_May2018\STRAN-2018-4929.pdf) | *COL4A1* | c.337C>T | Het | 47.64 | 254 | 34.64 | 254.03 | 10.83 | Accept | No |  |
| 113 | S-4386 | *COL4A2* | c.3205delC | Het | 52.46 | 183 | 33.76 | 254.00 | 2.97 | Accept | No |  |
| 114 | S-3030 | *COL4A3* | c.828+1G>T | Hom | 100.00 | 270 | 33.00 | 254.00 | 0.00 | Accept | No |  |
| 115 | S-3912 | *COL6A1* | c.297delC | Hom | 97.41 | 309 | 35.31 | 254.00 | 1.21 | **\*Accept** | **Yes** | Concordant |
| 116 | S-528 | *COL6A1* | c.850G>A | Het | 38.46 | 13 | 36.00 | 254.00 | 27.69 | Check | Yes | Concordant |
| 117 | S-2061 | *COL6A1* | c.868G>A | Het | 51.16 | 129 | 34.27 | 254.00 | 4.44 | Accept | No |  |
| 118 | S-4466 | *COL6A1* | c.877G>A | Het | 55.00 | 260 | 34.18 | 254.00 | 1.33 | **\*Accept** | **Yes** | Concordant |
| 119 | S-2761 | *COL6A1* | c.1675-1G>C | Hom | 99.45 | 181 | 35.52 | 254.03 | 0.50 | Accept | No |  |
| 120 | S-5468 | *COL6A2* | c.38delG | Hom | 97.56 | 41 | 32.50 | 254.02 | 1.46 | Accept | No |  |
| 121 | S-5310 | *COL6A2* | c.310C>T | Hom | 99.30 | 287 | 34.23 | 253.56 | 0.08 | Accept | No |  |
| 122 | S-1215 | *COL6A2* | c.875G>T | Het | 48.98 | 196 | 35.01 | 254.02 | 13.31 | Accept | No |  |
| 123 | S-1176 | *COL6A2* | c.1270-4\_1273dupGCAGGGGC | Het | 47.95 | 73 | 37.48 | 254.00 | 4.46 | **\*Accept** | **Yes** | Concordant |
| 124 | S-1176 | *COL6A2* | c.2864\_2865delAC | Het | 54.87 | 195 | 36.57 | 254.00 | 2.08 | **\*Accept** | **Yes** | Concordant |
| 125 | S-5445 | *COQ2* | c.1184dupT | Het | 41.41 | 99 | 32.61 | 254.00 | 19.76 | **\*Accept** | **Yes** | Concordant |
| 126 | S-3810 | *CSF1R* | c.2381T>C | Het | 41.33 | 150 | 34.45 | 254.02 | 8.43 | Accept | No |  |
| 127 | S-6075 | *CTC1* | c.1056\_1069delinsC | Het | 40.00 | 280 | 34.00 | 254.00 | 15.00 | Accept | No |  |
| 128 | S-2427 | *CUL4B* | c.1007\_1011delTTATA | Hemi | 98.86 | 88 | 33.53 | 254.00 | 0.50 | **\*Accept** | **Yes** | Concordant |
| 129 | S-3247 | *CYP27A1* | c.646+1G>A | Hom | 98.44 | 64 | 34.64 | 254.03 | 2.23 | Accept | No |  |
| 130 | S-2300 | *DCX* | c.667G>A | Hemi | 97.14 | 70 | 34.97 | 254.00 | 0.34 | Accept | No |  |
| 131 | S-3298 | *DMD* | c.724C>T | Hemi | 95.56 | 90 | 35.43 | 254.01 | 2.22 | Accept | No |  |
| 132 | S-381 | *DMD* | c.1412delC | Hemi | 97.92 | 96 | 33.44 | 254.01 | 0.49 | **\*Accept** | **Yes** | Concordant |
| 133 | S-4863 | *DMD* | c.1693dupA | Hemi | 100.00 | 37 | 34.97 | 254.00 | 0.00 | Accept | No |  |
| 134 | S-5408 | *DMD* | c.4018delA | Hemi | 93.75 | 192 | 34.46 | 254.01 | 0.97 | Accept | No |  |
| 135 | S-35 | *DMD* | c.4151delA | Hemi | 97.85 | 93 | 34.31 | 254.01 | 0.54 | Accept | No |  |
| 136 | S-5291 | *DMD* | c.4375C>T | Hemi | 97.92 | 96 | 35.02 | 254.01 | 0.31 | Accept | No |  |
| 137 | S-5788 | *DMD* | c.5140delG | Hemi | 100.00 | 42 | 32.45 | 254.02 | 0.00 | Accept | No |  |
| 138 | S-4249 | *DMD* | c.5899C>T | Hemi | 96.81 | 94 | 34.70 | 254.01 | 2.24 | Accept | No |  |
| 139 | S-4319 | *DMD* | c.5963\_5967delACATG | Hemi | 98.31 | 59 | 33.84 | 254.00 | 1.75 | Accept | No |  |
| 140 | S-870 | *DMD* | c.6238C>T | Het | 49.15 | 118 | 35.05 | 254.00 | 6.78 | **\*Accept** | **Yes** | Concordant |
| 141 | S-6071 | *DMD* | c.6283C>T | Hemi | 96.00 | 75 | 35.43 | 254.00 | 2.44 | Accept | No |  |
| 142 | [S-4445](file:///C:\..\..\aparna\AppData\Roaming\Microsoft\Excel\Reports\Hyperlinking_Aug2017_May2018\STRAN-2018-4545.pdf) | *DMD* | c.6819\_6826del | Hemi | 97.37 | 114 | 34.31 | 254.00 | 0.66 | **\*Accept** | **Yes** | Concordant |
| 143 | S-5878 | *DMD* | c.9568C>T | Hemi | 98.99 | 99 | 34.82 | 254.06 | 0.85 | Accept | No |  |
| 144 | S-1126 | *DNAJC6* | c.748C>T | Hom | 100.00 | 89 | 34.80 | 254.03 | 0.00 | Accept | No |  |
| 145 | S-4952 | *DOK7* | c.1121\_1124dupGCCT | Hom | 92.00 | 175 | 34.17 | 254.00 | 1.99 | Accept | No |  |
| 146 | S-2397 | *DSG2* | c.1021delG | Het | 41.33 | 75 | 33.10 | 254.00 | 10.41 | Accept | No |  |
| 147 | S-3604 | *DYSF* | c.1357C>T | Hom | 97.73 | 88 | 33.03 | 254.00 | 0.11 | **\*Accept** | **Yes** | Concordant |
| 148 | S-5728 | *DYSF* | c.5713C>T | Hom | 99.05 | 105 | 34.20 | 254.03 | 1.08 | Accept | No |  |
| 149 | S-2781 | *DYSF* | c.634delC | Het | 49.58 | 119 | 34.33 | 254.00 | 15.90 | Accept | No |  |
| 150 | S-2179 | *EIF2B2* | c.433+2T>G | Het | 55.00 | 120 | 33.83 | 254.03 | 7.88 | Accept | No |  |
| 151 | R-1349 | *ENAM* | c.1259\_1260insAG | Hom | 96.25 | 80 | 33.52 | 254.00 | 3.41 | Accept | No |  |
| 152 | S-1799 | *EPM2A* | c.690delG | Hom | 100.00 | 148 | 34.59 | 254.00 | 0.00 | Accept | No |  |
| 153 | R-0529 | *EPM2A* | c.929delT | Het | 57.14 | 112 | 33.66 | 254.00 | 2.68 | Accept | No |  |
| 154 | S-4506 | *ERCC6* | c.1012delA | Hom | 99.10 | 221 | 33.51 | 254.01 | 0.02 | **\*Accept** | **Yes** | Concordant |
| 155 | S-4707 | *ERCC6* | c.1536dupG | Het | 56.94 | 72 | 35.80 | 254.00 | 7.86 | Accept | No |  |
| 156 | S-4707 | *ERCC6* | c.4063-1G>C | Het | 51.74 | 201 | 35.05 | 254.01 | 4.51 | Accept | No |  |
| 157 | S-3631 | *ERCC8* | c.952delG | Het | 50.84 | 179 | 34.31 | 254.01 | 0.50 | **\*Accept** | **Yes** | Concordant |
| 158 | S-1614 | *EXOSC3* | c.395A>C | Hom | 77.69 | 121 | 34.66 | 247.41 | 6.51 | Check | Yes | Concordant |
| 159 | S-1080 | *FKRP* | c.1343C>T | Het | 56.82 | 176 | 35.01 | 254.00 | 5.68 | Accept | No |  |
| 160 | S-1961 | *FKRP* | c.1343C>T | Hom | 100.00 | 170 | 35.31 | 254.00 | 0.00 | Accept | No |  |
| 161 | S-378 | *FKRP* | c.1343C>T | Hom | 98.25 | 171 | 34.89 | 254.01 | 1.27 | **\*Accept** | **Yes** | Concordant |
| 162 | S-3420 | *FUS* | c.1520G>A | Het | 54.84 | 31 | 35.06 | 254.18 | 6.07 | Accept | No |  |
| 163 | S-3714 | *GAA* | c.1927G>A | Het | 55.00 | 120 | 31.64 | 254.00 | 12.58 | Accept | No |  |
| 164 | S-4990 | *GALC* | c.1230delC | Het | 50.59 | 85 | 34.53 | 254.00 | 5.58 | Accept | No |  |
| 165 | S-410 | *GAMT* | c.491dupG | Hom | 95.00 | 144 | 34.41 | 254.01 | 2.58 | Accept | No |  |
| 166 | S-710 | *GDAP1* | c.128dupT | Hom | 98.86 | 176 | 33.67 | 251.96 | 0.68 | Accept | No |  |
| 167 | S-3459 | *GDAP1* | c.358C>T | Het | 47.30 | 74 | 35.77 | 254.00 | 17.92 | Accept | No |  |
| 168 | S-5873 | *GDAP1* | c.769C>T | Hom | 98.62 | 290 | 34.37 | 254.04 | 0.09 | **\*Accept** | **Yes** | Concordant |
| 169 | S-5083 | *GDAP1* | c.802\_803delTG | Het | 45.45 | 110 | 32.98 | 254.05 | 10.55 | Accept | No |  |
| 170 | S-3908 | *GFAP* | c.262C>T | Het | 40.19 | 107 | 35.30 | 254.00 | 3.26 | Accept | No |  |
| 171 | S-3380 | *GJB1* | c.43C>T | Hemi | 100.00 | 30 | 35.73 | 254.00 | 0.00 | Accept | No |  |
| 172 | S-2754 | *GJC2* | c.508delG | Hom | 100.00 | 44 | 33.80 | 254.02 | 0.00 | Accept | No |  |
| 173 | S-5966 | *GJC2* | c.982delG | Het | 36.36 | 11 | 27.75 | 254.00 | 27.27 | Check | Yes | Concordant |
| 174 | S-4159 | *GLB1* | c.145C>T | Het | 41.90 | 105 | 35.41 | 254.05 | 6.67 | Accept | No |  |
| 175 | [S-4159](file:///C:\..\..\aparna\AppData\Roaming\Microsoft\Excel\Reports\Hyperlinking_Aug2017_May2018\STRAN-2018-5011.pdf) | *GLB1* | c.1325G>A | Het | 44.53 | 137 | 35.43 | 254.03 | 3.78 | Accept | No |  |
| 176 | S-5514 | *GLDC* | c.1786C>T | Het | 49.49 | 99 | 34.78 | 248.92 | 9.32 | Accept | No |  |
| 177 | S-4666 | *GLRA1* | c.839G>A | Hom | 98.06 | 258 | 34.63 | 253.51 | 0.85 | Accept | No |  |
| 178 | S-4657 | *GM2A* | c.244-2A>G | Hom | 97.47 | 79 | 32.62 | 254.00 | 0.10 | Accept | No |  |
| 179 | S-4296 | *GNE* | c.478C>T | Het | 53.33 | 210 | 34.62 | 254.00 | 1.55 | Accept | No |  |
| 180 | S-4544 | *GNE* | c.478C>T | Het | 49.70 | 501 | 34.94 | 254.01 | 3.79 | Accept | No |  |
| 181 | S-439 | *GNE* | c.478C>T | Het | 55.06 | 385 | 35.14 | 254.00 | 2.90 | Accept | No |  |
| 182 | S-1191 | *GNE* | c.1760T>C | Hom | 97.61 | 167 | 37.22 | 254.00 | 1.27 | Accept | No |  |
| 183 | S-5444 | *GNPTG* | c.189\_192dupTCTC | Hom | 94.70 | 132 | 34.35 | 254.00 | 0.23 | Accept | No |  |
| 184 | S-760 | *GRIN2A* | c.2764delG | Het | 56.20 | 395 | 33.35 | 253.46 | 5.18 | Accept | No |  |
| 185 | [S-5420](file:///C:\..\..\aparna\AppData\Roaming\Microsoft\Excel\Reports\Hyperlinking_Aug2017_May2018\STRAN-2018-4696.pdf) | *GRM1* | c.748delC | Hom | 93.61 | 313 | 33.01 | 254.02 | 0.43 | Accept | No |  |
| 186 | S-5250 | *HEXA* | c.964G>T | Hom | 96.43 | 112 | 35.48 | 254.00 | 0.66 | Accept | No |  |
| 187 | S-5286 | *HEXA* | c.1444G>A | Hom | 97.98 | 247 | 34.91 | 254.01 | 0.90 | Accept | No |  |
| 188 | S-37 | *HGSNAT* | c.1019G>A | Hom | 98.13 | 107 | 35.04 | 254.00 | 0.84 | Accept | No |  |
| 189 | S-1643 | *HPD* | c.1005C>G | Het | 53.40 | 103 | 34.95 | 254.00 | 10.52 | Accept | No |  |
| 190 | S-2482 | *HPRT1* | c.532+5G>A | Hemi | 97.06 | 68 | 35.11 | 254.00 | 0.80 | Accept | No |  |
| 191 | S-5906 | *IGHMBP2* | c.958C>T | Hom | 97.92 | 48 | 34.85 | 254.00 | 1.95 | Accept | No |  |
| 192 | R-0606 | *IGHMBP2* | c.2430delT | Hom | 96.94 | 261 | 31.23 | 254.00 | 0.60 | **\*Accept** | **Yes** | Concordant |
| 193 | S-5963 | *IQSEC2* | c.2682delC | Het | 46.43 | 224 | 32.01 | 254.01 | 8.72 | Accept | No |  |
| 194 | S-1059 | *IQSEC2* | c.2890-2\_2898del | Hemi | 98.25 | 114 | 33.46 | 254.02 | 1.79 | **\*Accept** | **Yes** | Concordant |
| 195 | S-4543 | *KARS* | c.223C>T | Het | 44.01 | 284 | 34.61 | 254.00 | 2.53 | Accept | No |  |
| 196 | S-898 | *KCNJ10* | c.168G>A | Het | 45.28 | 265 | 35.16 | 254.02 | 7.80 | Accept | No |  |
| 197 | S-4014 | *KCNQ1* | c.613G>A | Het | 42.40 | 217 | 34.60 | 254.00 | 4.01 | Accept | No |  |
| 198 | S-4698 | *KCNQ1* | c.1552C>T | Het | 61.97 | 71 | 35.05 | 254.00 | 3.78 | Accept | No |  |
| 199 | S-3640 | *KCNQ2* | c.629G>C | Het | 22.58 | 62 | 34.50 | 254.00 | 12.44 | Check | Yes | Concordant |
| 200 | S-566 | *KCNQ2* | c.997C>T | Het | 42.67 | 75 | 35.12 | 254.00 | 21.83 | Accept | No |  |
| 201 | S-4083 | *KCNQ2* | c.1041C>G | Het | 46.02 | 226 | 34.61 | 254.02 | 3.45 | Accept | No |  |
| 202 | S-1375 | *KIAA2022* | c.2749G>T | Het | 47.98 | 371 | 34.98 | 254.01 | 2.98 | Accept | No |  |
| 203 | S-3449 | *KIF1A* | c.296C>T | Het | 49.27 | 205 | 35.44 | 254.01 | 4.29 | **\*Accept** | **Yes** | Concordant |
| 204 | S-1143 | *KMT2D* | c.303dupG | Het | 44.88 | 283 | 33.91 | 254.00 | 4.39 | Accept | No |  |
| 205 | S-2052 | *KMT2D* | c.7456delC | Het | 58.62 | 29 | 33.94 | 254.00 | 1.22 | **\*Accept** | **Yes** | Concordant |
| 206 | S-4848 | *KMT2D* | c.14516-2A>C | Het | 51.63 | 215 | 34.74 | 254.01 | 8.80 | Accept | No |  |
| 207 | S-80 | *L1CAM* | c.604G>T | Hemi | 97.58 | 165 | 34.66 | 254.02 | 0.20 | **\*Accept** | **Yes** | Concordant |
| 208 | S-4873 | *L1CAM* | c.2278C>T | Hemi | 96.82 | 220 | 35.21 | 254.01 | 1.66 | **\*Accept** | **Yes** | Concordant |
| 209 | S-5306 | *L2HGDH* | c.829C>T | Hom | 100.00 | 79 | 35.43 | 254.00 | 0.00 | Accept | No |  |
| 210 | S-4167 | *LAMA2* | c.250C>T | Het | 43.48 | 46 | 33.10 | 254.00 | 22.61 | **\*Accept** | **Yes** | Concordant |
| 211 | S-4928 | *LAMA2* | c.286\_287delAG | Het | 51.17 | 213 | 34.69 | 254.01 | 1.86 | Accept | No |  |
| 212 | S-5626 | *LAMA2* | c.910-2A>G | Het | 48.57 | 70 | 34.47 | 254.00 | 16.64 | Accept | No |  |
| 213 | S-3611 | *LAMA2* | c.1307-1G>C | Hom | 98.97 | 195 | 34.06 | 254.00 | 0.81 | Accept | No |  |
| 214 | S-3227 | *LAMA2* | c.1749C>G | Hom | 96.84 | 95 | 32.34 | 254.00 | 0.71 | Accept | No |  |
| 215 | S-2689 | *LAMA2* | c.2538delG | Hom | 100.00 | 30 | 32.30 | 254.00 | 0.00 | Accept | No |  |
| 216 | S-246 | *LAMA2* | c.2749+4\_2749+15del | Het | 74.00 | 35 | 35.00 | 254.00 | 6.15 | Accept | No |  |
| 217 | [S-2749](file:///C:\..\..\aparna\AppData\Roaming\Microsoft\Excel\Reports\Hyperlinking_Aug2017_May2018\STRAN-2018-4706.pdf) | *LAMA2* | c.4048C>T | Hom | 92.59 | 54 | 35.06 | 254.02 | 0.44 | Accept | No |  |
| 218 | S-2392 | *LAMA2* | c.4198C>T | Hom | 100.00 | 162 | 35.39 | 254.01 | 0.00 | Accept | No |  |
| 219 | R-0569 | *LAMA2* | c.4348C>T | Het | 29.73 | 37 | 33.91 | 254.00 | 35.38 | Check | Yes | Concordant |
| 220 | S-4167 | *LAMA2* | c.5360G>A | Het | 44.59 | 74 | 33.82 | 254.00 | 1.64 | **\*Accept** | **Yes** | Concordant |
| 221 | S-5563 | *LAMA2* | c.7147C>T | Het | 40.57 | 106 | 34.79 | 254.02 | 5.09 | **\*Accept** | **Yes** | Concordant |
| 222 | S-369 | *LAMA2* | c.7816delA | Het | 51.40 | 107 | 35.25 | 254.04 | 12.71 | Accept | No |  |
| 223 | S-1525 | *LAMA2* | c.8072\_8073delCT | Hom | 97.47 | 79 | 33.33 | 254.00 | 0.66 | **\*Accept** | **Yes** | Concordant |
| 224 | S-369 | *LAMA2* | c.8703+1G>A | Het | 52.38 | 63 | 35.15 | 254.00 | 1.44 | Accept | No |  |
| 225 | S-5563 | *LAMA2* | c.8767G>T | Het | 47.06 | 340 | 34.36 | 254.04 | 7.57 | **\*Accept** | **Yes** | Concordant |
| 226 | S-1553 | *LDLR* | c.1060+2T>G | Het | 57.50 | 80 | 33.91 | 254.07 | 10.98 | Accept | No |  |
| 227 | S-6242 | *LMNA* | c.1157G>A | Het | 49.52 | 105 | 34.04 | 254.06 | 14.21 | Accept | No |  |
| 228 | S-1358 | *MECP2* | c.316C>T | Het | 33.09 | 136 | 35.13 | 254.00 | 17.22 | Accept | No |  |
| 229 | S-1726 | *MECP2* | c.316C>T | Het | 52.71 | 129 | 34.59 | 254.01 | 1.14 | Accept | No |  |
| 230 | S-346 | *MECP2* | c.397C>T | Het | 54.00 | 50 | 35.37 | 254.00 | 6.81 | Accept | No |  |
| 231 | S-4174 | *MECP2* | c.455C>G | Het | 49.59 | 121 | 34.17 | 254.00 | 3.61 | Accept | No |  |
| 232 | S-4299 | *MECP2* | c.455C>G | Het | 44.63 | 177 | 33.67 | 254.00 | 7.45 | Accept | No |  |
| 233 | S-2350 | *MECP2* | c.455C>G | Het | 56.79 | 81 | 35.09 | 254.02 | 2.74 | Accept | No |  |
| 234 | S-1838 | *MECP2* | c.473C>T | Het | 52.14 | 117 | 34.90 | 254.05 | 4.04 | Accept | No |  |
| 235 | S-3104 | *MECP2* | c.473C>T | Het | 40.37 | 161 | 34.94 | 254.00 | 0.61 | Accept | No |  |
| 236 | S-4200 | *MECP2* | c.502C>T | Het | 43.44 | 221 | 33.04 | 254.00 | 3.63 | Accept | No |  |
| 237 | S-2738 | *MECP2* | c.611dupC | Het | 51.61 | 124 | 31.67 | 254.00 | 3.63 | **\*Accept** | **Yes** | Concordant |
| 238 | S-2801 | *MECP2* | c.645\_646delAA | Het | 45.45 | 88 | 30.74 | 254.00 | 5.45 | Accept | No |  |
| 239 | S-886 | *MECP2* | c.695delG | Het | 63.37 | 202 | 33.53 | 254.01 | 11.85 | Accept | No |  |
| 240 | S-3305 | *MECP2* | c.763C>T | Het | 47.52 | 141 | 34.81 | 254.00 | 11.60 | Accept | No |  |
| 241 | S-3408 | *MECP2* | c.763C>T | Het | 42.86 | 133 | 34.44 | 254.02 | 25.06 | **\*Accept** | **Yes** | Concordant |
| 242 | S-5585 | *MECP2* | c.808C>T | Het | 42.67 | 150 | 33.73 | 254.05 | 15.62 | Accept | No |  |
| 243 | S-5361 | *MECP2* | c.917G>A | Het | 47.86 | 234 | 35.50 | 254.01 | 4.35 | Accept | No |  |
| 244 | S-2951 | *MECP2* | c.1450\_1453delAGAG | Het | 60.34 | 58 | 34.24 | 254.00 | 2.17 | Accept | No |  |
| 245 | S-5116 | *MEN1* | c.1350+1G>A | Het | 68.00 | 22 | 35.73 | 254.00 | 0.61 | Check | Yes | Concordant |
| 246 | S-4628 | *MFN2* | c.1090C>T | Het | 38.13 | 257 | 34.75 | 254.05 | 8.66 | **\*Accept** | **Yes** |  |
| 247 | S-1912 | *MFSD8* | c.217dupA | Hom | 100.00 | 14 | 36.93 | 254.00 | 0.00 | Check | Yes | Concordant |
| 248 | S-4504 | *MFSD8* | c.590dupG | Hom | 94.32 | 88 | 34.07 | 254.00 | 1.20 | Accept | No |  |
| 249 | S-2284 | *MFSD8* | c.694delC | Hom | 100.00 | 49 | 34.90 | 254.00 | 0.00 | Accept | No |  |
| 250 | S-6 | *MFSD8* | c.699-1G>A | Hom | 97.85 | 93 | 35.21 | 254.03 | 0.31 | **\*Accept** | **Yes** | Concordant |
| 251 | S-1925 | *MFSD8* | c.1338delA | Hom | 97.62 | 42 | 35.49 | 254.00 | 1.39 | Accept | No |  |
| 252 | S-2324 | *MLC1* | c.959C>A | Hom | 97.58 | 165 | 35.19 | 254.01 | 0.02 | Accept | No |  |
| 253 | S-5680 | *MLC1* | c.135dupC | Hom | 100.00 | 77 | 33.69 | 254.00 | 0.00 | Accept | No |  |
| 254 | S-5406 | *MMACHC* | c.394C>T | Hom | 98.41 | 126 | 34.66 | 254.02 | 0.44 | **\*Accept** | **Yes** | Concordant |
| 255 | S-6030 | *MTHFR* | c.1262G>C | Hom | 98.53 | 136 | 33.96 | 254.01 | 0.20 | Accept | No |  |
| 256 | S-3769 | *MTM1* | c.1420C>T | Hemi | 100.00 | 83 | 33.87 | 254.00 | 0.00 | **\*Accept** | **Yes** | Concordant |
| 257 | S-5539 | *MTMR2* | c.1734dupA | Hom | 99.13 | 115 | 32.94 | 254.00 | 1.10 | Accept | No |  |
| 258 | S-329 | *MTTP* | c.1092delC | Hom | 98.99 | 198 | 34.21 | 254.00 | 0.79 | Accept | No |  |
| 259 | S-4525 | *MUSK* | c.308A>G | Hom | 98.01 | 351 | 35.12 | 254.01 | 0.43 | Accept | No |  |
| 260 | S-5257 | *MYBPC3* | c.383\_384insGCCCCAAGCTG | Het | 30.56 | 72 | 32.89 | 254.00 | 52.02 | Accept | No |  |
| 261 | S-4875 | *MYH2* | c.2377C>T | Hom | 100.00 | 154 | 34.99 | 254.01 | 0.00 | Accept | No |  |
| 262 | S-5165 | *MYH7* | c.4850\_4852delAGA | Het | 54.48 | 268 | 31.98 | 236.76 | 1.26 | Accept | No |  |
| 263 | S-3632 | *MYH7* | c.5746C>T | Het | 59.00 | 100 | 35.05 | 254.00 | 2.64 | Accept | No |  |
| 264 | S-832 | *NDUFV2* | c.1A>G | Het | 51.72 | 29 | 35.20 | 254.00 | 17.47 | Check | Yes | Concordant |
| 265 | S-1310 | *NDUFV2* | c.427C>T | Het | 34.62 | 52 | 35.56 | 254.00 | 14.96 | Accept | No |  |
| 266 | S-2616 | *NEB* | c.1470+2T>C | Het | 58.82 | 34 | 34.25 | 254.00 | 8.82 | Accept | No |  |
| 267 | S-2616 | *NEB* | c.18676C>T | Het | 36.89 | 103 | 31.63 | 254.00 | 17.12 | Accept | No |  |
| 268 | S-2135 | *NEB* | c.25137delT | Het | 45.33 | 75 | 33.00 | 254.09 | 16.31 | Accept | No |  |
| 269 | S-567 | *NEFL* | c.1261C>T | Hom | 98.61 | 72 | 34.68 | 254.01 | 0.90 | Accept | No |  |
| 270 | S-1143 | *NF2* | c.1021C>T | Het | 52.24 | 134 | 33.69 | 254.00 | 4.18 | Accept | No |  |
| 271 | S-5215 | *NHLRC1* | c.468\_469delAG | Het | 61.65 | 133 | 33.00 | 254.08 | 13.02 | Accept | No |  |
| 272 | S-5215 | *NHLRC1* | c.793C>T | Het | 46.32 | 367 | 33.45 | 254.05 | 3.80 | Accept | No |  |
| 273 | S-4709 | *NIPBL* | c.2689C>T | Het | 51.33 | 150 | 34.79 | 254.00 | 3.48 | Accept | No |  |
| 274 | S-4473 | *NIPBL* | c.5272C>T | Het | 43.71 | 151 | 35.02 | 254.00 | 3.73 | Accept | No |  |
| 275 | S-3171 | *NKX2-1* | c.650C>A | Het | 38.28 | 128 | 34.63 | 251.49 | 13.33 | Accept | No |  |
| 276 | S-5530 | *NOTCH3* | c.544C>T | Het | 49.54 | 109 | 34.37 | 254.07 | 1.05 | Accept | No |  |
| 277 | S-2000 | *NPC1* | c.2604+1G>A | Het | 70.97 | 31 | 34.13 | 254.00 | 5.87 | Accept | No | Concordant |
| 278 | S-453 | *NSD1* | c.1318C>T | Het | 51.66 | 271 | 35.42 | 254.00 | 2.22 | Accept | No |  |
| 279 | S-5998 | *NSD1* | c.1855delG | Het | 51.40 | 107 | 33.93 | 254.00 | 4.42 | Accept | No |  |
| 280 | S-342 | *NSD1* | c.6010-2A>C | Het | 46.00 | 291 | 33.88 | 254.00 | 5.40 | Accept | No |  |
| 281 | S-695 | *NSD1* | c.6049C>T | Het | 41.90 | 253 | 35.59 | 252.83 | 7.46 | Accept | No |  |
| 282 | R-0604 | *NSUN2* | c.560delC | Hom | 99.49 | 194 | 34.20 | 254.00 | 0.53 | Accept | No |  |
| 283 | S-5459 | *NTRK1* | c.404\_405delTG | Het | 54.72 | 212 | 33.36 | 254.02 | 3.61 | Accept | No |  |
| 284 | S-5459 | *NTRK1* | c.1787G>A | Het | 53.06 | 49 | 33.77 | 254.00 | 5.65 | Accept | No |  |
| 285 | S-6052 | *NTRK1* | c.2115G>A | Hom | 96.77 | 62 | 35.02 | 254.00 | 0.11 | Accept | No |  |
| 286 | S-3192 | *OCRL* | c.827T>C | Hemi | 97.58 | 124 | 31.46 | 254.00 | 0.35 | **\*Accept** | **Yes** | Concordant |
| 287 | S-5835 | *OSTM1* | c.518\_521delATTG | Hom | 97.44 | 78 | 32.70 | 254.00 | 2.29 | **\*Accept** | **Yes** | Concordant |
| 288 | S-5508 | *PANK2* | c.828\_829delTG | Hom | 98.88 | 179 | 33.28 | 254.01 | 0.92 | Accept | No |  |
| 289 | S-5519 | *PANK2* | c.856C>T | Het | 51.22 | 205 | 34.74 | 254.02 | 6.55 | Accept | No |  |
| 290 | S-4691 | *PCDH19* | c.94G>T | Het | 43.56 | 101 | 34.32 | 251.14 | 16.25 | Accept | No |  |
| 291 | S-3346 | *PCDH19* | c.339\_355delinsTCGT | Het | 45.00 | 170 | 31.00 | 254.00 | 4.00 | Accept | No |  |
| 292 | S-4839 | *PCNT* | c.5767C>T | Het | 46.40 | 125 | 35.50 | 254.00 | 0.83 | Accept | No |  |
| 293 | S-4592 | *PDHA1* | c.483C>T | Het | 56.31 | 206 | 34.12 | 254.02 | 4.54 | Accept | No |  |
| 294 | S-340 | *PHYH* | c.679-2A>C | Het | 63.79 | 58 | 34.40 | 254.00 | 8.01 | **\*Accept** | **Yes** | Concordant |
| 295 | S-1781 | *PKD2* | c.1003C>T | Het | 46.64 | 238 | 35.93 | 254.00 | 5.10 | Accept | No |  |
| 296 | S-19 | *PLA2G6* | c.210-2A>G | Hom | 100.00 | 191 | 34.49 | 254.00 | 0.00 | **\*Accept** | **Yes** | Concordant |
| 297 | R-0832 | *PLA2G6* | c.1077G>A | Het | 53.23 | 62 | 34.91 | 254.00 | 4.30 | Accept | No |  |
| 298 | S-3802 | *PLA2G6* | c.1077G>A | Hom | 99.46 | 185 | 34.71 | 254.00 | 0.39 | Accept | No |  |
| 299 | S-4155 | *PLA2G6* | c.1999G>T | Hom | 98.77 | 162 | 34.64 | 254.00 | 0.12 | **\*Accept** | **Yes** | Concordant |
| 300 | R-0832 | *PLA2G6* | c.2222G>A | Het | 44.12 | 68 | 34.77 | 254.00 | 10.98 | Accept | No |  |
| 301 | S-1249 | *PLA2G6* | c.2222G>A | Hom | 99.53 | 213 | 35.66 | 253.41 | 0.51 | Accept | No |  |
| 302 | S-4795 | *PLA2G6* | c.2222G>A | Het | 54.49 | 156 | 34.94 | 254.01 | 6.50 | Accept | No |  |
| 303 | S-2562 | *PNKP* | c.936+1G>T | Het | 46.89 | 177 | 34.41 | 254.04 | 0.94 | Accept | No |  |
| 304 | S-339 | *PNPO* | c.346C>T | Het | 47.00 | 123 | 35.00 | 254.00 | 2.37 | Accept | No |  |
| 305 | S-1439 | *POLG* | c.911T>G | Hom | 99.35 | 153 | 34.99 | 254.03 | 0.83 | Accept | No |  |
| 306 | S-4266 | *POMGNT1* | c.617G>A | Hom | 99.49 | 195 | 35.10 | 254.01 | 0.54 | Accept | No |  |
| 307 | S-2947 | *POMT2* | c.1057G>A | Hom | 99.38 | 160 | 34.78 | 254.01 | 0.63 | Accept | No |  |
| 308 | S-5455 | *PPT1* | c.451C>T | Hom | 98.39 | 62 | 34.54 | 254.00 | 1.59 | Accept | No |  |
| 309 | S-5616 | *PRX* | c.231C>A | Hom | 99.21 | 253 | 35.23 | 254.00 | 0.54 | Accept | No |  |
| 310 | S-495 | *PTEN* | c.304A>T | Het | 51.79 | 168 | 35.07 | 206.20 | 1.19 | Check | Yes | Concordant |
| 311 | S-485 | *PYGM* | c.1098G>A | Het | 52.22 | 203 | 35.38 | 254.00 | 2.06 | Accept | No |  |
| 312 | S-3829 | *RAB3GAP1* | c.28G>T | Hom | 100.00 | 183 | 34.38 | 254.03 | 0.00 | Accept | No |  |
| 313 | S-2690 | *RAB3GAP2* | c.1937\_1938delAT | Hom | 97.09 | 206 | 34.71 | 254.00 | 0.91 | Accept | No |  |
| 314 | S-4788 | *RNASEH2A* | c.322C>T | Hom | 98.99 | 199 | 33.38 | 254.01 | 0.08 | **\*Accept** | **Yes** | Concordant |
| 315 | S-1987 | *RNASEH2C* | c.205C>T | Hom | 96.77 | 31 | 35.07 | 254.00 | 0.86 | **\*Accept** | **Yes** | Concordant |
| 316 | [S-4256](file:///C:\..\..\aparna\AppData\Roaming\Microsoft\Excel\Reports\Hyperlinking_Aug2017_May2018\STRAN-2018-4778.pdf) | *RNASEH2C* | c.205C>T | Het | 48.78 | 82 | 33.42 | 254.00 | 13.78 | Accept | No |  |
| 317 | S-2825 | *RNASEH2C* | c.205C>T | Hom | 100.00 | 47 | 33.85 | 254.00 | 0.00 | Accept | No |  |
| 318 | S-5194 | *RYR1* | c.3748G>T | Het | 39.71 | 136 | 34.65 | 254.02 | 9.20 | Accept | No |  |
| 319 | S-334 | *RYR1* | c.10825-2A>T | Het | 50.00 | 168 | 35.18 | 254.01 | 0.00 | Accept | No |  |
| 320 | S-5217 | *SCN1A* | c.265-1G>T | Het | 48.80 | 125 | 35.28 | 254.03 | 0.10 | Accept | No |  |
| 321 | S-3764 | *SCN1A* | c.302G>A | Het | 46.02 | 176 | 35.02 | 254.00 | 0.80 | Accept | No |  |
| 322 | S-3457 | *SCN1A* | c.602+1G>A | Het | 44.79 | 192 | 35.16 | 254.00 | 0.97 | Accept | No |  |
| 323 | S-4925 | *SCN1A* | c.602+1G>A | Het | 49.34 | 152 | 35.23 | 254.00 | 1.26 | Accept | No |  |
| 324 | S-4940 | *SCN1A* | c.955C>T | Het | 53.26 | 92 | 34.76 | 254.00 | 3.73 | Accept | No |  |
| 325 | S-5248 | *SCN1A* | c.964+2T>G | Het | 48.21 | 56 | 34.89 | 254.00 | 1.59 | Accept | No |  |
| 326 | S-5621 | *SCN1A* | c.1284delA | Het | 50.00 | 152 | 33.43 | 250.84 | 0.00 | Accept | No |  |
| 327 | S-2483 | *SCN1A* | c.2505+3A>T | Het | 60.00 | 45 | 35.70 | 254.19 | 13.33 | Accept | No |  |
| 328 | S-5561 | *SCN1A* | c.2589+3A>T | Het | 60.78 | 51 | 34.35 | 254.16 | 26.44 | Accept | No |  |
| 329 | S-1532 | *SCN1A* | c.2624C>A | Het | 47.74 | 155 | 35.31 | 247.47 | 1.90 | **\*Accept** | **Yes** | Concordant |
| 330 | S-4101 | *SCN1A* | c.3733C>T | Het | 58.89 | 90 | 35.47 | 254.00 | 9.27 | Accept | No |  |
| 331 | S-3756 | *SCN1A* | c.4250delA | Het | 48.95 | 190 | 34.27 | 254.00 | 1.54 | Accept | No |  |
| 332 | S-4722 | *SCN1A* | c.4338+1G>A | Het | 47.22 | 36 | 35.76 | 254.00 | 15.03 | **\*Accept** | **Yes** | Concordant |
| 333 | S-255 | *SCN1A* | c.4853-1G>C | Het | 51.69 | 207 | 34.94 | 236.77 | 2.93 | Accept | No |  |
| 334 | [S-5280](file:///C:\..\..\aparna\AppData\Roaming\Microsoft\Excel\Reports\Hyperlinking_Aug2017_May2018\STRAN-2018-4333.pdf) | *SCN1A* | c.4942C>T | Het | 49.56 | 226 | 35.43 | 240.00 | 8.63 | Accept | No |  |
| 335 | S-3088 | *SCN1B* | c.254G>A | Hom | 98.65 | 74 | 35.07 | 254.01 | 0.00 | Accept | No |  |
| 336 | S-1722 | *SCN2A* | c.4471delA | Het | 48.15 | 108 | 34.71 | 254.00 | 2.71 | Accept | No |  |
| 337 | S-361 | *SEPN1* | c.249\_250dupGG | Het | 36.90 | 65 | 33.15 | 254.00 | 8.90 | **\*Accept** | **Yes** | Concordant |
| 338 | S-361 | *SEPN1* | c.999delG | Het | 45.00 | 90 | 32.00 | 254.00 | 16.50 | **\*Accept** | **Yes** | Concordant |
| 339 | S-1372 | *SEPN1* | c.1282-2A>C | Het | 56.00 | 85 | 34.80 | 254.00 | 1.57 | Accept | No |  |
| 340 | S-2844 | *SETX* | c.718+1G>A | Hom | 99.30 | 143 | 35.30 | 251.32 | 1.03 | Accept | No |  |
| 341 | S-2642 | *SETX* | c.5475dupA | Het | 46.58 | 73 | 35.03 | 254.00 | 0.56 | Accept | No |  |
| 342 | S-4214 | *SETX* | c.5927T>G | Het | 48.81 | 84 | 34.29 | 254.00 | 0.17 | **\*Accept** | **Yes** | Concordant |
| 343 | S-3821 | *SGCA* | c.197T>A | Het | 54.27 | 293 | 33.33 | 254.00 | 2.93 | Accept | No |  |
| 344 | S-3821 | *SGCA* | c.220C>T | Het | 44.02 | 234 | 32.08 | 254.00 | 6.55 | Accept | No |  |
| 345 | S-3707 | *SGCA* | c.409G>A | Het | 49.03 | 259 | 35.61 | 254.00 | 3.81 | Accept | No |  |
| 346 | [S-3707](file:///C:\..\..\aparna\AppData\Roaming\Microsoft\Excel\Reports\Hyperlinking_Aug2017_May2018\STRAN-2018-4940.pdf) | *SGCA* | c.614C>A | Het | 58.74 | 143 | 35.73 | 254.00 | 6.14 | Accept | No |  |
| 347 | S-4778 | *SGCB* | c.271C>T | Het | 52.05 | 171 | 34.94 | 254.01 | 4.60 | Accept | No |  |
| 348 | S-1651 | *SGCG* | c.848G>A | Hom | 100.00 | 136 | 35.12 | 254.02 | 0.00 | Accept | No |  |
| 349 | S-4682 | *SGSH* | c.571G>A | Het | 51.30 | 74 | 34.21 | 254.00 | 12.23 | Accept | No |  |
| 350 | S-1739 | *SH3TC2* | c.385+1G>A | Hom | 94.52 | 73 | 35.64 | 254.01 | 2.50 | Accept | No |  |
| 351 | S-3102 | *SH3TC2* | c.931A>T | Hom | 100.00 | 153 | 35.53 | 254.01 | 0.00 | Accept | No |  |
| 352 | S-246 | *SH3TC2* | c.2491\_2492delAG | Het | 46.00 | 150 | 32.70 | 254.00 | 2.49 | Accept | No |  |
| 353 | S-3696 | *SIL1* | c.767+2T>A | Hom | 100.00 | 68 | 35.38 | 254.00 | 0.00 | Accept | No |  |
| 354 | S-3981 | *SIL1* | c.1030-18G>A | Hom | 88.24 | 34 | 30.73 | 254.00 | 0.00 | Check | Yes | Concordant |
| 355 | S-5077 | *SIL1* | c.1260\_1279delinsCAC | Hom | 98.00 | 157 | 33.00 | 254.00 | 0.70 | Accept | No |  |
| 356 | S-722 | *SLC12A6* | c.2655dupT | Hom | 97.00 | 100 | 34.70 | 251.43 | 0.16 | Accept | No |  |
| 357 | S-2432 | *SLC16A2* | c.972G>A | Hemi | 100.00 | 81 | 34.06 | 254.00 | 0.00 | **\*Accept** | **Yes** | Concordant |
| 358 | S-3475 | *SLC19A3* | c.4delG | Hom | 98.25 | 57 | 30.61 | 254.00 | 1.63 | **\*Accept** | **Yes** | Concordant |
| 359 | S-3778 | *SLC22A5* | c.1195C>T | Het | 53.23 | 248 | 34.92 | 254.01 | 6.60 | Accept | No |  |
| 360 | S-3173 | *SLC25A22* | c.202+1dupG | Hom | 94.76 | 248 | 32.68 | 252.91 | 1.59 | **\*Accept** | **Yes** | Concordant |
| 361 | S-4145 | *SLC25A22* | c.818+1G>C | Hom | 96.43 | 140 | 33.79 | 254.04 | 0.90 | Accept | No |  |
| 362 | S-898 | *SLC26A4* | c.1334T>G | Het | 49.19 | 124 | 34.97 | 254.00 | 16.31 | Accept | No |  |
| 363 | S-5628 | *SLC2A1* | c.195G>A | Het | 52.99 | 117 | 34.69 | 254.00 | 13.95 | **\*Accept** | **Yes** | Concordant |
| 364 | S-1891 | *SLC2A1* | c.553delC | Het | 46.32 | 95 | 34.41 | 254.00 | 10.57 | Accept | No |  |
| 365 | S-3231 | *SLC2A1* | c.844delC | Het | 57.14 | 56 | 33.22 | 254.00 | 5.36 | Accept | No |  |
| 366 | S-221 | *SLC2A1* | c.997C>T | Het | 54.93 | 71 | 34.75 | 254.05 | 6.79 | Accept | No |  |
| 367 | S-4302 | *SLC2A1* | c.1105\_1112del | Het | 38.46 | 91 | 32.97 | 254.00 | 1.32 | **\*Accept** | **Yes** | Concordant |
| 368 | S-3390 | *SLC6A5* | c.679+2T>C | Hom | 97.83 | 46 | 34.76 | 254.07 | 1.93 | Accept | No |  |
| 369 | S-5036 | *SMC1A* | c.2394delA | Het | 56.25 | 160 | 32.42 | 254.02 | 3.89 | **\*Accept** | **Yes** | Concordant |
| 370 | S-5409 | *SMC1A* | c.3305\_3312del | Het | 63.27 | 147 | 32.52 | 254.03 | 0.97 | **\*Accept** | **Yes** | Concordant |
| 371 | S-5854 | *SNAP29* | c.487dupA | Het | 49.57 | 117 | 31.29 | 254.00 | 4.57 | Accept | No |  |
| 372 | S-5854 | *SNAP29* | c.586C>T | Het | 42.52 | 214 | 34.82 | 254.00 | 4.56 | Accept | No |  |
| 373 | S-4250 | *SPG11* | c.2146C>T | Hom | 95.83 | 120 | 34.69 | 254.02 | 0.51 | Accept | No |  |
| 374 | S-7220 | *SPG11* | c.3018T>G | Hom | 98.4 | 189 | 34.44 | 254.00 | 0.31 | Accept | No |  |
| 375 | S-4523 | *SPG11* | c.6100C>T | Hom | 96.80 | 125 | 35.09 | 254.00 | 2.53 | Accept | No |  |
| 376 | S-4351 | *SPG11* | c.6856C>T | Hom | 99.28 | 138 | 34.00 | 254.00 | 0.65 | Accept | No |  |
| 377 | S-4130 | *SQSTM1* | c.712\_713insTCCTCCGAGTGTGAATTTCCTGA | Hom | 98.80 | 174 | 33.82 | 254.03 | 1.95 | Accept | No |  |
| 378 | S-2368 | *SQSTM1* | c.823\_824delAG | Hom | 99.07 | 107 | 33.63 | 254.00 | 0.85 | Accept | No |  |
| 379 | S-2673 | *STXBP1* | c.841\_845delCTGGA | Het | 47.66 | 214 | 33.81 | 254.00 | 9.44 | Accept | No |  |
| 380 | S-3287 | *STXBP1* | c.841\_845delCTGGA | Het | 51.22 | 164 | 33.93 | 254.00 | 11.15 | Accept | No |  |
| 381 | S-3458 | *STXBP1* | c.1439C>T | Het | 48.18 | 137 | 34.89 | 254.00 | 20.15 | **\*Accept** | **Yes** | Concordant |
| 382 | S-3181 | *SURF1* | c.535dupA | Hom | 98.05 | 205 | 32.44 | 254.00 | 0.01 | Accept | No |  |
| 383 | S-3233 | *SURF1* | c.751C>T | Hom | 100.00 | 122 | 34.64 | 254.00 | 0.00 | Accept | No |  |
| 384 | S-776 | *SYNE1* | c.16015C>T | Het | 68.85 | 61 | 35.31 | 251.00 | 10.54 | Accept | No |  |
| 385 | S-2171 | *SYNGAP1* | c.1591\_1592delTG | Het | 44.00 | 92 | 32.00 | 254.00 | 13.00 | Accept | No |  |
| 386 | S-1331 | *SYNGAP1* | c.2474\_2477dupCGGA | Het | 48.07 | 362 | 34.00 | 254.00 | 4.88 | Accept | No |  |
| 387 | R-0840 | *TCF4* | c.550-2A>C | Het | 46.00 | 50 | 35.48 | 254.00 | 6.26 | Accept | No |  |
| 388 | S-5111 | *TK2* | c.416C>T | Hom | 100.00 | 70 | 35.00 | 254.03 | 0.00 | Accept | No |  |
| 389 | S-3945 | *TMEM43* | c.316\_317delTA | Het | 37.80 | 164 | 34.82 | 254.00 | 4.09 | Accept | No |  |
| 390 | S-3156 | *TNNT1* | c.32+1G>A | Hom | 100.00 | 120 | 34.70 | 254.00 | 0.00 | Accept | No |  |
| 391 | S-2314 | *TNNT1* | c.750+1G>A | Hom | 97.54 | 122 | 35.01 | 254.00 | 2.00 | Accept | No |  |
| 392 | S-2587 | *TPP1* | c.496dupC | Het | 46.05 | 152 | 32.79 | 254.00 | 2.89 | Accept | No |  |
| 393 | S-2587 | *TPP1* | c.622C>T | Het | 42.66 | 143 | 35.00 | 254.02 | 18.34 | Accept | No |  |
| 394 | S-1079 | *TPP1* | c.1146-1G>A | Hom | 98.97 | 97 | 33.06 | 254.00 | 0.97 | Accept | No |  |
| 395 | S-2691 | *TPP1* | c.1264C>T | Hom | 98.02 | 101 | 34.61 | 254.02 | 0.62 | Accept | No |  |
| 396 | [S-2684](file:///C:\..\..\aparna\AppData\Roaming\Microsoft\Excel\Reports\Hyperlinking_Aug2017_May2018\STRAN-2018-4657.pdf) | *TTN* | c.72686dupC | Het | 45.04 | 262 | 33.10 | 254.00 | 8.47 | Accept | No |  |
| 397 | S-3342 | *TTPA* | c.26C>A | Hom | 100.00 | 114 | 34.89 | 254.01 | 0.00 | **\*Accept** | **Yes** | Concordant |
| 398 | S-3161 | *UBE3A* | c.1480\_1481delCA | Het | 43.00 | 194 | 33.50 | 254.00 | 0.50 | Accept | No |  |
| 399 | S-1732 | *UBE3A* | c.1811\_1812delGT | Het | 43.30 | 97 | 35.43 | 242.17 | 17.23 | **\*Accept** | **Yes** | Concordant |
| 400 | S-6581 | *UNC80* | c.8215\_8227del | Hom | 99.40 | 169 | 33.30 | 254.03 | 0.42 | Accept | No |  |
| 401 | S-569 | *VRK1* | c.1159+1G>A | Hom | 96.88 | 64 | 35.29 | 254.02 | 1.41 | **\*Accept** | **Yes** | Concordant |
| 402 | S-96 | *WDR45* | c.1007\_1008delAT | Het | 55.39 | 204 | 34.00 | 232.49 | 1.61 | Accept | No |  |
| 403 | S-2563 | *WDR62* | c.1959-2A>G | Hom | 98.23 | 113 | 34.93 | 254.03 | 0.33 | **\*Accept** | **Yes** | Concordant |
| 404 | S-5186 | *WWOX* | c.735dupT | Hom | 99.09 | 110 | 31.24 | 254.01 | 0.65 | Accept | No |  |
| 405 | S-2908 | *ZEB2* | c.1791\_1813dup | Het | 33.00 | 260 | 33.97 | 254.15 | 1.54 | Accept | No |  |
| 406 | S-2656 | *ZFYVE26* | c.6692T>A | Hom | 97.89 | 95 | 34.42 | 254.00 | 0.34 | Accept | No |  |

**Abbreviations:**

ABQ-Average base quality; AMQ-Average mapping quality; Hemi- Hemizygous; Het-Heterozygous; Hom-Homozygous; SB-Strand bias; SR-Supporting reads; TR-Total reads

\*: Sanger already performed as this sample was used as a positive control for Sanger sequencing of family members

**Table S2: List of structural variants identified in the study along with NGS quality parameters. All the variants were assessed for Zscore and copy number value. Variants with the following quality parameters were classified as high confidence variants; Zscore ≥ 5, and CNV ≤1.2 (for a Het call) and ≤0.5 (for a Hom/Hemi call), and these variants were excluded from second method validation (SMV) confirmation.**

|  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Sl.No.** | **Sample ID** | **Gene/**  **Genomic region** | **Chr. coordinates** | **Ex(s)/Gene(s)** | **Zygosity** | **Z-score** | **CN-values** | **Analysis Label** | **Decision on SMV** | **SMV Status** | **SMV Result** |
| 1 | S-2588 | *ATM* | chr11:108137898-?\_108138069+?del | Ex 17 del | Het | 9.78 | 0.65 | Genuine | Accept | No |  |
| 2 | S-3831 | *ATP8A2* | chr13:26273311-?\_26436546+?del | Ex 25-33 del | Hom | 14.8 | 0.06 | Genuine | Accept | No |  |
| 3 | S-4670 | *BCL11A* | chr2:60687539-?\_60780405+?del | Ex 1-4 del | Het | 8.2 | 0.73 | Genuine | **\*Accept** | **Yes** | Concordant |
| 4 | S-246 | *CAPN3* | chr15:42701501-?\_42703971+?del | Ex 17-24 del | Het | 5.5 | 0.96 | Genuine | Accept | No |  |
| 5 | S-4663 | *CLN3* | chr16:28493426-?\_28493993+?del | Ex 11-14 del | Hom | 14.34 | 0.0001 | Genuine | Accept | No |  |
| 6 | R-0109 | *CYB5R3* | chr22:43019795-?\_43024287+?del | Ex 5-8 del | Hom | 12.9 | 0.0002 | Genuine | Accept | No |  |
| 7 | S-3869 | *DMD* | chrX:31947713-?\_31986631+?del | Ex 45-47 del | Hemi | 10.6 | 0.3 | Genuine | Accept | No |  |
| 8 | S-5422 | *DMD* | chrX:31947713-?\_31986631+?del | Ex 45-47 del | Hemi | 12.7 | 0.18 | Genuine | **\*Accept** | **Yes** | Concordant |
| 9 | S-1083 | *DMD* | chrX:31947713-?\_31986631+?del | Ex 45-47 del | Hemi | 12.6 | 0.13 | Genuine | Accept | No |  |
| 10 | S-4696 | *DMD* | chrX:31893305-?\_31986631+?del | Ex 45-48 del | Hemi | 16.5 | 0.03 | Genuine | Accept | No |  |
| 11 | S-1130 | *DMD* | chrX:31854835-?\_31986631+?del | Ex 45-49 del | Hemi | 12.8 | 0.15 | Genuine | Accept | No |  |
| 12 | S-224 | *DMD* | chrX:31747748-?\_31950344+?del | Ex 46-52 del | Hemi | 13.02 | 0.17 | Genuine | Accept | No |  |
| 13 | S-4073 | *DMD* | chrX:31645790-?\_31697703+?del | Ex 53-55 del | Hemi | 8.17 | 0.18 | Genuine | Accept | No |  |
| 14 | S-4015 | *DMD* | chrX:31525398-?\_31525570+?del | Ex 56 del | Hemi | 5.7 | 0.0002 | Genuine | Accept | No |  |
| 15 | S-130 | *DMD* | chrX:31196786-?\_31341775+?del | Ex 62-70 del | Hemi | 8.33 | 0.06 | Genuine | Accept | No |  |
| 16 | S-776 | *EMD* Ex 2-6 del along with 17 bp insertion and *FLNA* Ex 14-47 inverted dup | chrX:153608050?\_153609557+?delinsTGCAGTTCACTGCAATCinsNC\_000023.10:g.153577217-?\_153592740inv | *EMD* Ex 2-6 del along with 17 bp insertion and *FLNA* Ex 14-47 inverted dup | Hemi | 7.3 (EMD), 13.3 (FLNA) | 0.034  (EMD),  4.35  (FLNA) | Likely to be genuine | Check | Yes | Concordant |
| 17 | S-4491 | *GALC* | chr14:88401076-?\_88417092+?del | Ex 11-17 del | Hom | 14.1 | 0.34 | Genuine | **\*Accept** | **Yes** | Concordant |
| 18 | S-1035 | *IGHMBP2* | chr11:68696651-?\_68707199+?del | Ex 8-15 del | Hom | 16.69 | 0.0001 | Genuine | **\*Accept** | **Yes** | Concordant |
| 19 | [S-5508](file:///C:\..\aparna\Reports\Hyperlinking_Aug2017_May2018\STRAN-2018-5011.pdf) | *MECP2* | chrX:153295818-?\_153298008+?del | Ex 3-4 del | Het | 10.76 | 1.21 | Genuine | Accept | No |  |
| 20 | S-2564 | *NIPBL* | chr5:36958206-?\_36958333+?del | Ex 4 del | Het | 7.8 | 0.94 | Genuine | **\*Accept** | **Yes** | Concordant |
| 21 | R-0520 | *NIPBL* | chr5:36995724-?\_37038840+?del | Ex 11-34 del | Het | 5.8 | 1.25 | Genuine | **\*Accept** | **Yes** | Concordant |
| 22 | S-5367 | *NPHP1* | chr2:110881368-?\_110962545+?del | Ex 1-20 del | Hom | 12.2 | 0.13 | Genuine | **\*Accept** | **Yes** | Concordant |
| 23 | S-6113 | *NRXN1* | chr2:50692580-?\_51149018+?del | Ex 6-18 del | Het | 6.81 | 1.09 | Genuine | Accept | No |  |
| 24 | S-5906 | *NSD1* | chr5:176636637-?\_176639196+?del | Ex 5 del | Het | 14.3 | 1.03 | Genuine | Accept | No |  |
| 25 | S-2769 | *PAFAH1B1* | chr17:2541583-?\_2585096+?del | Ex 2-11 del | Het | 6.15 | 1.04 | Genuine | Accept | No |  |
| 26 | S-6624 | *PLP1* | chrX:103031924-?\_103045526+?dup | Ex 1-7 dup (whole gene dup) | Hemi | 12.92 | 4.3 | Genuine | Accept | No |  |
| 27 | R-0536 | *ROGDI* | chr16:4847461?\_4848668+?delinsCAG | Ex 7-11 del | Hom | 9.3 | 0.05 | Genuine | **\*Accept** | **Yes** | Concordant |
| 28 | S-2919 | *SCN1A* | chr2:166908229-?\_166915198+?del | Ex 2-6 del | Het | 5.02 | 0.86 | Genuine | Accept | No |  |
| 29 | S-3157 | *TRIP12* | chr2:230656869-?\_230744795+?dup | Ex 2-27 dup | Het | 7.05 | 2.88 | Genuine | Accept | No |  |
| 30 | S-3882 | *TSEN2* | chr3:12531300-?\_12531488+?del | Ex 2 del | Het | 6.6 | 1.16 | Likely to be genuine | Check | Yes | Concordant |
| 31 | S-4436 | *TUSC3* | chr8:15508206-?\_15621757+?del | Ex 3-11 del | Hom | 12.26 | 0.032 | Genuine | Accept | No |  |
| 32 | S-2889 | chr1p36.31-p36.22 | chr1:5923325-?\_12267077+?del | *NPHP4, KCNAB2, ESPN, TNFRSF25, PLEKHG5, TAS1R1, CAMTA1, PER3, UTS2, PARK7, ENO1, CA6, H6PD, PIK3CD, NMNAT1, KIF1B, PGD, PEX14, TARDBP, MASP2, UBIAD1, MTHFR, NPPA, NPPB, PLOD1, MFN2, MIIP* and *TNFRSF1B* | Het | 5.25 | 1 | Genuine | Accept | No |  |
| 33 | S-2857 | chr1p36.33-1p36.23 | chr1:955553-?\_8934967+?del | *AGRN, TNFRSF4, B3GALT6, TAS1R3, CDK11A, GABRD, SKI, PEX10, MMEL1, TP73, NPHP4, KCNAB2, ESPN, TNFRSF25, PLEKHG5, TAS1R1, CAMTA1, PER3, UTS2, PARK7* and *ENO1* | Het | 7.8 | 0.95 | Genuine | Accept | No |  |
| 34 | S-5116 | chr1q43-q44 | chr1:243419476-?\_245027609+?del | *SDCCAG8, AKT3, ZBTB18* and *HNRNPU* | Het | 6.03 | 1.01 | Genuine | **\*Accept** | **Yes** | Concordant |
| 35 | S-2836 | chr2q24.3 | chr2:165946660-?\_169313128+?del | *SCN3A, SCN2A, GALNT3, TTC21B, SCN1A, SCN9A, STK39* and *CERS6* | Het | 8.9 | 1.09 | Genuine | Accept | No |  |
| 36 | S-3496 | chr2q24.3 | chr2:165946660-?\_169038600+?del | *SCN3A, SCN2A, GALNT3, TTC21B, SCN1A, SCN9A* and *STK39* | Het | 7.41 | 0.95 | Genuine | Accept | No |  |
| 37 | S-1887 | chr2q24.3 | chr2:166605291-?\_167168266+?del | *GALNT3, TTC21B, SCN1A* and *SCN9A* | Het | 6.86 | 1.13 | Genuine | Accept | No |  |
| 38 | S-594 | chr2q37.1-q37.3 | chr2:232879545-?\_242795132+?del | *DIS3L2, PRSS56, CHRND, CHRNG, GIGYF2, KCNJ13, NEU2, ATG16L1, SAG, DGKD, UGT1A8, UGT1A10, UGT1A9, UGT1A7, UGT1A6, UGT1A5, UGT1A4, UGT1A3, UGT1A1, COL6A3, MLPH, HES6, PER2, TRAF3IP1, TWIST2, HDAC4, NDUFA10, CAPN10, KIF1A, AGXT, PASK, ANO7, HDLBP, D2HGDH* and *PDCD1* | Het | 6.5 | 1.01 | Genuine | Accept | No |  |
| 39 | S-5221 | chr4q21.21-q22.3 | chr4:78978724-?\_96256782+?del | *FRAS1, BMP2K, ANTXR2, COQ2, HELQ, WDFY3, ARHGAP24, MAPK10, PTPN13, DSPP, DMP1, IBSP, SPP1, PKD2, ABCG2, SNCA, SMARCAD1, PDLIM5, BMPR1B* and *UNC5C* | Het | 6.02 | 1.06 | Genuine | **\*Accept** | **Yes** | Concordant |
| 40 | S-4861 | chr5q14.3 | chr5:86564269-?\_90459717+?del | *RASA1, MEF2C* and *GPR98* | Het | 5.8 | 1.15 | Genuine | **\*Accept** | **Yes** | Concordant |
| 41 | [S-5188](file:///C:\..\aparna\Reports\Hyperlinking_Aug2017_May2018\STRAN-2018-4752.pdf) | chr5q14.3-q15 | chr5:89854713-?\_96253309+?del | *GPR98, NR2F1, TTC37, PCSK1, CAST, ERAP1* and *ERAP2* | Het | 6.55 | 1.06 | Genuine | Accept | No |  |
| 42 | S-582 | chr5q35.2-q35.3 | chr5:175815821-?\_177422934+?del | *HIGD2A, SNCB, TSPAN17, UIMC1, FGFR4, NSD1, SLC34A1, F12, B4GALT7* and *PROP1* | Het | 6.8 | 1.03 | Genuine | Accept | No |  |
| 43 | S-5411 | chr6q27 | chr6:170591962-?\_170881353+?del | *DLL1* and *TBP* | Het | 6.8 | 1.02 | Genuine | **\*Accept** | **Yes** | Concordant |
| 44 | S-3030 | chr7p14.1 | chr7:40172658-?\_40314255+?del | *MPLKIP* and *SUGCT* | Hom | 10.8 | 0.3 | Genuine | Accept | No |  |
| 45 | S-5204 | chr7q11.23 | chr7:72742578-?\_74016760+?dup | *FKBP6, FZD9, MLXIPL, STX1A, ELN, LIMK1, RFC2* and *GTF2IRD1* | Het | 5.74 | 2.95 | Likely to be genuine | Check | Yes | Concordant |
| 46 | S-1592 | chr7q31.2-q31.32 | chr7:116165117-?\_122635688+?del | *CAV1, MET, ST7, CFTR, CTTNBP2, KCND2, TSPAN12, AASS* and *TAS2R16* | Het | 6.52 | 1.06 | Genuine | Accept | No |  |
| 47 | S-4962 | chr8p23.3-p23.1 | chr8:1496860-?\_6728348+?del | *DLGAP2, CLN8, ARHGEF10, CSMD1, MCPH1* and *DEFB1* | Het | 7.9 | 1.01 | Genuine | **\*Accept** | **Yes** | Concordant |
| 48 | S-4289 | chr15q11.2-q13.1 | chr15:23810930-?\_28544662+?del | *MKRN3, MAGEL2, NDN, PWRN1, NPAP1, SNURF, SNRPN, SNORD116-1, SNORD116-10, SNORD115-1, UBE3A, ATP10A, GABRB3, GABRA5, GABRG3, OCA2* and *HERC2* | Het | 8.06 | 1.01 | Genuine | Accept | No |  |
| 49 | S-177 | chr15q24.1-q25.1 | chr15:74630313-?\_78927929+?del | *CYP11A1, CYP1A1, CYP1A2, MPI, NEIL1, ETFA, PSTPIP1, CIB2, DNAJA4, CHRNA5, CHRNA3* and *CHRNB4* | Het | 5.3 | 1.06 | Genuine | Accept | No |  |
| 50 | S-3969 | chr16p13.11-p12.3 | chr16:15758636-?\_17564653+?del | *NDE1, MYH11, ABCC1, ABCC6* and *XYLT1* | Het | 9.3 | 0.8 | Genuine | Accept | No |  |
| 51 | S-3888 | chr17p11.2 | chr17:16842861-?\_19871774+?del | *TNFRSF13B, FLCN, PEMT, RAI1, SREBF1, ATPAF2, MYO15A, LLGL1, SHMT1, B9D1, SLC47A1, ALDH3A2, SLC47A2* and *AKAP10* | Het | 7.3 | 1.01 | Genuine | Accept | No |  |
| 52 | S-3209 | chr17p12 | chr17:14110127-?\_15164044+?del | *COX10, PMP22* | Het | 9.2 | 0.8 | Genuine | Accept | No |  |
| 53 | S-6580 | chr17p12 | chr17:14110127-?\_15162510+?dup | *COX10, PMP22* | Het | 5.76 | 2.8 | Genuine | Accept | No |  |
| 54 | S-6503 | chr17p12 | chr17:14110127-?\_15162510+?dup | *COX10, PMP22* | Het | 7.45 | 3.04 | Genuine | Accept | No |  |
| 55 | S-5648 | chr17p13.3-p13.2 | chr17:2568666-?\_3422144+?del | *PAFAH1B1, ASPA* and *TRPV3* | Het | 6.13 | 1.02 | Genuine | **\*Accept** | **Yes** | Concordant |
| 56 | S-3191 | chr18q12.1-q12.2 | chr18:28574436-?\_33848648+?del | *DSC3, DSC2, DSG1, DSG4, DSG2, TTR, MEP1B, DTNA, ZNF24, ELP2* and *MOCOS* | Het | 6.43 | 1.03 | Genuine | Accept | No |  |
| 57 | [S-6017](file:///C:\..\aparna\Reports\Hyperlinking_Aug2017_May2018\STRAN-2018-4545.pdf) | chr18q21.2 | chr18:50278424-?\_53303128+?del | *DCC* and *TCF4* | Het | 6.99 | 1.02 | Genuine | **\*Accept** | **Yes** | Concordant |
| 58 | S-3206 | chr19p13.3 | chr19:3586493-?\_4517716+?del | *GIPC3, TBXA2R, PIP5K1C, RAX2, ATCAY, MAP2K2, CREB3L3, SH3GL1* and *PLIN4* | Het | 5.973 | 1.00 | Genuine | Accept | No |  |
| 59 | S-338 | chr22q13.31-q13.33 | chr22:44221881-?\_51169740+?del | *SULT4A1, PNPLA3, UPK3A, SMC1B, FBLN1, ATXN10, PPARA,TRMU, CELSR1, BRD1, ALG12, IL17REL, MLC1,TUBGCP6, SCO2, TYMP, CPT1B, CHKB, ARSA* and *SHANK3* | Het | 5.9 | 1.1 | Genuine | Accept | No |  |
| 60 | S-4467 | chrXp22.13 | chrX:18525217-?\_18690188+?del | *CDKL5* and *RS1* | Het | 9.3 | 0.99 | Genuine | Accept | No |  |
| 61 | S-4223 | chrXq22.1-q22.2 | chrX:102471082-?\_103268232+?dup | *BEX4, PLP1* and *H2BFWT* | Hemi | 9.2 | 3.97 | Genuine | Accept | No |  |

**Abbreviations:**

CN-copy number; Del-Deletion; Dup-Duplication; Hemi-Hemizygous; Het-Heterozygous; Hom-Homozygous

\*: Quantitative polymerase chain reaction (qPCR) already performed as this sample was used as a positive control for qPCR of family members

**Table S3: List of ‘pathogenic’/‘likely pathogenic’ single nucleotide and indel variants detected in the study.**

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Sl. No** | **Sample ID** | **Disease conditions** | **Gene** | **Ex/**  **Int** | **Nucleotide** | **Protein** | **VT** | | **CS** | **Zygosity** | **Control** | **Clinical details** | **Sv** | **F** | **M** | **Sibl** | **MOI** | **Reference** |
| 1 | R-4266 | AAA syndromeo | *AAAS* | Ex 1 | c.43C>A | p.Gln15Lys | MS | | P | Hom | 4θ/ >2500 | Features suggestive of AAA syndrome | - | - | - | - | AR | \*15666842 |
| 2 | S-3752 | AAA syndromeo | *AAAS* | Ex 9 | c.855\_856delinsTT | p.Arg286Ter | NS | | P | Hom | 0/ >2500 | Decreased lacrimation, adrenal insufficiency and achalasia | - | - | - | - | AR | \*11159947 |
| 3 | S-2407 | Leukodystrophy | *ABCD1* | Ex 1 | c.443A>G | p.Asn148Ser | MS | | LP | Hemi | 0/ >2500 | Hyperpigmentation, memory loss and elevated VLCFA. MRI showed leukodystrophy | - | - | - | - | XL | \*11063720 |
| 4 | S-2545 | Leukodystrophy | *ABCD1* | Ex 1 | c.459C>A | p.Tyr153Ter | NS | | P | Hemi | 0/ >2500 | Addison disease, high VLCFA and features suggestive of X-linked adrenoleukodystrophy | - | - | - | - | XL | In this study |
| 5 | S-5919 | Leukodystrophy | *ABCD1* | Ex 1 | c.712delG | p.Ala238ProfsTer98 | Indel | | LP | Hemi | 0/ >2500 | Minimal hyperpigmentation, short stature and impending adrenal insufficiency | - | - | - | - | XL | In this study |
| 6 | S-6559 | Leukodystrophy, ID | *ABCD1* | Ex 1 | c.796G>A | p.Gly266Arg | MS | | P | Hemi | 0/ >2500 | Encephalopathy, delayed motor development, dysarthria, global developmental delay, failure to thrive, spasticity, cognitive deficit, ID, optic atrophy and hearing loss. Suspected of adrenoleukodystrophy | Yes | - | - | Ref (u) | XL | \*21700483 |
| 7 | S-3648 | Leukodystrophy | *ABCD1* | Int 1 | c.901-1G>A | NA | SS | | P | Hemi | 0/ >2500 | Progressive darkening of skin, deterioration of scholastic performance, leukodystrophy, worsening of handwriting and increased levels of VLCFA indicative of leukodystrophy | Yes | - | Het | - | XL | \*26454440 |
| 8 | S-2936 | Microcephaly | *ADAR* | Ex 2 | c.788\_791delACAG | p.Asp263ValfsTer31 | Indel | | LP | Het | 0/ >2500 | Developmental delay, microcephaly and Dandy-Walker malformations | - | - | - | - | AD | In this study |
| 9 | S-5239 | Epilepsy, Neuropathy | *ADSL* | Ex 12 | c.1339T>C | p.Ser447Pro | MS | | LP | Het | 0/ >2500 | Motor delay, hypotonia, recurrent chesty episode, severe motor axonal neuropathy, weakness in left leg, central hypothyroidism and seizures | - | - | - | - | AR | \*12016589 |
| Second variant not detected | | | | | | | | | | | | | | | |
| 10 | S-5981 | Joubert syndromeo | *AHI1* | Ex 8 | c.988delinsTAAATAATA | p.Asp330Ter | NS | | P | Hom | 0/ >2500 | Abnornal eye movements diffuse hypotonia, developmental delay and MRI revealed cerebellar vermian hypoplasia and molar tooth sign | Yes | - | - | - | AR | #rs777241163 |
| 11 | S-1948 | Joubert syndromeo | *AHI1* | Int 8 | c.1152-1G>C | NA | SS | | LP | Hom | 0/ >2500 | Global developmental delay, microcephaly, central hypotonia, oculomotor apraxia, poor visual function. MRI showed molar tooth sign and features suggestive of Joubert syndrome | - | - | - | - | AR | In this study |
| 12 | S-4566 | Joubert syndromeo | *AHI1* | Ex 15 | c.2168G>A | p.Arg723Gln | MS | | P | Het | 0/ >2500 | Ataxia and brain malformation | Yes | Het | Ref | - | AR | \*16453322 |
|  | c.2212C>T | p.Arg738Ter | NS | | P | Het | 0/ >2500 | Yes | Ref | Het | - | AR | \*16453322 |
| 13 | S-4350 | SLSo | *ALDH3A2* | Ex 1 | c.126delG | p.Thr43ArgfsTer64 | Indel | | P | Hom | 1θ/ >2500 | Features suggestive of SLS | - | - | - | - | AR | In this study |
| 14 | S-4199 | Epilepsy | *ALDH7A1* | Int 1 | c.193-12T>G | NA | SS | | VUS | Het | 0/ >2500 | Seizures | - | - | - | - | AR | #rs770632526 |
| Ex 15 | c.1411\_1412insG | p.Leu471ArgfsTer4 | Indel | | LP | Het | 0/ >2500 | - | - | - | - | AR | #rs772766995 |
| 15 | S-2952 | Epilepsy | *ALDH7A1* | Ex 14 | c.1279G>C | p.Glu427Gln | MS | | P | Het | 0/ >2500 | Epilepsy | - | - | - | - | AR | \*19128417 |
| Second variant not detected | | | | | | | | | | | | | | | |
| 16 | S-4679 | Epilepsy, ASD | *ALG13* | Ex 3 | c.320A>G | p.Asn107Ser | MS | | P | Het | 0/ >2500 | Refractory epilepsy, infantile spasms, atonic falls, secondary autism, developmental delay and hypotonia during infancy | - | - | - | - | XL | \*26138355 |
| 17 | S-2593 | Leukodystrophy, Spasticity | *ALS2* | Ex 4 | c.880\_881delCT | p.Leu294CysfsTer4 | Indel | | LP | Hom | 0/ >2500 | Progressive dystonia, difficulty in swallowing, slurring of speech, difficulty in walking, bilateral hypertonia, brisk tendon reflexes and bilateral ankle clonus | Yes | Het | Het | Hom (2a) | AR | In this study |
| 18 | S-1091 | Spasticity | *ALS2* | Ex 21 | c.3415C>T | p.Arg1139Ter | NS | | P | Hom | 0/ >2500 | Delayed walking, progressive gait and bilateral spastic paraplegia | Yes | Het | Het | - | AR | \*27159321 |
| 19 | S-2989 | Dystonia | *ANG* | Ex 2 | c.186\_190dupCTGCA | p.Lys64ThrfsTer32 | Indel | | LP | Het | 2µ/ >2500 | Mild global developmental delay, febrile illness, dyskinesia, dystonia and regression of milestones | - | - | - | - | AD | #rs747227363 |
| 20 | S-3198 | Muscular dystrophy | *ANO5* | Ex 16 | c.1733T>C | p.Phe578Ser | MS | | LP | Hom | 3θ/ >2500 | Proximal muscle weakness, calf muscle hypertrophy, inability to walk and increased CPK levels suggestive of LGMD | - | - | - | - | AR | \*25891276 |
| 21 | S-3469 | Epilepsy, Microcephaly, ID | *AP4M1* | Int 1 | c.59-1G>C | NA | SS | | P | Het | 0/ >2500 | Global developmental delay, microcephaly, ID, intrauterine growth retardation, prominent eye, preauricular skin tag in right ear and seizure disorder | Yes | - | - | - | AR | #rs761873506 |
| Ex 11 | c.901delT | p.Ser301LeufsTer15 | Indel | | P | Het | 0/ >2500 | Yes | - | - | - | AR | In this study |
| 22 | S-3687 | Ataxia | *APTX* | Ex 7 | c.596delG | p.Arg199LeufsTer15 | Indel | | LP | Hom | 2θ/ >2500 | Delayed milestones, gait difficulty and swaying while walking by 6 years of age, foot deformity and squint by 14 years of age and speech difficulty by 16 years of age | - | - | - | - | AR | #rs770007531 |
| 23 | S-5910 | Ataxia | *APTX* | Ex 7 | c.596delG | p.Arg199LeufsTer15 | Indel | | LP | Hom | 2θ/ >2500 | Generalised hypotonia, poor clarity in speech and ataxia. MRI of brain showed cerebellar and brainstem atrophy | - | - | - | - | AR | #rs770007531 |
| 24 | S-3919 | Ataxia | *APTX* | Ex 7 | c.596delG | p.Arg199LeufsTer15 | Indel | | LP | Hom | 2θ/ >2500 | Ataxia, oculomotor apraxia and abnormal gait | - | - | - | - | AR | #rs770007531 |
| 25 | S-753 | Ataxia | *APTX* | Int 8 | c.875-2A>G | NA | SS | | LP | Hom | 0/ >2500 | Cerebellar ataxia with oculomotor apraxia | Yes | Het | Het | - | AR | In this study |
| 26 | S-1347 | Coffin-Siris syndromeo | *ARID1B* | Ex 20 | c.5547delC | p.Leu1850Ter | NS | | P | Het | 0/ >2500 | Features suggestive of Coffin-Siris syndrome | - | - | - | - | AD | In this study |
| 27 | S-1763 | Leukodystrophy | *ARSA* | Ex 2 | c.346C>T | p.Arg116Ter | NS | | P | Hom | 1θ/ >2500 | Delayed milestones and features suggestive of leukodystrophy | - | - | - | - | AR | \*19021637 |
| 28 | S-2325 | Leukodystrophy | *ARSA* | Ex 5 | c.877C>T | p.Arg293Ter | NS | | P | Hom | 0/ >2500 | Regression of motor milestones. MRI was suggestive of Krabbe disease | - | - | - | - | AR | \*26553228 |
| 29 | S-2212 | Ataxia, Leukodystrophy | *ARSA* | Ex 5 | c.917C>T | p.Thr306Met | MS | | P | Hom | 0/ >2500 | Neuroregression with frequent falls, ataxia, speech regression, nystagmus and seizures | Yes | Het | Het | - | AR | \*26915897 |
| 30 | S-3364 | Leukodystrophy | *ARSA* | Ex 5 | c.917C>T | p.Thr306Met | MS | | P | Hom | 0/ >2500 | Developmental delay, severe neuroregression, loss of head control, seizures and inability to walk. MRI was suggestive of leukodystrophy | Yes | Het | Het | - | AR | \*26915897 |
| 31 | S-4598 | Leukodystrophy | *ARSA* | Ex 5 | c.917C>T | p.Thr306Met | MS | | P | Het | 0/ >2500 | Regression in motor milestones and speech, spasticity and hypertonia. | Yes | Ref | Het | - | AR | \*26915897 |
| Ex 5 | c.928G>A | p.Gly310Ser | MS | | VUS | Het | 1θ/ >2500 | Yes | Het | Ref | - | AR | In this study |
| 32 | S-3145 | Leukodystrophy | *ARSA* | Ex 5 | c.931G>A | p.Gly311Ser | MS | | LP | Hom | 0/ >2500 | Hypotonia, neuroregression and typical MRI pattern suggestive of metachromatic leukodystrophy | - | - | - | - | AR | \*28670130 |
| 33 | S-3827 | Epilepsy | *ARX* | Ex 4 | c.1425\_1446del | p.Phe476GlyfsTer9 | Indel | | LP | Het | 0/ >2500 | Seizures, static encephalopathy, global developmental delay, intellectual and cortical visual dysfunction, latent nystagmus and esotropia | - | - | Het | - | XL | In this study |
| 34 | S-6032 | Epilepsy, Microcephaly | *ASNS* | Int 11 | c.1321-2A>G | NA | SS | | LP | Hom | 0/ >2500 | Microcephaly, generalized seizures, hypomyelination, global developmental delay, hyperglycemia, hypoinsulinemia, respiratory distress and succumbed on 65th day. | Yes | Het | Het | Het (u) | AR | In this study |
| 35 | S-887 | Microcephaly | *ASPM* | Ex 1 | c.277delC | p.Gln93SerfsTer26 | Indel | | LP | Hom | 0/ >2500 | Microcephaly, mental retardation, unclear speech, behavioural problems and impaired motor functions | Yes | - | - | Het (u) | AR | In this study |
| 36 | S-1362 | Microcephaly | *ASPM* | Ex 3 | c.900delC | p.Asn301ThrfsTer6 | Indel | | P | Het | 0/ >2500 | Features suggestive of primary microcephaly | - | - | - | - | AR | In this study |
| *ASPM* | Ex 3 | c.1729\_1730delAG | p.Ser577ArgfsTer33 | Indel | | P | Het | 0/ >2500 | - | - | - | - | AR | \*14574646 |
| 37 | S-1339 | Microcephaly | *ASPM* | Ex 18 | c.7125\_7128dupACTG | p.Gln2377ThrfsTer26 | Indel | | P | Hom | 1θ/ >2500 | Primary microcephaly, speech delay and hyperactive with DQ of 67-74 | - | - | - | - | AR | ΔRCV000145177.4 |
| 38 | S-5189 | Ataxia | *ATM* | Ex 5 | c.387delA | p.Asp130IlefsTer23 | Indel | | P | Hom | 0/ >2500 | Gait imbalance, weakness, slurry speech, drooling. Brain MRI suggestive of cerebral atrophy | - | - | - | - | AR | ΔRCV000255124.1 |
| 39 | S-5948 | Ataxia | *ATM* | Int 32 | c.4909+1G>A | NA | SS | | P | Hom | 0/ >2500 | Truncal ataxia, hypotonia, developmental delay and hypoplastic right vertebral artery | - | - | - | - | AR | \*9887333 |
| 40 | S-4309 | Ataxia-telangiectasia | *ATM* | Int 41 | c.6096-2A>G | NA | SS | | LP | Hom | 0/ >2500 | Problems with balance and hand coordination, involuntary jerky movements and lung infection | - | - | - | - | AR | In this study |
| 41 | S-2588 | Ataxia-telangiectasia | *ATM* | Ex 58 | c.8545C>T | p.Arg2849Ter | NS | | P | Het | 0/ >2500 | Delayed onset of walking and inability to balance and speak | - | - | - | - | AR | \*17124347 |
| Ex 17 del | c.(2466+1\_24671)\_(2638+1\_2639-1)del | NA | Large del | | NA | Het | 0/ >2500 | - | - | - | - | AR | In this study |
| 42 | S-2051 | Ataxia-telangiectasia | *ATM* | Ex 62 | c.8873\_8874delTT | p.Phe2958Terfs | Indel | | P | Hom | 0/ >2500 | Delayed motor milestones, progressive ataxia, oculomotor apraxia, generalized dystonia, hyporeflexia, recurrent lower respiratory tract infections and ocular telangiectasia | - | - | - | - | AR | ΔRCV000206073.1 |
| 43 | R-0549 | Lissencephalyo | *ATP1A2* | Ex 20 | c.2723G>A | p.Arg908Gln | MS | | LP | Het | 0/ >2500 | Sudden onset of seizures at three months of age, with delayed milestones. MRI suggestive of pachygyria | - | - | - | - | AD | \*25948653 |
| 44 | R-4319 | AHCo | *ATP1A3* | Ex 17 | c.2401G>A | p.Asp801Asn | MS | | P | Het | 0/ >2500 | Hypotonia, significant speech and motor delay suggestive of AHC | Yes | Ref | Ref | - | AD | \*22850527 |
| 45 | S-590 | Epilepsy | *ATP1A3* | Ex 17 | c.2401G>A | p.Asp801Asn | MS | | P | Het | 0/ >2500 | Refractory left focal seizures and global developmental delay | - | - | - | - | AD | \*22850527 |
| 46 | S-4078 | AHCe | *ATP1A3* | Ex 17 | c.2401G>A | p.Asp801Asn | MS | | P | Het | 0/ >2500 | Seizures, postictal hemiparesis, developmental delay and transient weakness of the upper limb. | Yes | Ref | Ref | - | AD | \*24842602 |
| 47 | S-2402 | AHCa,e | *ATP1A3* | Ex 18 | c.2443G>A | p.Glu815Lys | MS | | P | Het | 0/ >2500 | Ataxia, epilepsy, alternating episodes of hemiplegia and cognitive impairment. | - | - | - | - | AD | \*24842602 |
| 48 | S-2223 | AHCo | *ATP1A3* | Ex 21 | c.2839G>A | p.Gly947Arg | MS | | P | Het | 0/ >2500 | Features suggestive of AHC | - | - | - | - | AD | \*26410222 |
| 49 | S-2993 | Epilepsy | *ATP1A3* | Ex 21 | c.2839G>A | p.Gly947Arg | MS | | P | Het | 0/ >2500 | Encephalopathy, global developmental delay, hypertonia, delayed speech, cerebral palsy, lethargy, round facies and seizures | - | - | - | - | AD | \*26410222 |
| 50 | S-3386 | Dystonia, ID | *ATP8A2* | Int 31 | c.3019-1G>A | NA | SS | | LP | Hom | 0/ >2500 | Global developmental delay and dystonia | - | - | - | - | AR | In this study |
| 51 | S-2049 | Dystonia | *AUH* | Int 4 | c.505+1G>C | NA | SS | | LP | Het | 1θ/ >2500 | Slurring of speech and multifocal dystonia | - | - | - | - | AR | #rs773652620 |
| Second variant not detected | | | | | | | | | | | | | | | |
| 52 | R-0597 | Costello syndromeo | *BRAF* | Ex 16 | c.1914T>G | p.Asp638Glu | MS | | LP | Het | 0/ >2500 | Developmental delay, failure to thrive, seizures, ID, frontal bossing, macrocephaly and occular nystagmus. | Yes | - | Ref | - | AD | \*21063443 |
| 53 | S-4749 | Microcephaly, Dystonia | *BTD* | Ex 2 | c.98\_104delinsTCC | p.Cys33PhefsTer36 | Indel | | P | Het | 1θ/ >2500 | Cerebellar atrophy, hypomyelination, infantile spasm, global developmental delay, dystonia, hypotonia, microcephaly, visual impairment and hearing loss | - | - | - | - | AR | \*28640880 |
| Second variant not detected | | | | | | | | | | | | | | | |
| 54 | S-2918 | Epilepsy | *BUB1B* | Ex 7 | c.933\_951del | p.Arg317LeufsTer8 | Indel | | LP | Het | 0/ >2500 | Seizures since two years of age, developmental delay and nystagmus. MRI shows cerebellar atrophy | - | - | - | - | AD | In this study |
| 55 | S-2832 | ASD | *C12orf57* | Ex1 | c.19C>T | p.Gln7Ter | NS | | LP | Hom | 0/ >2500 | Global developmental delay, seizures, upslanting of eyes, bilateral partial ptosis, hypertelorism, hyperactivity and autistic traits. | - | - | - | - | AR | #rs782502903 |
| 56 | S-3353 | Neuropathy, Spasticity | *C12orf65* | Ex 3 | c.346delG | p.Val116Ter | NS | | P | Hom | 0/ >2500 | Visual disturbance, frequent falls, progressive gait difficulty, oculomotor apraxia, spasticity, cognitive deficit and peripheral neuropathy | - | - | - | - | AR | \*24198383 |
| 57 | S-3673 | Muscular dystrophy | *CAPN3* | Ex 3 | c.383delA | p.Asp128AlafsTer11 | Indel | | LP | Hom | 0/ >2500 | Atrophy of deltoid muscles, lower motor neuron syndrome, weakness of both upper and lower limbs. Muscle biopsy showed normal staining for dystrophin, sarcoglycan, dysferlin and merosin. | - | - | - | - | AR | In this study |
| 58 | S-5902 | Muscular dystrophy | *CAPN3* | Ex 5 | c.701G>A | p.Gly234Glu | MS | | P | Het | 0/ >2500 | Proximal muscle weakness, particularly in pelvis and shoulder girdle group. Muscle biopsy revealed dysferlinopathy | - | - | - | - | AR | \*15689361 |
| Ex 8 | c.1063C>T | p.Arg355Trp | MS | | P | Het | 0/ >2500 | - | - | - | - | AR | \*17994539 |
| 59 | S-3642 | Muscular dystrophy | *CAPN3* | Int 5 | c.802-9G>A | NA | SS | | P | Hom | 0/ >2500 | Difficulty getting up from the floor and climbing stairs, waddling gait, mild pseudo-hypertrophy of calf muscle, hypertelorism, low set ears, and prominent cheeks. Muscle biopsy was indicative of muscular dystrophy | - | - | - | - | AR | \*17979987 |
| 60 | S-4649 | Muscular dystrophy | *CAPN3* | Int 18 | c.2051-1G>T | NA | SS | | P | Het | 1θ/ >2500 | Muscular dystrophies and congenital myopathies | - | - | - | - | AR | \*23666804 |
| Ex 22 | c.2338G>C | p.Asp780His | MS | | P | Het | 3θ/ >2500 | - | - | - | - | AR | \*23666804 |
| 61 | S-4102 | Muscular dystrophy | *CAPN3* | Int 20 | c.2185-2A>G | NA | SS | | P | Hom | 0/ >2500 | Muscular dystrophies | - | - | - | - | AR | \*10330340 |
| 62 | S-3597 | Muscular dystrophy, Dystonia | *CAPN3* | Ex 21 | c.2243G>A | p.Arg748Gln | MS | | P | Hom | 1θ/ >2500 | Generalised dystonia, behavioural issues, anger outbursts and progressive pancerebellar features | - | - | - | - | AR | \*27020652 |
| 63 | S-3859 | LGMDmd | *CAPN3* | Ex 21 | c.2243G>A | p.Arg748Gln | MS | | P | Hom | 1θ/ >2500 | Toe walking and other features suggestive of LGMD | - | - | - | - | AR | \*9762961 |
| 64 | S-707 | Muscular dystrophy | *CAPN3* | Ex 22 | c.2338G>C | p.Asp780His | MS | | P | Hom | 3θ/ >2500 | Slowly progressive proximal myopathy and hyperthyroidism | - | - | - | - | AR | \*23666804 |
| 65 | S-2936 | Microcephaly | *CASK* | Ex 2 | c.79C>T | p.Arg27Ter | NS | | P | Het | 0/ >2500 | Developmental delay, microcephaly and Dandy-Walker malformations | - | - | - | - | XL | \*21735170 |
| 66 | S-4099 | Neuropathy | *CD59* | Ex 4 | c.21delT | p.Val8SerfsTer72 | Indel | | LP | Het | 0/ >2500 | Thalamic hyperintensity, hypotonia, recurrent flaccid weakness triggered by fever, inability to move upper and lower limbs, areflexia, gaze palsy and recurrent neuropathy | - | - | - | - | AR | \*\*gnomAD |
| CD59 | Ex 6 | c.221\_223delACG | p.Asp74del | Indel | | VUS | Het | 0/ >2500 | - | - | - | - | AR | In this study |
| 67 | S-4086 | Epilepsy, Microcephaly | *CDK5RAP2* | Ex 12 | c.1139C>A | p.Ser380Ter | NS | | LP | Het | 0/ >2500 | Global developmental delay, microcephaly, spastic quadriparesis and failure to thrive, suspected of West syndrome | Yes | Ref | Het | - | AR | In this study |
| Second variant not detected | | | | | | | | | | | | | | | |
| 68 | S-488 | Microcephaly, ASD | *CDKL5* | Ex 12 | c.1842T>A | p.Tyr614Ter | NS | | LP | Het | 0/ >2500 | Global developmental delay, seizures, poor eye contact, autistic traits, microcephaly, short nose and short philtrum | - | - | - | - | XL | In this study |
| 69 | S-4826 | Epilepsy, Microcephaly | *CEP135* | Int 4 | c.473-1G>C | NA | SS | | LP | Het | 2 θ/ >2500 | Global developmental delay, seizures and microcephaly. Brain MRI revealed cerebral atrophy | - | - | - | - | AR | #rs367650085 |
| Second variant not detected | | | | | | | | | | | | | | | |
| 70 | S-1775 | Joubert syndromeo | *CEP290* | Ex 22 | c.2365C>T | p.Gln789Ter | NS | | P | Het | 0/ >2500 | Manifested features suggestive of Joubert syndrome. Brain MRI showed cerebellar vermis agenesis | - | - | - | - | AR | In this study |
| Ex 41 | c.5668G>T | p.Gly1890Ter | NS | | P | Het | 3θ/ >2500 | - | - | - | - | AR | \*17564967 |
| 71 | S-1400 | Joubert syndromeo | *CEP290* | Ex 33 | c.4219delT | p.Trp1407GlyfsTer12 | Indel | | LP | Hom | 0/ >2500 | Global developmental delay since early infancy with delayed speech, nystagmus, no vision since 5 months of age, marked hypotonia and pigmentary dystrophy; ultrasound suggestive of renal disease | - | - | - | - | AR | In this study |
| 72 | S-1848 | Joubert syndromeo | *CEP290* | Ex 36 | c.4792A>T | p.Lys1598Ter | NS | | LP | Hom | 0/ >2500 | Developmental delay, hypotonia and seizure disorders. MRI suggestive of Jouberts syndrome | Yes | Het | Het | - | AR | In this study |
| 73 | S-4631 | Joubert syndromeo | *CEP290* | Ex 41 | c.5649delA | p.Lys1883AsnfsTer2 | Indel | | LP | Hom | 1θ/ >2500 | Developmental delay, dysmorphism, exotropia, polycystic kidneys, third nerve palsy and hypertelorism. | Yes | Het | Het | - | AR | In this study |
| 74 | S-2563 | Joubert syndromeo | *CEP290* | Ex 42 | c.5766\_5769delAGAA | p.Ile1922MetfsTer7 | Indel | | LP | Hom | 0/ >2500 | Features indicative of Joubert syndrome. | - | - | - | Hom (unk) | AR | In this study |
| 75 | S-6496 | Epilepsy | *CHD2* | Ex 38 | c.4909C>T | p.Arg1637Ter | NS | | P | Het | 0/ >2500 | Delay in milestones and complex partial seizures | - | - | - | - | AD | #rs864309547 |
| 76 | S-3110 | CHARGE syndromeo | *CHD7* | Ex 2 | c.1611G>A | p.Trp537Ter | NS | | P | Het | 0/ >2500 | Features suggestive of CHARGE syndrome | - | - | - | - | AD | \*18074359 |
| 77 | S-3773 | CHARGE syndromeo | *CHD7* | Ex 21 | c.4756\_4757delAA | p.Lys1586AlafsTer11 | Indel | | LP | Het | 0/ >2500 | Features suggestive of CHARGE syndrome | - | - | - | - | AD | In this study |
| 78 | S-3056 | Myasthenic syndromeo | *CHRNE* | Ex 2 | c.183\_187dupCTCAC | p.Leu63ProfsTer3 | Indel | | P | Hom | 0/ >2500 | Exercise intolerance, muscle wasting, downslanting palpebral fissures, opthalmoplegia and ptosis | - | - | - | - | AR | \*9158150 |
| 79 | S-5167 | Myasthenic syndromeo | *CHRNE* | Ex 2 | c.183\_187dupCTCAC | p.Leu63ProfsTer3 | Indel | | P | Hom | 0/ >2500 | Droopy eyelids and increased fatigability in the evenings | - | - | - | - | AR | \*9158150 |
| 80 | S-2875 | Myasthenic syndromeo | *CHRNE* | Ex 7 | c.799C>T | p.Gln267Ter | NS | | P | Hom | 0/ >2500 | Features suggestive of congenital myasthenic syndrome | - | - | - | - | AR | In this study |
| 81 | S-3586 | Myasthenic syndromeo | *CHRNE* | Ex 8 | c.878\_887dup | p.Thr297AsnfsTer103 | Indel | | LP | Hom | 0/ >2500 | Features suggestive of congenital myasthenic syndrome | - | - | - | - | AR | In this study |
| 82 | S-876 | Myasthenic syndromeo | *CHRNE* | Ex 10-Int 10 | c.1216\_1219+19del | NA | SS | | LP | Hom | 0/ >2500 | Ptosis, progressive developmental delay, external ophthalmoplegia and recurrent falls | - | - | - | - | AR | In this study |
| 83 | S-1157 | Myasthenic syndromeo | *CHRNE* | Ex 12 | c.1327delG | p.Glu443Lysfs | Indel | | P | Hom | 1θ/ >2500 | Features suggestive of congenital myasthenic syndrome | - | - | - | - | AR | \*10514102 |
| 84 | S-1632 | Myasthenic syndromeo | *CHRNE* | Ex 12 | c.1327delG | p.Glu443Lysfs | Indel | | P | Hom | 1θ/ >2500 | Features suggestive of congenital myasthenic syndrome | - | - | - | - | AR | \*10514102 |
| 85 | S-705 | Myasthenic syndromeo | *CHRNE* | Ex 12 | c.1327delG | p.Glu443Lysfs | Indel | | P | Hom | 1θ/ >2500 | Muscle weakness, abnormal eye movements, drooping eyelids, thinning of facial muscles | - | - | - | - | AR | \*10514102 |
| 86 | S-168 | Myasthenic syndromeo | *CHRNE* | Ex 12 | c.1371delC | p.Cys458Alafs | Indel | | LP | Hom | 0/ >2500 | Ptosis and proximal muscle weakness | - | - | - | - | AR | In this study |
| 87 | S-1860 | Myasthenic syndromeo | *CHRNE* | Ex 12 | c.1371delC | p.Cys458Alafs | Indel | | LP | Hom | 0/ >2500 | Ptosis, double vision, external ophthalmoparesis and proximal lower limb weakness. | - | - | - | - | AR | In this study |
| 88 | S-4641 | ID | *CLCN1* | Ex 1 | c.47G>A | p.Trp16Ter | NS | | LP | Het | 1µ/ >2500 | Developmental delay | - | - | - | - | AD | #rs769092535 |
| 89 | R-0515 | Myotoniao | *CLCN1* | Ex 7 | c.803C>T | p.Thr268Met | MS | | LP | Hom | 0/ >2500 | Delay in walk since birth, low grasping, walk/ speech/ ocular spasm and slow progression | Yes | - | - | - | AR | \*25088311 |
| 90 | S-4721 | Myotoniao | *CLCN1* | Ex 8 | c.908G>A | p.Trp303Ter | NS | | P | Hom | 0/ >2500 | Pain over both lower limbs and hypertrophied calf muscles and myotonia | - | - | - | - | AR | \*24349310 |
| 91 | S-4681 | Muscle hypertrophyo | *CLCN1* | Ex 10 | c.1129C>T | p.Arg377Ter | NS | | P | Hom | 0/ >2500 | Pseudohypertrophy with recent onset weakness, muscle hypertrophy, loss of fat pads and short stature | - | - | - | - | AR | \*17932099 |
| 92 | S-2685 | Myotoniao | *CLCN1* | Ex 14 | c.1495G>A | p.Gly499Arg | MS | | LP | Hom | 0/ >2500 | Muscle stiffness, suggestive of myotonia congenita | Yes | - | - | Ref (u) | AR | \*10644771 |
| 93 | S-690 | Myotoniao | *CLCN1* | Ex 15 | c.1667T>A | p.Ile556Asn | MS | | P | Het | 1θ/ >2500 | Hypertrophy of the calf and biceps, myotonia and brachioradialis suggestive of myotonia congenita | - | - | - | - | AR | \*9736777 |
| Ex 21 | c.2472\_2473dupTC | p.Pro825LeufsTer28 | Indel | | P | Het | 2θ/ >2500 | - | - | - | - | AR | \*9566422 |
| 94 | S-2058 | Muscular dystrophy | *CLCN1* | Ex 16 | c.1876C>T | p.Arg626Ter | NS | | P | Hom | 3θ/ >2500 | Hypotonia, muscle atrophy and Gowers' sign | - | - | - | - | AR | \*23113340 |
| 95 | S-2577 | Myotoniao | *CLCN1* | Ex 21 | c.2472\_2473dupTC | p.Pro825LeufsTer28 | Indel | | P | Hom | 2θ/ >2500 | Difficulty in walking and initiating movements | - | - | - | - | AR | #rs768133575 |
| 96 | S-5686 | Leukodystrophy | *CLCN2* | Ex 18 | c.2068G>T | p.Glu690Ter | NS | | LP | Hom | 0/ >2500 | Brain MRI shows diffuse symmetric T2-hyperintensities involving supratentorial subcortical white matter, internal capsule, corpus callosum and corticospinal tract | - | - | - | - | AR | In this study |
| 97 | S-4390 | Epilepsy | *CLN5* | Ex 3 | c.595C>T | p.Arg199Ter | NS | | P | Hom | 0/ >2500 | Myoclonic jerks, progressive visual blurring and vision loss | - | - | - | - | AR | \*23374165 |
| 98 | S-3224 | NCLe | *CLN5* | Ex 4 | c.746\_747insCA | p.Lys249AsnfsTer16 | Indel | | LP | Hom | 0/ >2500 | Abnormal gait, generalized seizures, ataxia, hypertonia, neuronal regression, progressive vision loss and retinitis pigmentosa | - | - | - | - | AR | In this study |
| 99 | S-3555 | Epilepsy, Ataxia | *CLN6* | Ex 4 | c.476C>T | p.Pro159Leu | MS | | LP | Hom | 0/ >2500 | Myoclonic epilepsy and cerebellar atrophy | - | - | - | - | AR | \*19201763 |
| 100 | S-3731 | NCLe | *CLN6* | Ex 7 | c.775G>C | p.Gly259Arg | MS | | LP | Hom | 0/ >2500 | Neuroregression, cerebellar ataxia, cerebral atrophy, abnormal gait and myoclonic jerks | - | - | - | - | AR | In this study |
| 101 | S-4672 | NCLe | *CLN8* | Ex 2 | c.1A>C | p.Met1? | MS | | LP | Hom | 0/ >2500 | Seizures, regression and imbalance while walking. Brain MRI shows cerebellar atrophy | Yes | - | - | - | AR | In this study |
| 102 | S-5622 | Spasticity | *COL4A1* | Ex 6 | c.337C>T | p.Gln113Ter | NS | | LP | Het | 0/ >2500 | Generalized spasticity, more in the lower limbs and mild speech and cognitive deficit, suggestive of hereditary spastic paraplegia | - | - | - | - | AD | In this study |
| 103 | S-4386 | Neuropathy | *COL4A2* | Ex 34 | c.3205delC | p.Arg1069GlyfsTer29 | Indel | | P | Het | 0/ >2500 | Signs suggestive of Charcot-Marie-Tooth disease | - | - | - | - | AD | \*22333902 |
| 104 | S-3030 | Microcephaly | *COL4A3* | Int 14 | c.828+1G>T | NA | SS | | LP | Hom | 0/ >2500 | Mild developmental delay, trichorrhexis nodosa and facial features indicative of autosomal recessive primary microcephaly. | - | - | - | - | AR | In this study |
| 105 | S-3912 | UCMDmd | *COL6A1* | Ex 3 | c.297delC | p.Ile100SerfsTer48 | Indel | | LP | Hom | 0/ >2500 | Dysmorphic facies, kyphoscoliosis, hyperextensibility of joints and severe muscle wasting, suggestive of UCMD | Yes | Het | Het | - | AR | In this study |
| 106 | S-528 | Muscular dystrophy | *COL6A1* | Ex 9 | c.850G>A | p.Gly284Arg | MS | | P | Het | 0/ >2500 | Delay in attaining motor milestones, history of recurrent falls, and high CPK levels, suggestive of muscular dystrophy. | Yes | - | - | - | AD | \*16130093 |
| 107 | S-2061 | Muscular dystrophy, Myopathy | *COL6A1* | Ex 10 | c.868G>A | p.Gly290Arg | MS | | P | Het | 0/ >2500 | Muscle weakness, myopathy and muscular dystrophy, suggestive of UCMD or Bethlem myopathy. | - | - | - | - | AD | \*19564581 |
| 108 | S-4466 | Myopathy | *COL6A1* | Ex 10 | c.877G>A | p.Gly293Arg | MS | | LP | Het | 0/ >2500 | Motor delay and muscle weakness | Yes | Ref | Ref | - | AD | \*20976770 |
| 109 | S-2761 | Myopathy | *COL6A1* | Int 25 | c.1675-1G>C | NA | SS | | LP | Hom | 0/ >2500 | Delayed motor milestones, hypotonia, proximal muscle weakness, hyperextensible joints and myopathic facies | - | - | - | - | AR | #rs112965881 |
| 110 | S-5468 | Muscular dystrophy | *COL6A2* | Ex 2 | c.38delG | p.Gly13GlufsTer58 | Indel | | LP | Hom | 0/ >2500 | Congenital muscular dystrophy, developmental delay, hypotonia and contractures at knees and elbow | - | - | - | - | AR | In this study |
| 111 | S-5310 | UCMDmd | *COL6A2* | Ex 3 | c.310C>T | p.Gln104Ter | NS | | P | Hom | 0/ >2500 | Myopathic facies, arachnodactyly, high arched palate, hypotonia, elbow and hip contractures | - | - | - | - | AR | \*\*gnomAD |
| 112 | S-1215 | UCMDmd | *COL6A2* | Ex 7 | c.875G>T | p.Gly292Val | MS | | LP | Het | 0/ >2500 | Delayed walking, abnormal gait, recurrent falls and hypotonia | - | - | - | - | AR | \*17785673 |
| Ex 15 | c.1312G>A | p.Asp438Asn | MS | | VUS | Het | 1θ/ >2500 | - | - | - | - | AR | ΔRCV000383359.1 |
| 113 | S-1176 | Muscular dystrophy | *COL6A2* | Ex 15 | c.1270-4\_1273dup  GCAGGGGC | p.Asp428AlafsTer120 | Indel | | LP | Het | 0/ >2500 | Myopathic facies, high arched groove palate, flexon deformity, hyperextensibility of fingers, prominent fetal finger pads, restriction of shoulder joints, narrow chest, proximal contracture. | Yes | Het | Ref | - | AR | In this study |
| Ex 28 | c.2864\_2865delAC | p.Asp955GlufsTer37 | Indel | | LP | Het | 0/ >2500 | Yes | Ref | Het | - | AR | In this study |
| 114 | S-5445 | Periodic paralysiso | *COQ2* | Ex 7 | c.1184dupT | p.Leu395PhefsTer24 | Indel | | LP | Het | 0/ >2500 | Muscle weakness and wheelchair bound | Yes | - | - | - | AD | #rs763059203 |
| 115 | S-3810 | Leukodystrophy | *CSF1R* | Ex 18 | c.2381T>C | p.Ile794Thr | MS | | P | Het | 0/ >2500 | Signs suggestive of leukodystrophy | - | - | - | - | AD | \*27680516 |
| 116 | S-6075 | Krabbe diseasel | *CTC1* | Ex 6 | c.1056\_1069delinsC | p.Ser353ProfsTer10 | Indel | | P | Het | 0/ >2500 | Features suggestive of leukodystrophy | - | - | - | - | AR | In this study |
| Ex 10 | c.1673A>G | p.Glu558Gly | MS | | VUS | Het | 4θ/ >2500 | - | - | - | - | AR | #rs371121503 |
| 117 | S-2427 | Epilepsy, ASD | *CUL4B* | Ex 8 | c.1007\_1011delTTATA | p.Ile336LysfsTer2 | Indel | | LP | Hemi | 0/ >2500 | Neuroregression, global developmental delay, autistic features and mild dysmorphism. | Yes | - | Ref | - | XL | \*17236139 |
| 118 | S-3247 | Ataxia, Neuropathy | *CYP27A1* | Int 3 | c.646+1G>A | NA | SS | | LP | Hom | 0/ >2500 | Tendon xanthomas, bilateral cataract, ataxia, peripheral sensory neuropathy, pes cavus and pancerebellar signs. | - | - | - | - | AR | In this study |
| 119 | S-2300 | Epilepsy | *DCX* | Ex 3 | c.667G>A | p.Gly223Arg | MS | | LP | Hemi | 0/ >2500 | Seizures, developmental delay, ptosis, posteriorly rotated ears and hypotonia | - | - | - | - | XL | ΔRCV000483287.1 |
| 120 | S-3298 | Muscular dystrophy | *DMD* | Ex 8 | c.724C>T | p.Gln242Ter | NS | | P | Hemi | 0/ >2500 | Muscular dystrophy | - | - | - | - | XL | \*20485447 |
| 121 | S-381 | DMDmd | *DMD* | Ex 12 | c.1412delC | p.Thr471LysfsTer16 | Indel | | P | Hemi | 0/ >2500 | Calf hypertrophy, and difficulty in getting up, running, jumping, climbing stairs, suggestive of DMD | Yes | - | Ref | - | XL | \*25612904 |
| 122 | S-4863 | LGMDmd | *DMD* | Ex 14 | c.1693dupA | p.Thr565AsnfsTer2 | Indel | | P | Hemi | 0/ >2500 | Mild motor delay, toe walking and calf muscle hypertrophy, suggestive of LGMD | - | - | - | - | XL | In this study |
| 123 | S-5408 | DMDmd | *DMD* | Ex 29 | c.4018delA | p.Ile1340SerfsTer20 | Indel | | P | Hemi | 0/ >2500 | Pseudohypertrophy of calf muscles and increased CPK levels, suggestive of LGMD | - | - | - | - | XL | In this study |
| 124 | S-35 | DMDmd | *DMD* | Ex 30 | c.4151delA | p.Glu1384GlyfsTer34 | Indel | | P | Hemi | 0/ >2500 | Waddling gait and Gowers' sign, suggestive of DMD | - | - | - | - | XL | \*15643612 |
| 125 | S-5291 | DMDmd | *DMD* | Ex 32 | c.4375C>T | p.Arg1459Ter | NS | | P | Hemi | 0/ >2500 | Increased CPK and features suggestive of DMD | - | - | - | - | XL | \*23453023 |
| 126 | S-5788 | Muscular dystrophy | *DMD* | Ex 36 | c.5140delG | p.Glu1714LysfsTer7 | Indel | | P | Hemi | 0/ >2500 | Gower's sign and bilateral calf muscle hypertrophy suggestive of dystrophinopathies | - | - | - | - | XL | \*20485447 |
| 127 | S-4249 | DMDmd | *DMD* | Ex 41 | c.5899C>T | p.Arg1967Ter | NS | | P | Hemi | 0/ >2500 | Difiiculty in climbing stairs, bilateral calf muscle hypertrophy and increased CPK levels | - | - | - | - | XL | \*20485447 |
| 128 | S-4319 | DMDmd | *DMD* | Ex 42 | c.5963\_5967delACATG | p.Asp1988AlafsTer14 | Indel | | P | Hemi | 0/ >2500 | Signs suggestive of DMD | - | - | - | - | XL | In this study |
| 129 | S-870 | DMDmd | *DMD* | Ex 43 | c.6238C>T | p.Gln2080Ter | NS | | P | Het | 0/ >2500 | Waddling gait and difficulty in getting up from chair and difficulty in climbing stairs | Yes | - | - | - | XL | \*17041906 |
| 130 | S-6071 | DMDmd | *DMD* | Ex 43 | c.6283C>T | p.Arg2095Ter | NS | | P | Hemi | 0/ >2500 | Difficulty in ambulation and standing up | - | - | - | - | XL | \*20485447 |
| 131 | S-4445 | Muscular dystrophy | *DMD* | Ex 47 | c.6819\_6826del | p.Gly2274ArgfsTer14 | Indel | | P | Hemi | 0/ >2500 | Signs suggestive of DMD | Yes | - | - | - | XL | In this study |
| 132 | S-5878 | Muscular dystrophy | *DMD* | Ex 66 | c.9568C>T | p.Arg3190Ter | NS | | P | Hemi | 0/ >2500 | Progressive proximal muscle weakness and calf hypertrophy | - | - | - | - | XL | \*10909857 |
| 133 | S-1126 | Microcephaly, Spasticity | *DNAJC6* | Ex 6 | c.748C>T | p.Arg250Ter | NS | | LP | Hom | 0/ >2500 | Microcephaly and subtle dysmorphism, spasticity with bipyramidal and extrapyramidal involvement and neuroregression | - | - | - | - | AR | In this study |
| 134 | S-4952 | Epilepsy, Microcephaly | *DOK7* | Ex 7 | c.1121\_1124dupGCCT | p.Ala378SerfsTer30 | Indel | | P | Hom | 5θ/ >2500 | Seizures, dolicocephaly, microcephaly, hypotonia, macroglossia, clinodactyly, low set ears, high arched palate and failure to thrive | - | - | - | - | AR | \*18626973 |
| 135 | S-2781 | LGMDmd | *DYSF* | Ex 6 | c.634delC | p.Leu212CysfsTer15 | Indel | | LP | Het | 0/ >2500 | Difficulty in walking and standing and expressed elevated CPK levels | - | - | - | - | AR | In this study |
| Int 15 | c.1397+2dupT | NA | SS | | VUS | Het | 0/ >2500 | - | - | - | - | AR | ΔRCV000656701.1 |
| 136 | S-3604 | Muscular dystrophy | *DYSF* | Ex 15 | c.1357C>T | p.Pro453Ser | MS | | LP | Hom | 0/ >2500 | Progressive wasting of both upper limb and lower limb muscles, absent deep tendon reflexes and poor eye closure | Yes | - | - | Het (u) | AR | \*17070050 |
| 137 | S-5728 | Muscular dystrophy | *DYSF* | Ex 51 | c.5713C>T | p.Arg1905Ter | NS | | P | Hom | 1θ/ >2500 | Difficulty in climbing and jumping. Muscle biopsy was suggestive of muscular dystrophy | - | - | - | - | AR | \*16087766 |
| 138 | S-2179 | Leukodystrophy | *EIF2B2* | Int 3 | c.433+2T>G | NA | SS | | LP | Het | 0/ >2500 | MRI of brain was suggestive of cystic leukoencephalopathy features suggestive of megalencephalic leukoencephalopathy or RNAase T2 deficiency cystic leukoencephalopathy | - | - | - | - | AR | In this study |
| Second variant not detected | | | | | | | | | | | | | | | |
| 139 | R-1349 | Developmental delay | *ENAM* | Ex 9 | c.1259\_1260insAG | p.Pro422ValfsTer27 | Indel | | LP | Hom | 2θ/ >2500 | Neuroregression, frequent falls, slurred speech, cognitive decline, drooling and mild cerebral atrophy | - | - | - | - | AR | \*17125728 |
| 140 | S-1799 | Epilepsy | *EPM2A* | Ex 3 | c.690delG | p.Trp230Terfs | Indel | | LP | Hom | 0/ >2500 | Progressive myoclonic epilepsy | - | - | - | - | AR | In this study |
| 141 | R-0529 | Microcephaly | *EPM2A* | Ex 4 | c.929delT | p.Leu310Trpfs | Indel | | LP | Het | 0/ >2500 | Microcephaly, big and low set ears , seizures and delayed milestones | - | - | - | - | AR | In this study |
| Second variant not detected | | | | | | | | | | | | | | | |
| 142 | S-4506 | Cockayne syndromeo | *ERCC6* | Ex 5 | c.1012delA | p.Arg338GlyfsTer22 | Indel | P | | Hom | 0/ >2500 | Neuroregression, nystagmus, brachydactyly and failure to thrive, suggestive of Cockayne syndrome | Yes | - | Het | Hom (a) | AR | In this study |
| 143 | S-4707 | Spasticity, ID, Microcephaly | *ERCC6* | Ex 7 | c.1536dupG | p.Thr513AspfsTer4 | Indel | P | | Het | 0/ >2500 | Abnormal gait, spasticity, cognitive deficit, ID, microcephaly and short stature | - | - | - | - | AR | In this study |
| Int 20 | c.4063-1G>C | NA | SS | P | | Het | 0/ >2500 | - | - | - | - | AR | \*19894250 |
| 144 | S-3631 | Cerebral atrophyo | *ERCC8* | Int 2 | c.173+3\_173+6delAAGT | NA | SS | VUS | | Het | 0/ >2500 | Cerebral atrophy and pigmentation changes in the retinal fundus | Yes | Het | Ref | - | AR | In this study |
| Ex 10 | c.952delG | p.Val318PhefsTer10 | Indel | LP | | Het | 0/ >2500 | Yes | Ref | Het | - | AR | #rs766565870 |
| 145 | S-1614 | Encephalopathyo | *EXOSC3* | Ex 2 | c.395A>C | p.Asp132Ala | MS | P | | Hom | 0/ >2500 | Drooping neck, upper limb hypotonia, brisk reflex in lower limb, delay in walking, motor and language delay and recurrent respiratory tract infection. MRI showed white matter volume loss. | Yes | - | - | - | AR | \*23975261 |
| 146 | S-1080 | Muscular dystrophy | *FKRP* | Ex 4 | c.934C>A | p.Arg312Ser | MS | VUS | | Het | 0/ >2500 | Delayed motor development, abnormal gait, hypotonia, muscle weakness and wasting, calf hypertrophy and abnormal gait. | Yes | - | - | - | AR | In this study |
| Ex 4 | c.1343C>T | p.Pro448Leu | MS | P | | Het | 0/ >2500 | - | - | - | - | AR | \*27711214 |
| 147 | S-1961 | Muscular dystrophy | *FKRP* | Ex 4 | c.1343C>T | p.Pro448Leu | MS | P | | Hom | 0/ >2500 | Difficulty in walking, waddling gait with toe walking and diminished muscle power in upper and lower limbs | - | - | - | - | AR | \*27711214 |
| 148 | S-378 | Muscular dystrophy | *FKRP* | Ex 4 | c.1343C>T | p.Pro448Leu | MS | P | | Hom | 0/ >2500 | Delayed crawling, motor delay, frequent falls and elevated CPK | Yes | Het | Het | - | AR | \*27711214 |
| 149 | S-3420 | ALSo | *FUS* | Ex 14 | c.1520G>A | p.Gly507Asp | MS | LP | | Het | 0/ >2500 | Proximal and distal muscle weakness with wasting of right lower limbs, cramps and fasciculations | - | - | - | - | AD | \*23056579 |
| 150 | S-3714 | Developmental delay | *GAA* | Ex 4 | c.857C>T | p.Thr286Met | MS | VUS | | Het | 3 θ/>2500 | Floppiness, global developmental delay, brisk deep tendon reflex, head lag, plagiocephaly, bilateral subdural hydroma and hypoxic ischemic encephalopathy. | - | - | - | - | AR | #rs375310352 |
| Ex 14 | c.1927G>A | p.Gly643Arg | MS | P | | Het | 0/ >2500 | - | - | - | - | AR | \*9521422 |
| 151 | S-4990 | Krabbe diseasel | *GALC* | Ex 9 | c.956A>G | p.Tyr319Cys | MS | VUS | | Het | 76 θ/ >2500 | Milestone regression | - | - | - | - | AR | \*22115770 |
| Ex 11 | c.1230delC | p.Phe411LeufsTer46 | Indel | P | | Het | 0/ >2500 | Milestone regression | - | - | - | - | AR | In this study |
| 152 | S-410 | Dystonia | *GAMT* | Ex 5 | c.491dupG | p.Val165ArgfsTer26 | Indel | P | | Hom | 0/ >2500 | Dystonia and convulsions | - | - | - | - | AR | \*28438604 |
| 153 | S-710 | Neuropathy | *GDAP1* | Ex 2 | c.128dupT | p.Ile44AsnfsTer3 | Indel | LP | | Hom | 0/ >2500 | Features suggestive of neuropathies | - | - | - | - | AR | In this study |
| 154 | S-3459 | Charcot-Marie-Tooth diseasen | *GDAP1* | Ex 3 | c.358C>T | p.Arg120Trp | MS | P | | Het | 0/ >2500 | Delayed motor milestones, distal lower limb weakness, upper limb weakness, thenar and hypothenar wasting, distal limb wasting with arreflexia | - | - | - | - | AR | \*21753178 |
| Ex 6 | c.866T>C | p.Phe289Ser | MS | VUS | | Het | 0/ >2500 | - | - | - | - | AR | In this study |
| 155 | S-5873 | Neuropathy | *GDAP1* | Ex 6 | c.769C>T | p.Arg257Ter | NS | P | | Hom | 0/ >2500 | Distal weakness and features suggestive of hereditary motor sensory neuropathy | Yes | - | - | Het (u) | AR | \*21322820 |
| 156 | S-5083 | ASD | *GDAP1* | Ex 6 | c.802\_803delTG | p.Trp268GlyfsTer22 | Indel | LP | | Het | 0/ >2500 | Features suggestive of ASD | - | - | - | - | AD | #rs765346218 |
| 157 | S-3908 | Alexander diseasel | *GFAP* | Ex 1 | c.262C>T | p.Arg88Cys | MS | P | | Het | 0/ >2500 | Developmental delay and seizures | - | - | - | - | AD | \*17318298 |
| 158 | S-3380 | Neuropathy | *GJB1* | Ex 2 | c.43C>T | p.Arg15Trp | MS | P | | Hemi | 0/ >2500 | Progressive thinning and weakness of feet and hands, burning paresthesias in bilateral foot and palms, recurrent falls, hypotonia, absence of deep tendon reflexes, bilateral dorsiflexion and small hand muscle wasting | - | - | - | - | XL | \*11835375 |
| 159 | S-5966 | Leukodystrophy | *GJC2* | Ex 2 | c.138C>G | p.Ile46Met | MS | VUSD | | Het | 0/ >2500 | Developmental delay with regression, ataxic gait, dysarthria, nystagmus, hypotonia and leukodystrophy. | - | - | - | Not tested | AR | \*19423250 |
| Ex 2 | c.982delG | p.Ala328Profs | Indel | LP | | Het | 0/ >2500 | Yes | - | - | Het (u) | AR | In this study |
| 160 | S-2754 | Leukodystrophy | *GJC2* | Ex 2 | c.508delG | p.Glu170ArgfsTer40 | Indel | P | | Hom | 0/ >2500 | Spasticity, global developmental delay, regression of milestones, nystagmus and leukodystrophy | - | - | - | - | AR | In this study |
| 161 | S-4159 | Dystonia | *GLB1* | Ex 2 | c.145C>T | p.Arg49Cys | MS | P | | Het | 0/ >2500 | Dystonia and developmental delay | - | - | - | - | AR | \*25936995 |
| Ex 13 | c.1325G>A | p.Arg442Gln | MS | LP | | Het | 5 θ/ >2500 | - | - | - | - | AR | \*15365997 |
| 162 | S-5514 | Epilepsy, ASD | *GLDC* | Ex 15 | c.1786C>T | p.Arg596Ter | NS | P | | Het | 0/ >2500 | Global developmental delay, autistic traits and multiple episodes of seizures | - | - | - | - | AR | \*17361008 |
| 163 | S-4666 | Microcephaly | *GLRA1* | Ex 7 | c.839G>A | p.Arg280His | MS | LP | | Hom | 0/ >2500 | Global developmental delay and microcephaly | - | - | - | - | AR | \*10514101 |
| 164 | S-4657 | Spasticity | *GM2A* | Int 2 | c.244-2A>G | NA | SS | LP | | Hom | 0/ >2500 | Developmental arrest and regression, exaggerated startle response, hypotonia, spasticity, and cherry red spot | - | - | - | - | AR | In this study |
| 165 | S-4296 | Muscular dystrophy, Myopathy | *GNE* | Ex 3 | c.478C>T | p.Arg160Ter | NS | P | | Het | 0/ >2500 | Proximal muscular dystrophy and difficulty in walking | - | - | - | - | AR | \*24796702 |
| Ex 12 | c.2179G>A | p.Val727Met | MS | VUSD | | Het | 94θ/ >2500 | - | - | - | - | AR | \*21708040 |
| 166 | S-4544 | Myopathy | *GNE* | Ex 3 | c.478C>T | p.Arg160Ter | NS | P | | Het | 0/ >2500 | Proximal muscular dystrophy and difficulty in walking | - | - | - | - | AR | \*21708040 |
| Ex 12 | c.2179G>A | p.Val727Met | MS | VUSD | | Het | 94θ/ >2500 | - | - | - | - | AR | \*24005727 |
| 167 | S-439 | Myopathy | *GNE* | Ex 3 | c.478C>T | p.Arg160Ter | NS | P | | Het | 0/ >2500 | Bilateral lower limb weakness, bilateral scapular winging, facial muscle weakness and bilateral calf pseudohypertrophy | - | - | - | - | AR | \*25182749 |
| Ex 12 | c.2179G>A | p.Val727Met | MS | VUSD | | Het | 94θ/ >2500 | - | - | - | - | AR | \*24005727 |
| 168 | S-1191 | Myopathy | *GNE* | Ex 10 | c.1760T>C | p.Ile587Thr | MS | LP | | Hom | 0/ >2500 | Progressive weakness, writing problem, difficulty in breathing while walking and talking, sleep difficulties, weakness around finger tips, clawing of toes, pes cavus and thinning of forearms | - | - | - | - | AR | \*12497639 |
| 169 | S-5444 | Macrocephalyo | *GNPTG* | Ex 4 | c.189\_192dupTCTC | p.Phe65SerfsTer19 | Indel | LP | | Hom | 0/ >2500 | Gait imbalance. Brain MRI showed Arnold-Chiari malformation with skeletal features of mucopolysaccharidoses | - | - | - | - | AR | In this study |
| 170 | S-760 | Epilepsy | *GRIN2A* | Ex 14 | c.2764delG | p.Ala922LeufsTer20 | Indel | LP | | Het | 0/ >2500 | Global developmental delay, seizures, shock sepsis, metabolic aciduria, hypoglycemia, anemia, thrombocytopenia, nephrocalcinosis and hypervitaminosis | - | - | - | - | AD | In this study |
| 171 | S-5420 | Ataxia, Microcephaly | *GRM1* | Ex 2 | c.748delC | p.Gln250ArgfsTer45 | Indel | LP | | Hom | 0/ >2500 | Global developmental delay, microcephaly, quadriparesis, ataxia and strabismus | - | - | - | - | AR | In this study |
| 172 | S-5250 | Epilepsy | *HEXA* | Ex 8 | c.964G>T | p.Asp322Tyr | MS | P | | Hom | 0/ >2500 | Febrile encephalopathy, neuroregression, seizures, cherry red spot, hepatomegaly, macrocephaly and coarse facies | - | - | - | - | AR | \*23852624 |
| 173 | S-5286 | Epilepsy | *HEXA* | Ex 13 | c.1444G>A | p.Glu482Lys | MS | LP | | Hom | 0/ >2500 | Febrile seizures, delayed motor development, failure to thrive, milestone regression. Biochemical test indicated elevated ammonia levels | - | - | - | - | AR | \*16088929 |
| 174 | S-37 | Developmental delay | *HGSNAT* | Ex 11 | c.1019G>A | p.Trp340Ter | NS | LP | | Hom | 0/ >2500 | Global developmental delay, hypertonia, cerebral atrophy, encephalopthy, coarse facies, behavioural abnormalities, deaf mutism and stereotypic movements | - | - | - | - | AR | In this study |
| 175 | S-1643 | Epilepsy | *HPD* | Ex 7 | c.398T>C | p.Phe133Ser | MS | VUS | | Het | 14 θ/ >2500 | Epileptic encephalopathy and seizures since 2nd day of birth, did not attain milestones | - | - | - | - | AR | #rs545913399 |
| Ex 13 | c.1005C>G | p.Ile335Met | MS | LP | | Het | 42 θ/ >2500 | - | - | - | - | AR | \*10942115 |
| 176 | S-2482 | Dystonia | *HPRT1* | Int 7 | c.532+5G>A | NA | SS | LP | | Hemi | 0/ >2500 | Features suggestive of dystonia | - | - | - | - | XL | \*2347587 |
| 177 | S-5906 | Neuropathy | *IGHMBP2* | Ex 7 | c.958C>T | p.Arg320Ter | NS | P | | Hom | 0/ >2500 | Features suggestive of Charcot-Marie-Tooth disease and spinal muscular atrophy | - | - | - | - | AR | \*14506069 |
| 178 | R-0606 | Neuropathy | *IGHMBP2* | Ex 13 | c.2430delT | p.Thr811ProfsTer20 | Indel | LP | | Hom | 0/ >2500 | Developmental delay and an electromyography/nerve conduction velocity test was suggestive of hereditary motor and sensory neuropathy. She also underwent a corrective surgery for congenital talipes equino varus | Yes | Het | Het | - | AR | In this study |
| 179 | S-5963 | ASD | *IQSEC2* | Ex 8 | c.2682delC | p.Asp894GlufsTer13 | Indel | LP | | Het | 0/ >2500 | Developmental delay, speech regression, seizures, sterotypic hand movements and poor eye contact | - | - | - | - | XL | In this study |
| 180 | S-1059 | Microcephaly, ASD | *IQSEC2* | Int 9-Ex10 | c.2890-2\_2898del | NA | SS | LP | | Hemi | 0/ >2500 | Hand flapping movements, microcephaly, tongue protrusion and autistic features | Yes | - | Ref | - | XL | In this study |
| 181 | S-4543 | Neuropathy | *KARS* | Ex 3 | c.223C>T | p.Gln75Ter | NS | LP | | Het | 0/ >2500 | Axonal neuropathy | - | - | - | - | AR | In this study |
| Second variant not detected | | | | | | | | | | | | | | | |
| 182 | S-898 | Epilepsy | *KCNJ10* | Ex 2 | c.168G>A | p.Trp56Ter | NS | | LP | Het | 0/ >2500 | Seizures, global developmental delay, sparse scalp hair, no social smile and eye contact. Electroencephalogram revealed hypsarrhythmia, suggesting epileptic encephalopathy | - | - | - | - | AR | In this study |
| Second variant not detected | | | | | | | | | | | | | | | |
| 183 | S-3640 | Epilepsy | *KCNQ2* | Ex4 | c.629G>C | p.Arg210Pro | MS | | LP | Het | 0/ >2500 | Neonatal onset epilepsy, features suggestive of pyridoxal phosphate dependent seizures | Yes | - | - | - | AD | ΔRCV000454413.1 |
| 184 | S-566 | Epilepsy | *KCNQ2* | Ex 7 | c.997C>T | p.Arg333Trp | MS | | P | Het | 0/ >2500 | Seizures and focal heterotopia | - | - | - | - | AD | \*23621294 |
| 185 | S-4083 | Epilepsy | *KCNQ2* | Ex 8 | c.1041C>G | p.Tyr347Ter | NS | | LP | Het | 0/ >2500 | Seizures | - | - | - | - | AD | In this study |
| 186 | S-1375 | ASD | *KIAA2022* | Ex 3 | c.2749G>T | p.Gly917Ter | NS | | LP | Het | 0/ >2500 | Features suggestive of Rett syndrome | - | - | - | - | XL | In this study |
| 187 | S-3449 | Epilepsy, ID | *KIF1A* | Ex 3 | c.296C>T | p.Thr99Met | MS | | P | Het | 1 θ/ >2500 | Regression since early infancy, ID, impaired vision, myoclonic epilepsy, spastic quadriparesis and cerebellar atrophy | Yes | Ref | Ref | - | AD | \*26125038 |
| 188 | S-1143 | Kabuki syndromeo | *KMT2D* | Ex 3 | c.303dupG | p.Ser102GlufsTer6 | Indel | | P | Het | 0/ >2500 | Global developmental delay, failure to gain weight, long face, flared pinna, shallow orbits, nystagmus, cataract and cardiac abnormalities | - | - | - | - | AD | ΔRCV000522326.1 |
| 189 | S-2052 | Kabuki syndromeo | *KMT2D* | Ex 31 | c.7456delC | p.His2486ThrfsTer57 | Indel | | LP | Het | 0/ >2500 | Long palpebral fissures, low set ears, flat nose tip and small little finger | Yes | - | - | - | AD | In this study |
| 190 | S-4848 | Kabuki syndromeo | *KMT2D* | Int 46 | c.14516-2A>C | NA | SS | | LP | Het | 0/ >2500 | Features suggestive of Kabuki syndrome | - | - | - | - | AD | In this study |
| 191 | S-80 | Hydrocephaluso | *L1CAM* | Ex 6 | c.604G>T | p.Asp202Tyr | MS | | P | Hemi | 0/ >2500 | Hydrocephalus, aqueductal stenosis and adducted thumb | Yes | - | - | - | XL | \*26891472 |
| 192 | S-4873 | Hydrocephaluso | *L1CAM* | Ex 18 | c.2278C>T | p.Arg760Ter | NS | | P | Hemi | 0/ >2500 | Plagiocephaly, hypertelorism, depressed nasal bridge, midfacial hypoplasia and adducted thumb | Yes | Ref | Het | - | XL | \*19846429 |
| 193 | S-5306 | Leukodystrophy | *L2HGDH* | Ex 7 | c.829C>T | p.Arg277Ter | NS | | P | Hom | 1 θ/ >2500 | Global developmental delay, seizures, spastic gait, brisk distal tendon reflex in the lower limbs and bilateral ankle clonus | - | - | - | - | AR | \*20052767 |
| 194 | S-4928 | Myopathy | *LAMA2* | Ex 2 | c.176G>A | p.Gly59Glu | MS | | VUS | Het | 0/ >2500 | Developmental delay, partially achieved milestones and global hypotonia, EMG findings were indicative of myopathy | - | - | - | - | AR | #rs748303070 |
| Ex 3 | c.286\_287delAG | p.Arg96ThrfsTer8 | Indel | | LP | Het | 0/ >2500 | - | - | - | - | AR | In this study |
| 195 | S-4167 | Muscular dystrophy | *LAMA2* | Ex 2 | c.250C>T | p.Arg84Ter | NS | | P | Het | 0/ >2500 | Features suggestive of MD-CMD | Yes | - | - | - | AR | #rs868019991 |
| Ex 37 | c.5360G>A | p.Trp1787Ter | NS | | P | Het | 0/ >2500 | Yes | - | - | - | AR | In this study |
| 196 | S-5626 | Muscular dystrophy | *LAMA2* | Int 6 | c.910-2A>G | NA | SS | | LP | Het | 0/ >2500 | Motor developmental delay, decreased muscle tone in upper limbs and had a history of difficulty in feeding and apnoea | - | - | - | - | AR | In this study |
| Ex 22 | c.3174G>A | p.Lys1058= | SS | | VUS | Het | 0/ >2500 | - | - | - | - | AR | In this study |
| 197 | S-3611 | Muscular dystrophy | *LAMA2* | Int 7 | c.1307-1G>C | NA | SS | | P | Hom | 0/ >2500 | Merosin negative congenital muscular dystrophy and delayed motor milestones. Electromyography suggests mild myopathic pattern | - | - | - | - | AR | In this study |
| 198 | S-3227 | Muscular dystrophy | *LAMA2* | Ex 12 | c.1749C>G | p.Tyr583Ter | NS | | LP | Hom | 0/ >2500 | Features suggestive of MD-CMD | - | - | - | - | AR | In this study |
| 199 | S-2689 | Muscular dystrophy | *LAMA2* | Ex 19 | c.2538delG | p.Arg846SerfsTer42 | Indel | | LP | Hom | 0/ >2500 | Significant motor delay, good higher mental functions and cognition | - | - | - | - | AR | In this study |
| 200 | S-246 | Developmental delay | *LAMA2* | Ex 19 | c.2749+4\_2749+15del | NA | SS | | LP | Het | 0/ >2500 | Developmental delay, inability to walk or sit without support, pigmented sparse hair, hypotonia and pyloric stenosis | - | - | - | - | AR | In this study |
| Second variant not detected | | | | | | | | | | | | | | | |
| 201 | R-0569 | Muscular dystrophy | *LAMA2* | Ex 21 | c.2961C>G | p.Cys987Trp | MS | | VUS | Het | 0/ >2500 | Developmental delay and paucity of movements of all four limbs | Yes | Ref | Het | - | AR | In this study |
| Ex 30 | c.4348C>T | p.Arg1450Ter | NS | | LP | Het | 1θ/ >2500 | Yes | Het | Ref | - | AR | #rs200923373 |
| 202 | S-2749 | Muscular dystrophy | *LAMA2* | Ex 27 | c.4048C>T | p.Arg1350Ter | NS | | P | Hom | 0/ >2500 | Global developmental delay, motor delay and features suggestive of congenital muscular dystrophy | - | - | - | - | AR | \*23326386 |
| 203 | S-2392 | Muscular dystrophy, Leukodystrophy | *LAMA2* | Ex 29 | c.4198C>T | p.Arg1400Ter | NS | | P | Hom | 1θ/ >2500 | Delayed motor development, hypotonia, muscle wasting, macrocephaly, joint contractures and myopathic facies. MRI suggestive of leukodystrophy | - | - | - | - | AR | \*24225367 |
| 204 | S-5563 | Muscular dystrophy | *LAMA2* | Ex 50 | c.7147C>T | p.Arg2383Ter | NS | | P | Het | 0/ >2500 | Gross motor delay and muscular dystrophy | Yes | Ref | Het | - | AR | \*11071490 |
| Ex 62 | c.8767G>T | p.Glu2923Ter | NS | | P | Het | 0/ >2500 | Yes | Het | Ref | - | AR | In this study |
| 205 | S-369 | MD-CMDmd | *LAMA2* | Ex 56 | c.7816delA | p.Met2606Ter | NS | | P | Het | 0/ >2500 | Gross motor delay and increased CPK. Brain MRI and muscle biopsy were suggestive of MD-CMD | - | - | - | - | AR | In this study |
| Int 61 | c.8703+1G>A | NA | SS | | P | Het | 0/ >2500 | - | - | - | - | AR | \*27708273 |
| 206 | S-1525 | Muscular dystrophy | *LAMA2* | Ex 57 | c.8072\_8073delCT | p.Ser2691CysfsTer15 | Indel | | P | Hom | 0/ >2500 | Features suggestive of muscular dystrophy | Yes | Het | Het | - | AR | In this study |
| 207 | S-6242 | LGMDmd | *LMNA* | Ex 6 | c.1157G>A | p.Arg386Lys | MS | | P | Het | 0/ >2500 | Waddling gait, muscle weakness, joint contractures of hamstrings, elbow and tendon achilles. Biochemical analysis showed elevated levels of CPK and muscle biopsy was indicative of muscular dystrophy | - | - | - | - | AD | \*20848652 |
| 208 | S-1726 | Rett syndromeasd,m | *MECP2* | Ex 3 | c.316C>T | p.Arg106Trp | MS | | P | Het | 0/ >2500 | Microcephaly, bilateral flat feet, short neck, global developmental delay, repetitive hand clapping, body rocking, stereotypic movements and autistic features. MRI is normal. EEG suggested epileptic activity in central and parietal regions | - | - | - | - | AD | \*16965328 |
| 209 | S-1358 | Rett syndromeasd | *MECP2* | Ex 3 | c.316C>T | p.Arg106Trp | MS | | P | Het | 0/ >2500 | Features suggestive of Rett syndrome. | - | - | - | - | XL | \*10767337 |
| 210 | S-346 | Rett syndromeasd | *MECP2* | Ex 4 | c.397C>T | p.Arg133Cys | MS | | P | Het | 0/ >2500 | Club foot, regression of milestones, autistic features, atonic seizures followed by generalized seizures, bowel and bladder incontinence, and mouthing of hands | - | - | - | - | XL | \*18332345 |
| 211 | S-4174 | Rett syndromeasd | *MECP2* | Ex 4 | c.455C>G | p.Pro152Arg | MS | | P | Het | 0/ >2500 | Global developmental delay, microcephaly, hypotonia, and hand stereotypes | - | - | - | - | XL | \*12966523 |
| 212 | S-4299 | Ataxia, Microcephaly, ASD | *MECP2* | Ex 4 | c.455C>G | p.Pro152Arg | MS | | P | Het | 0/ >2500 | Microcephaly, limb length discrepancy, ataxia, spasticity, flappy hands, delayed walking, cognitive delay, delayed and bisyllabic speech, laughter spells, inward strabismus, no eye contact, hyperactivity, repetitive hand clapping and absence seizures. EEG showed abnormal multifocal epileptic activity | - | - | - | - | XL | \*12966523 |
| 213 | S-2350 | ASD | *MECP2* | Ex 4 | c.455C>G | p.Pro152Arg | MS | | P | Het | 0/ >2500 | Features suggestive of ASD | - | - | - | - | XL | \*18989701 |
| 214 | S-1838 | Rett syndromeasd | *MECP2* | Ex 4 | c.473C>T | p.Thr158Met | MS | | P | Het | 0/ >2500 | Neurocognitive regression, loss of speech and communication, loss of useful hand movements, stereotypic behaviour and hand movements, and bruxism | - | - | - | - | XL | \*12567420 |
| 215 | S-3104 | Rett syndromeasd | *MECP2* | Ex 4 | c.473C>T | p.Thr158Met | MS | | P | Het | 0/ >2500 | Global developmental delay with microcephaly | - | - | - | - | XL | \*12567420 |
| 216 | S-4200 | ASD | *MECP2* | Ex 4 | c.502C>T | p.Arg168Ter | NS | | P | Het | 0/ >2500 | Global developmental delay, regression at 18 to 24 months, autistic features and hand wringing | - | - | - | - | XL | \*24283265 |
| 217 | S-2738 | Rett syndromeasd,m | *MECP2* | Ex 4 | c.611dupC | p.Glu205ArgfsTer31 | Indel | | P | Het | 0/ >2500 | Developmental delay, poor eye contact, no speech, right sided paresis, history of right sided focal seizures, microcephaly and autistic behaviour | Yes | - | Ref | - | AD | In this study |
| 218 | S-2801 | Epilepsy, Microcephaly, ASD | *MECP2* | Ex 4 | c.645\_646delAA | p.Pro217TrpfsTer18 | Indel | | LP | Het | 0/ >2500 | Autistic regression, epilepsy and microcephaly | - | - | - | - | XL | In this study |
| 219 | S-886 | Rett syndromeasd | *MECP2* | Ex 4 | c.695delG | p.Gly232AlafsTer16 | Indel | | P | Het | 0/ >2500 | Global developmental delay, spasticity, seizures, microcephaly and hypertonia | - | - | - | - | XL | \*15737703 |
| 220 | S-3305 | Rett syndromeasd | *MECP2* | Ex 4 | c.763C>T | p.Arg255Ter | NS | | P | Het | 0/ >2500 | Global developmental delay and autistic features. | - | - | - | - | XL | \*28347601 |
| 221 | S-3408 | Rett syndromeasd,m | *MECP2* | Ex 4 | c.763C>T | p.Arg255Ter | NS | | P | Het | 0/ >2500 | Autistic features and microcephaly | Yes | - | Ref | - | XL | \*28347601 |
| 222 | S-5585 | Epilepsy | *MECP2* | Ex 4 | c.808C>T | p.Arg270Ter | NS | | P | Het | 0/ >2500 | Global developmental delay, regression, epilepsy and dystonic movements | - | - | - | - | XL | \*10814718 |
| 223 | S-5361 | Rett syndromeasd | *MECP2* | Ex 4 | c.917G>A | p.Arg306His | MS | | P | Het | 0/ >2500 | Developmental delay, no eye contact, hand flapping and stereotypes | - | - | - | - | XL | \*15057977 |
| 224 | S-2951 | Rett syndromeasd,m | *MECP2* | Ex 4 | c.1450\_1453delAGAG | p.Arg484Leufs | Indel | | P | Het | 0/ >2500 | Seizures, microcephaly, autistic features and regression of language milestones | - | - | - | - | XL | \*15737703 |
| 225 | S-4628 | Epilepsy | *MFN2* | Ex 11 | c.1090C>T | p.Arg364Trp | MS | | P | Het | 0/ >2500 | Infantile spasms, global developmental delay and focal myoclonic jerk. MRI showed cerebellar polymicrogyria | Yes | Ref | Ref | - | AD | \*28063088 |
| 226 | S-1912 | NCLe | *MFSD8* | Ex 5 | c.217dupA | p.Thr73AsnfsTer12 | Indel | | P | Hom | 0/ >2500 | Seizures, shivering, walking difficulty, loss of vision and progressive neuroregression | Yes | Het | Het | - | AR | ΔRCV000188195.1 |
| 227 | S-4504 | NCLe | *MFSD8* | Ex 7 | c.590dupG | p.Val198CysfsTer7 | Indel | | LP | Hom | 0/ >2500 | Progressive myoclonic epilepsy, unsteadiness while walking, regression of milestones and speech difficulty | - | - | - | - | AR | In this study |
| 228 | S-2284 | NCLa | *MFSD8* | Ex 7 | c.694delC | p.Leu232Terfs | Indel | | LP | Hom | 0/ >2500 | Developmental delay, myoclonic jerks, frequent falls and ataxia. MRI showed cerebellar atrophy | - | - | - | - | AR | In this study |
| 229 | S-6 | Leukodystrophy | *MFSD8* | Int 7 | c.699-1G>A | NA | SS | | LP | Hom | 0/ >2500 | Neuroregression, loss of balance while walking, drooling and delayed speech. MRI showed cerebellar atrophy. | Yes | - | - | Hom (a) | AR | In this study |
| 230 | S-1925 | NCLe | *MFSD8* | Ex 12 | c.1338delA | p.Pro447GlnfsTer11 | Indel | | LP | Hom | 0/ >2500 | Neuroregression with gait abnormality. MRI was suggestive of NCL | - | - | - | - | AR | In this study |
| 231 | S-5680 | Leukodystrophy | *MLC1* | Ex 2 | c.135dupC | p.Cys46LeufsTer34 | Indel | | P | Hom | 2θ/ >2500 | Macrocephaly and brain MRI showed dysmyelination and liquefaction of areas of the temporal lobe | - | - | - | - | AR | \*12189496 |
| 232 | S-2324 | Leukodystrophy | *MLC1* | Ex11 | c.959C>A | p.Thr320Lys | MS | | LP | Hom | 0/ >2500 | Macrocephaly, neuroregression and spasticity | - | - | - | - | AR | \*21555057 |
| 233 | S-5406 | Spasticity | *MMACHC* | Ex 3 | c.394C>T | p.Arg132Ter | NS | | P | Hom | 2θ/ >2500 | Global developmental delay, seizures, spasticity, tremors, pancytopenia, optic atrophy, alopecia, hyperpigmentation and recurrent respiratory tract infection. MRI showed white matter hyperintensities and mild prominence of bilateral subarachnoid spaces | Yes | Het | Het | - | AR | \*20924684 |
| 234 | S-6030 | Muscular dystrophy | *MTHFR* | Ex 8 | c.1262G>C | p.Trp421Ser | MS | | P | Hom | 0/ >2500 | Broad facies, nose abnormality, downslanting palpebral fissure, breathing and swallowing difficulties, large ears and failure to thrive. | - | - | - | - | AR | \*26025547 |
| 235 | S-3769 | Myopathy | *MTM1* | Ex 13 | c.1420C>T | p.Arg474Ter | NS | | P | Hemi | 0/ >2500 | Generalised hypotonia, frog leg position and pooling of secretions | Yes | - | Het | - | XL | \*11793470 |
| 236 | S-5539 | Neuropathy | *MTMR2* | Ex 14 | c.1734dupA | p.Tyr579IlefsTer25 | Indel | | LP | Hom | 0/ >2500 | Features suggestive of neuropathy | - | - | - | - | AR | In this study |
| 237 | S-329 | Ataxia | *MTTP* | Ex 10 | c.1092delC | p.Ser365LeufsTer8 | Indel | | LP | Hom | 0/ >2500 | Retinitis pigmentosa, progressive cerebellar and sensory ataxia, tremors and malabsorption with recurrent diarrhea. Peripheral smear showed acanthocytes. | - | - | - | - | AR | In this study |
| 238 | S-4525 | Myasthenic syndromeo | *MUSK* | Ex 3 | c.308A>G | p.Asn103Ser | MS | | LP | Hom | 0/ >2500 | Bilateral lower limb weakness since thirteen years of age, upper limb weakness since eighteen years of age, slow extra-ocular movement and bilateral restricted abduction | - | - | - | - | AR | \*25900532 |
| 239 | S-4875 | Myopathy | *MYH2* | Ex 21 | c.2377C>T | p.Arg793Ter | NS | | LP | Hom | 0/ >2500 | Alternating esotropia, myopathy and congenital myathenia | - | - | - | - | AR | #rs545623839 |
| 240 | S-5165 | Myopathy | *MYH7* | Ex 34 | c.4850\_4852delAGA | p.Lys1617del | Indel | | P | Het | 0/ >2500 | Distal weakness of dorsi flexors and finger extensors, and severe neck flexor weakness | - | - | - | - | AD | \*27081534 |
| 241 | S-3632 | Muscular dystrophy | *MYH7* | Ex 39 | c.5746C>T | p.Gln1916Ter | NS | | LP | Het | 0/ >2500 | Proximal muscle weakness. Muscle biopsy is negative. | - | - | - | - | AD | In this study |
| 242 | S-832 | Leukodystrophy | *NDUFV2* | Ex 1 | c.1A>G | p.Met1? | MS | | LP | Het | 2θ/ >2500 | Excess cry, sudden onset of irritability, increased tone in limbs, fever followed by left hemiparesis and lost all milestones. MRI revealed leukodystrophy. | Yes | Het | Ref | - | AR | In this study |
| Ex 6 | c.547G>A | p.Ala183Thr | MS | | VUS | Het | 0/ >2500 | Yes | Ref | Het | - | AR | #rs1428682980 |
| 243 | S-1310 | Ataxia, Leukodystrophy | *NDUFV2* | Ex 5 | c.427C>T | p.Arg143Ter | NS | | LP | Het | 0/ >2500 | Episodic vomiting, headache, irritability, global developmental delay and ataxia. | - | - | - | - | AR | #rs768703151 |
| Ex 8 | c.694A>C | p.Thr232Pro | MS | | VUS | Het | 0/ >2500 | - | - | - | - | AR | #rs534384070 |
| 244 | S-2616 | Myopathy | *NEB* | Int 16 | c.1470+2T>C | NA | SS | | LP | Het | 0/ >2500 | Persistent difficulty in climbing stairs, inability to get up from sitting position, frequent falls, and weakness while taking objects from heights. Power grading of limbs showed mild weakness in trunk and proximal muscles. | - | - | - | - | AR | In this study |
| Ex 119 | c.18676C>T | p.Gln6226Ter | NS | | LP | Het | 0/ >2500 | - | - | - | - | AR | \*16917880 |
| 245 | S-2135 | Myopathy | *NEB* | Ex 179 | c.25137delT | p.Asn8379LysfsTer38 | Indel | | LP | Het | 0/ >2500 | Congenital talipes equinovarus, patent ductus arteriosus, myopathic facies, high arched palate and undescended testis. | - | - | - | - | AR | In this study |
| Second variant not detected | | | | | | | | | | | | | | | |
| 246 | S-567 | Neuropathy | *NEFL* | Ex 3 | c.1261C>T | p.Arg421Ter | NS | | P | Hom | 0/ >2500 | Motor developmental delay and hypotonia. A nerve conduction velocity test was suggestive of HMSN. | - | - | - | - | AR | \*25264603 |
| 247 | S-5215 | Epilepsy | *NHLRC1* | Ex 1 | c.468\_469delAG | p.Gly158ArgfsTer17 | Indel | | P | Het | 0/ >2500 | Features suggestive of epilepsy | - | - | - | - | AR | \*12958597 |
| Ex 1 | c.793C>T | p.Arg265Ter | NS | | P | Het | 0/ >2500 | - | - | - | - | AR | \*15781812 |
| 248 | S-4709 | CdLSo | *NIPBL* | Ex 10 | c.2689C>T | p.Gln897Ter | NS | | P | Het | 0/ >2500 | Microcephaly, synophrys, low anterior hair line, long philtrum, anteverted nares, thin lip, depressed nose, high arched palate, short neck and micromelia | - | - | - | - | AD | In this study |
| 249 | S-4473 | CdLSo | *NIPBL* | Ex 27 | c.5272C>T | p.Arg1758Ter | NS | | P | Het | 0/ >2500 | Global developmental delay, microcephaly and dysmorphism | - | - | - | - | AD | \*15318302 |
| 250 | S-3171 | Ataxia | *NKX2-1* | Ex 3 | c.650C>A | p.Ser217Ter | NS | | P | Het | 0/ >2500 | Complex movement disorder involving motor delay, hypotonia, ataxia, myoclonus and choreoathetosis | - | - | - | - | AD | \*18661567 |
| 251 | S-5530 | Leukodystrophy | *NOTCH3* | Ex 4 | c.544C>T | p.Arg182Cys | MS | | P | Het | 0/ >2500 | Recurrent stroke, decreased neurocognition and periventricular lacunae | - | - | - | - | AD | \*26715087 |
| 252 | S-2000 | Ataxia | *NPC1* | Ex 13 | c.2068A>T | p.Ile690Phe | MS | | VUS | Het | 0/ >2500 | Difficulty in walking, slow response, ataxia, poor scholastic performance, extensor plantar reflex along with increased tone and reflex | - | - | - | - | AR | In this study |
| Int 17 | c.2604+1G>A | NA | SS | | LP | Het | 0/ >2500 | - | - | - | - | AR | ΔRCV000591979.1 |
| 253 | S-453 | Sotos syndromeo | *NSD1* | Ex 5 | c.1318C>T | p.Arg440Ter | NS | | P | Het | 0/ >2500 | Macrocephaly, broad frontal region, deep set nails, downslanting eyes, micrognathia and advanced bone age | - | - | - | - | AD | \*15942875 |
| 254 | S-5998 | Sotos syndromeo | *NSD1* | Ex 5 | c.1855delG | p.Val619Terfs | Indel | | P | Het | 0/ >2500 | Overgrowth, broad forehead, downslanting eyes, thinning of corpus callosum and large hands and feet | - | - | - | - | AD | In this study |
| 245 | S-342 | Sotos syndromeo | *NSD1* | Int 19 | c.6010-2A>C | NA | SS | | P | Het | 0/ >2500 | Generalized seizures, global developmental delay, loss of white matter, congnitive deficit, delayed speech, ID, anemia, tall stature, facial dysmorphism | - | - | - | - | AD | In this study |
| 246 | S-695 | Sotos syndromeo | *NSD1* | Ex 20 | c.6049C>T | p.Arg2017Trp | MS | | P | Het | 0/ >2500 | Overgrowth, scoliosis and pointed chin | - | - | - | - | AD | \*15942875 |
| 247 | R-0604 | ID | *NSUN2* | Ex 5 | c.560delC | p.Pro187LeufsTer8 | Indel | | LP | Hom | 0/ >2500 | ID and dysmorphism | - | - | - | - | AR | In this study |
| 248 | S-5459 | Neuropathy | *NTRK1* | Ex 4 | c.404\_405delTG | p.Val135AlafsTer37 | Indel | | P | Het | 0/ >2500 | Recurrent fracture of long bones, decreased or absent sweating and insensitivity to pain | - | - | - | - | AR | In this study |
| Ex 13 | c.1787G>A | p.Arg596Gln | MS | | P | Het | 1θ/ >2500 | - | - | - | - | AR | \*19651702 |
| 249 | S-6052 | Neuropathy | *NTRK1* | Ex 15 | c.2115G>A | p.Trp705Ter | NS | | LP | Hom | 0/ >2500 | Developmental delay, self mutilation, early exfoliation of teeth, dislocation of bilateral hip joint, temperature instability and decreased sweating | - | - | - | - | AR | In this study |
| 250 | S-3192 | Epilepsy, Neuropathy | *OCRL* | Ex 10 | c.827T>C | p.Phe276Ser | MS | | LP | Hemi | 0/ >2500 | Global developmental delay, congenital cataract, hypotonia, umbilical hernia, failure to thrive, vitamin D deficiency and seizures | Yes | - | Het | - | XL | \*16420990 |
| 251 | S-5835 | Epilepsy, Microcephaly | *OSTM1* | Ex 3 | c.518\_521delATTG | p.Asn173IlefsTer2 | Indel | | LP | Hom | 0/ >2500 | Encephalopathy, seizures, spasticity, delayed motor development, global developmental delay, failure to thrive, microcephaly, septicemia and lactic acidemia | Yes | Het | Het | - | AR | In this study |
| 252 | S-5508 | Dystonia | *PANK2* | Ex 2 | c.828\_829delTG | p.Cys276TrpfsTer15 | Indel | | P | Hom | 0/ >2500 | Global developmental delay, dystonia, tremor, chorea and focal seizures | - | - | - | - | AR | \*28567303 |
| 253 | S-5519 | Dystonia | *PANK2* | Ex 2 | c.856C>T | p.Arg286Cys | MS | | P | Het | 0/ >2500 | Progressive dystonia with oromotor dyskinesia | - | - | - | - | AR | \*22221393 |
| Ex 4 | c.1339G>T | p.Asp447Tyr | MS | | VUS | Het | 0/ >2500 | - | - | - | - | AR | In this study |
| 254 | S-4691 | Epilepsy, Microcephaly, ID | *PCDH19* | Ex 1 | c.94G>T | p.Glu32Ter | NS | | LP | Het | 0/ >2500 | Cerebellar atrophy, generalised seizures, global developmental delay, microcephaly, tremors, cognitive deficit, delayed speech and ID. | - | - | - | - | XL | In this study |
| 255 | S-3346 | Epilepsy | *PCDH19* | Ex 1 | c.339\_355delinsTCGT | p.Val114ArgfsTer5 | Indel | | LP | Het | 0/ >2500 | Multiple episodes of generalized tonic-clonic seizures. | - | - | - | - | XL | In this study |
| 256 | S-4839 | Microcephaly, ID | *PCNT* | Ex 28 | c.5767C>T | p.Arg1923Ter | NS | | P | Het | 0/ >2500 | Delayed motor development, obesity, global developmental delay, mild ID, microcephaly, hypertelorism, bilateral epicanthic fold, hirsutism, thick ear lobes, depressed nasal bridge and short neck with elevated total cholesterol, LDL and TSH levels | - | - | - | - | AR | \*18174396 |
| Second variant not detected | | | | | | | | | | | | | | | |
| 257 | S-4592 | Ataxia | *PDHA1* | Ex 5 | c.483C>T | p.Tyr161Tyr | MS | | LP | Het | 0/ >2500 | Cerebellar ataxia, repetitive eye movements, developmental delay, trouble eating and performing fine motor movements | - | - | - | - | XL | \*18023225 |
| 258 | S-340 | Muscle weaknesso | *PHYH* | Int 6 | c.679-2A>C | NA | SS | | LP | Het | 0/ >2500 | Frequent falls, abnormal gait, muscle weakness, attention deficit, round face, short neck and ichthyosis | Yes | - | - | - | AR | In this study |
| Second variant not detected | | | | | | | | | | | | | | | |
| 259 | S-1781 | Dementiao | *PKD2* | Ex 4 | c.1003C>T | p.Gln335Ter | NS | | LP | Het | 0/ >2500 | Reduced social interaction, delayed milestones, aphasia, stuttering and febrile seizures since age of three years. MRI indicative of diffuse neuroparenchymal atrophy, caudate atrophy and mineralization. Abdominal ultrasound suggestive of multiple cysts in both the kidneys and simple cyst in right lobe of liver | - | - | - | - | AD | In this study |
| 260 | S-19 | INADasd,d | *PLA2G6* | Int 2 | c.210-2A>G | NA | SS | | P | Hom | 0/ >2500 | Mild delay in milestones, regression, delayed motor development, autistic behavior, dystonia, optic nerve pallor and cerebellar atrophy | Yes | Het | Het | - | AR | In this study |
| 261 | S-3802 | Dystonia | *PLA2G6* | Ex 7 | c.1077G>A | p.Ser359= | SS | | P | Hom | 0/ >2500 | Features suggestive of INAD | - | - | - | - | AR | \*22934738 |
| 262 | R-0832 | Ataxia | *PLA2G6* | Ex 7 | c.1077G>A | p.Ser359= | SS | | P | Het | 0/ >2500 | Gait abnormalities, slurred speech and dysarthria. Brain MRI showed cerebellar atrophy | - | - | - | - | AR | \*22213678 |
| Ex 16 | c.2222G>A | p.Arg741Gln | MS | | LP | Het | 2θ/ >2500 | - | - | - | - | AR | \*27268037 |
| 263 | S-4155 | Ataxia | *PLA2G6* | Ex 14 | c.1999G>T | p.Glu667Ter | NS | | LP | Hom | 0/ >2500 | Developmental delay, motor regression, ataxia, nystagmus, cerebellar atrophy and generalised motor neuronal axonopathy | Yes | - | - | - | AR | In this study |
| 264 | S-4795 | Ataxia, Spasticity | *PLA2G6* | Ex 14 | c.1982C>T | p.Thr661Met | MS | | VUSD | Het | 0/ >2500 | Global developmental delay, abnormal gait, ataxia, spasticity, toe walking, brisk reflexes, cerebellar atrophy, behavioral disturbances and mild cognitive impairment | - | - | - | - | AR | \*29454663 |
| Ex 16 | c.2222G>A | p.Arg741Gln | MS | | LP | Het | 2θ/ >2500 | - | - | - | - | AR | \*27268037 |
| 265 | S-1249 | Dystonia | *PLA2G6* | Ex 16 | c.2222G>A | p.Arg741Gln | MS | | LP | Hom | 2θ/ >2500 | Fleeting depression symptoms since the last 5-6 years, tremors, stiffness, postural instability, dystonic posturing and urinary incontinence | - | - | - | - | AR | \*18570303 |
| 266 | S-2562 | Epilepsy, Microcephaly | *PNKP* | Ex 2 | c.35T>C | p.Leu12Pro | MS | | VUS | Het | 0/ >2500 | Microcephaly, global developmental delay, seizures and dysmorphism | - | - | - | - | AR | In this study |
| Int 10 | c.936+1G>T | NA | SS | | LP | Het | 0/ >2500 | - | - | - | - | AR | In this study |
| 267 | S-339 | Epilepsy | *PNPO* | Ex 3 | c.346C>T | p.Arg116Ter | NS | | P | Het | 0/ >2500 | Febrile seizures, macrostomia, short palpebral fissures, hypertelorism, facial dysmorphism, prominent ears and small eyes | - | - | - | - | AR | rs148839273 |
| Second variant not detected | | | | | | | | | | | | | | | |
| 268 | S-1439 | Epilepsy | *POLG* | Ex 4 | c.911T>G | p.Leu304Arg | MS | | P | Hom | 1θ/ >2500 | Mitochondrial cytopathy, basal ganglia abnormality, cerebral atrophy, encephalopathy, generalized seizures, epilepsy, delayed motor development, failure to thrive, regression of milestones since 15 months of age, hypotonia, and attention and cognitive deficit | - | - | - | - | AR | \*21880868 |
| 269 | S-4266 | Muscular dystrophy | *POMGNT1* | Ex 7 | c.617G>A | p.Trp206Ter | NS | | LP | Hom | 0/ >2500 | Features suggestive of muscular dystrophy. Brain MRI showed anterior pachygyria and pontine hypoplasia with cerebellar vermian atrophy. Biochemical tests showed elevated CPK and SGPT/SGOT enzymes. | - | - | - | - | AR | \*\*gnomAD |
| 270 | S-2947 | Muscular dystrophy | *POMT2* | Ex 9 | c.1057G>A | p.Gly353Ser | MS | | LP | Hom | 0/ >2500 | Hypotonia, developmental delay, difficulty in lifting head, poor axial control and increased CPK | - | - | - | - | AR | \*18513969 |
| 271 | S-5455 | Epilepsy | *PPT1* | Ex 5 | c.451C>T | p.Arg151Ter | NS | | P | Hom | 0/ >2500 | Features suggestive of epilepsy | - | - | - | - | AR | \*25205113 |
| 272 | S-5616 | Neuropathy | *PRX* | Ex 6 | c.231C>A | p.Tyr77Ter | NS | | LP | Hom | 0/ >2500 | Features suggestive of neuropathy | - | - | - | - | AR | In this study |
| 273 | S-495 | Epilepsy | *PTEN* | Ex 5 | c.304A>T | p.Lys102Ter | NS | | LP | Het | 0/ >2500 | Craniosynostosis, macrocephaly, seizures, behavioural problems, delayed speech, hypermetropia, papilliedema and slender fingers | Yes | Ref | Ref | - | AD | In this study |
| 274 | S-485 | Myopathy | *PYGM* | Ex 10 | c.1098G>A | p.Trp366Ter | NS | | P | Het | 0/ >2500 | Abnormal positioning of limbs, myopathy, recurrent cramps, infection triggered rhabdomyolysis with secondary renal and liver damage | - | - | - | - | AR | \*21802952 |
| Second variant not detected | | | | | | | | | | | | | | | |
| 275 | S-3829 | Warburg micro syndromeo | *RAB3GAP1* | Ex 2 | c.28G>T | p.Glu10Ter | NS | | LP | Hom | 0/ >2500 | Microcornea and thin corpus callosum | - | - | - | - | AR | In this study |
| 276 | S-2690 | Spasticity | *RAB3GAP2* | Ex 19 | c.1937\_1938delAT | p.Tyr646Terfs | Indel | | LP | Hom | 0/ >2500 | Bilateral congenital cataract, stiffness on both legs and toe walking | - | - | - | - | AR | In this study |
| 277 | S-1987 | AGSo | *RNASEH2C* | Ex 2 | c.205C>T | p.Arg69Trp | MS | | P | Hom | 7θ/ >2500 | Developmental delay, microcephaly, seizures and CT scan revealed diffuse calcification | Yes | Het | Het | - | AR | \*21177854 |
| 278 | S-4256 | Epilepsy, Microcephaly | *RNASEH2C* | Ex 2 | c.205C>T | p.Arg69Trp | MS | | P | Het | 7θ/ >2500 | Neurodevelopmental delay, seizures, strabismus, hypotonia, microcephaly, hypomyelination, spasticity, flat faces and visual impairment | - | - | - | - | AR | \*21177854 |
| Second variant not detected | | | | | | | | | | | | | | | |
| 279 | S-2825 | Leukodystrophy | *RNASEH2C* | Ex 2 | c.205C>T | p.Arg69Trp | MS | | P | Hom | 7θ/ >2500 | Calcification, glaucoma and white matter signal changes | - | - | - | - | AR | \*29150899 |
| 280 | S-4788 | Developmental delay | *RNASEH2A* | Ex 3 | c.322C>T | p.Arg108Trp | MS | | LP | Hom | 0/ >2500 | Global developmental delay and regression | Yes | Het | Het | - | AR | \*17846997 |
| 281 | S-5217 | Epilepsy | *SCN1A* | Int 1 | c.265-1G>T | NA | SS | | LP | Het | 0/ >2500 | Febrile and afebrile generalized seizures at 2.5 months of age | - | - | - | - | AD | In this study |
| 282 | S-3764 | Dravet syndromee | *SCN1A* | Ex 2 | c.302G>A | p.Arg101Gln | MS | | P | Het | 0/ >2500 | Seizures starting at the age of three months, developmental delay, hyperactivity, attention and cognitive deficit and is currently on multiple anti epileptic drugs | - | - | - | - | AD | \*20431604 |
| 283 | S-3457 | Epilepsy | *SCN1A* | Int 4 | c.602+1G>A | NA | SS | | P | Het | 0/ >2500 | Encephalopathy, seizures, regression of milestones and photosensitivity | - | - | - | - | AD | \*20562086 |
| 284 | S-4925 | Dravet syndromee | *SCN1A* | Int 4 | c.602+1G>A | NA | SS | | P | Het | 0/ >2500 | Febrile focal seizures | - | - | - | - | AD | \*24902755 |
| 285 | S-4940 | Epilepsy | *SCN1A* | Ex 6 | c.955C>T | p.Gln319Ter | NS | | P | Het | 0/ >2500 | Global developmental delay, both febrile and afebrile seizures since three months of age. Brain MRI showed unmyelinated white matter and FLAIR hyperintensities in bilateral parietal periventricular white matter | - | - | - | - | AD | In this study |
| 286 | S-5248 | Epilepsy | *SCN1A* | Int 6 | c.964+2T>G | NA | SS | | LP | Het | 0/ >2500 | Features of Dravet syndrome | - | - | - | - | AD | In this study |
| 287 | S-5621 | Epilepsy | *SCN1A* | Ex 9 | c.1284delA | p.Glu428AspfsTer20 | Indel | | P | Het | 0/ >2500 | Swelling on left side of neck at birth and seizures with clonic jerking | - | - | - | - | AD | In this study |
| 288 | S-2483 | Epilepsy | *SCN1A* | Int 14 | c.2589+3A>T | NA | SS | | P | Het | 0/ >2500 | Refractory epilepsy | - | - | - | - | AD | \*26096185 |
| 289 | S-5561 | Dravet syndromee | *SCN1A* | Int 14 | c.2589+3A>T | NA | SS | | P | Het | 0/ >2500 | Seizures | - | - | - | - | AD | \*26096185 |
| 290 | S-1532 | Dravet syndromee | *SCN1A* | Ex 15 | c.2624C>A | p.Thr875Lys | MS | | LP | Het | 0/ >2500 | Epilepsy and recurrent febrile seizures | Yes | - | Het (a) | - | AD | \*20522430 |
| 291 | S-4101 | Epilepsy, Microcephaly | *SCN1A* | Ex 19 | c.3733C>T | p.Arg1245Ter | NS | | P | Het | 0/ >2500 | Recurrent seizures, mild motor delay and mild microcephaly | - | - | - | - | AD | \*16458823 |
| 292 | S-3756 | Dravet syndromee | *SCN1A* | Ex 21 | c.4250delA | p.Asn1417MetfsTer2 | Indel | | P | Het | 0/ >2500 | Focal and myoclonic seizures | - | - | - | - | AD | In this study |
| 293 | S-4722 | Epilepsy, ASD | *SCN1A* | Int 22 | c.4338+1G>A | NA | SS | | P | Het | 0/ >2500 | Epilepsy, global developmental delay with hyperactivity and autistic features | Yes | - | - | - | AD | \*11359211 |
| 294 | S-255 | Epilepsy | *SCN1A* | Int 25 | c.4853-1G>C | NA | SS | | P | Het | 0/ >2500 | Epilepsy, myoclonic jerks in the upper limb, hypotonia, hyporeflexia, hypotension, metabolic acidosis, hepatic and renal failure | - | - | - | - | AD | \*17054684 |
| 295 | S-5280 | Epilepsy | *SCN1A* | Ex 26 | c.4942C>T | p.Arg1648Cys | MS | | P | Het | 0/ >2500 | Seizures from the age of 5 months | - | - | - | - | AD | \*10742094 |
| 296 | S-5194 | Myopathy | *RYR1* | Ex 27 | c.3748G>T | p.Glu1250Ter | NS | | LP | Het | 0/ >2500 | Weak neck flexors, waddling gait and difficulty in climbing stairs | - | - | - | - | AD | In this study |
| 297 | S-3088 | Dravet syndromee | *SCN1B* | Ex 3 | c.254G>A | p.Arg85His | MS | | LP | Hom | 0/ >2500 | Convulsions, myoclonic jerks, both febrile and afebrile siezures post DPT vaccination | - | - | - | - | AR | \*17020904 |
| 298 | S-1722 | Epilepsy, Ataxia, ASD | *SCN2A* | Ex 25 | c.4471delA | p.Thr1491GlnfsTer11 | Indel | | LP | Het | 0/ >2500 | Global developmental delay, intermittent ataxia, seizures and autistic features | - | - | - | - | AD | In this study |
| 299 | S-361 | Myopathy | *SEPN1* | Ex 2 | c.249\_250dupGG | p.Asp84GlyfsTer17 | Indel | | P | Het | 0/ >2500 | Delay in attaining motor milestones, floppiness of head and frequent falls suggestive of congenital myopathy | Yes | - | - | - | AR | In this study |
| Ex 7 | c.999delG | p.Asn335ThrfsTer5 | Indel | | P | Het | 0/ >2500 | Yes | - | - | - | AR | In this study |
| 300 | S-2642 | Ataxia | *SETX* | Ex 6 | c.502C>T | p.Arg168Trp | MS | | VUSD | Het | 0/ >2500 | Gait imbalance and dysarthria for the past 6 years. Alpha-fetoprotein was elevated and a CT scan showed cerebellar atrophy | - | - | - | - | AR | \*26811093 |
| Ex 12 | c.5475dupA | p.Glu1826ArgfsTer4 | Indel | | P | Het | 0/ >2500 | - | - | - | - | AR | In this study |
| 301 | S-1372 | Muscular dystrophy, myopathy | *SEPN1* | Ex 6 | c.827\_829dupCCT | p.Ala276\_Cys277insSer | Indel | | VUSD | Het | 0/ >2500 | Mild motor delay, kyphoscoliosis, hypotonia, hyperreflexia and proximal muscle weakness | - | - | - | - | AR | \*26780752 |
| Int 9 | c.1282-2A>C | NA | SS | | P | Het | 0/ >2500 | - | - | - | - | AR | \*21670436 |
| 302 | S-2844 | Ataxia, Neuropathy | *SETX* | Int 6 | c.718+1G>A | NA | SS | | LP | Hom | 0/ >2500 | Difficulty in walking, climbing stairs and speaking, tremors, nystagmus, pes cavus, hypertrophy of dorsal extension, mild scoliosis and was positive for Romberg's sign | - | - | - | - | AR | In this study |
| 303 | S-4214 | Spasticity | *SETX* | Ex 14 | c.5927T>G | p.Leu1976Arg | MS | | P | Het | 0/ >2500 | Spasticity of lower limbs, scoliosis and developmental delay | Yes | Het | Ref | - | AR | \*17159128 |
| Second variant not detected | | | | | | | | | | | | | | | |
| 304 | S-3821 | LGMDmd | *SGCA* | Ex 3 | c.197T>A | p.Leu66His | MS | | LP | Het | 1θ/ >2500 | Abnormal gait, frequent falls and proximal muscle weakness | - | - | - | - | AR | ΔRCV000384383.1 |
| Ex 3 | c.220C>T | p.Arg74Trp | MS | | P | Het | 1θ/ >2500 | - | - | - | - | AR | \*18285821 |
| 305 | S-3707 | LGMDmd | *SGCA* | Ex 5 | c.409G>A | p.Glu137Lys | MS | | P | Het | 0/ >2500 | Features suggestive of LGMD | - | - | - | - | AR | \*18285821 |
| Ex 6 | c.614C>A | p.Pro205His | MS | | P | Het | 0/ >2500 | - | - | - | - | AR | \*9032047 |
| 306 | S-4778 | Muscular dystrophy | *SGCB* | Ex 3 | c.271C>T | p.Arg91Cys | MS | | LP | Het | 1θ/ >2500 | Pain in lower limbs and pseudohypertrophy of calf muscles | - | - | - | - | AR | \*22095924 |
| Second variant not detected | | | | | | | | | | | | | | | |
| 307 | S-1651 | Muscular dystrophy | *SGCG* | Ex 8 | c.848G>A | p.Cys283Tyr | MS | | P | Hom | 0/ >2500 | Features suggestive of DMD | - | - | - | - | AR | \*10720277 |
| 308 | S-4682 | Developmental delay | *SGSH* | Ex 1 | c.83T>C | p.Leu28Pro | MS | | VUS | Het | 0/ >2500 | Delayed speech and motor development, cognitive regression, hyperactivity, hypertrichosis, short stature, coarse facies and mild dysostosis multiplex | - | - | - | Het (a) | AR | In this study |
| Ex 5 | c.571G>A | p.Gly191Arg | MS | | LP | Het | 0/ >2500 | - | - | - | Het (a) | AR | \*15146460 |
| 309 | S-1739 | Neuropathy | *SH3TC2* | Int 4 | c.385+1G>A | NA | SS | | LP | Hom | 0/ >2500 | Neuropathy and myopathy | - | - | - | - | AR | In this study |
| 310 | S-3102 | Neuropathy | *SH3TC2* | Ex 8 | c.931A>T | p.Lys311Ter | NS | | P | Hom | 0/ >2500 | Delayed motor development, abnormal gait, scoliosis and poor scholastic performance | - | - | - | - | AR | In this study |
| 311 | S-246 | Developmental delay | *SH3TC2* | Ex 11 | c.2491\_2492delAG | p.Leu832HisfsTer8 | Indel | | P | Het | 0/ >2500 | Developmental delay, inability to walk or sit without support, pigmented sparse hair, hypotonia and pyloric stenosis | - | - | - | - | AR | \*20301514 |
| Second variant not detected | | | | | | | | | | | | | | | |
| 312 | S-3696 | Ataxia | *SIL1* | Int 7 | c.767+2T>A | NA | SS | | LP | Hom | 0/ >2500 | Cataract, cerebellar ataxia and global developmental delay | - | - | - | - | AR | In this study |
| 313 | S-3981 | Ataxia | *SIL1* | Int 9 | c.1030-18G>A | NA | SS | | P | Hom | 0/ >2500 | Global developmental delay with features suggestive of cerebellar ataxia | Yes | Het | Het | - | AR | \*25473114 |
| 314 | S-5077 | Ataxia | *SIL1* | Ex 10 | c.1260\_1279delinsCAC | p.Arg420SerfsTer3 | Indel | | P | Hom | 0/ >2500 | Cerebellar ataxia and cataract | - | - | - | - | AR | In this study |
| 315 | S-722 | Ataxia, Neuropathy, ID | *SLC12A6* | Ex 20 | c.2655dupT | p.Ala886CysfsTer9 | Indel | | LP | Hom | 0/ >2500 | Neuropathy, sensory ataxia, proximal muscle weakness, tremors, delayed speech and ID | - | - | - | - | AR | In this study |
| 316 | S-2432 | Developmental delay | *SLC16A2* | Ex 3 | c.972G>A | p.Trp324Ter | NS | | LP | Hemi | 0/ >2500 | Cerebral atrophy and global developmental delay | Yes | - | Het | Hemi (a) | XL | In this study |
| 317 | S-3475 | Leigh syndromeo | *SLC19A3* | Ex 2 | c.4delG | p.Asp2IlefsTer7 | Indel | | LP | Hom | 0/ >2500 | Encephalopathy, seizures, microcephaly, lethargy and elevated levels of methylmalonic acid (MMA). Brain MRI revealed hyperintensities in putamen and swollen thalamus | Yes | - | - | - | AR | In this study |
| 318 | S-3778 | Myopathy | *SLC22A5* | Ex 6 | c.1007G>A | p.Arg336Gln | MS | | VUS | Het | 0/ >2500 | Congenital myopathy, hypotonia, motor development delay and floppy infant. | - | - | - | - | AR | #rs759529143 |
| Ex 7 | c.1195C>T | p.Arg399Trp | MS | | LP | Het | 1θ/ >2500 | - | - | - | - | AR | \*28841266 |
| 319 | S-3173 | Epilepsy | *SLC25A22* | Int 4 | c.202+1dupG | NA | SS | | LP | Hom | 0/ >2500 | Global developmental delay, seizures, hypoplasia of corpus callosum, coarse features, hirsuitism, dysmorphism, rocker bottom feet and hypospadias | Yes | Het | Het | - | AR | In this study |
| 320 | S-4145 | Epilepsy | *SLC25A22* | Int 9 | c.818+1G>C | NA | SS | | LP | Hom | 0/ >2500 | Generalized seizures, global developmental delay, hypotonia, dyskinesia, cognitive deficit and speech delay | - | - | - | - | AR | In this study |
| 321 | S-898 | Epilepsy | *SLC26A4* | Ex 11 | c.1334T>G | p.Leu445Trp | MS | | P | Het | 0/ >2500 | Seizures, global developmental delay, sparse scalp hair, no social smile and eye contact. Electroencephalogram revealed hypsarrhythmia | - | - | - | - | AR | \*18285825 |
| Second variant not detected | | | | | | | | | | | | | | | |
| 322 | S-5628 | Epilepsy | *SLC2A1* | Ex 3 | c.195G>A | p.Trp65Ter | NS | | P | Het | 0/ >2500 | Seizures, spastic quadriplegia, walking and speech difficulties, vacant and unresponsive stare, episodes of excessive sleep and delayed developmental milestones. | Yes | Ref | Ref | - | AD | \*26193382 |
| 323 | S-1891 | Epilepsy, Microcephaly | *SLC2A1* | Ex 5 | c.553delC | p.Leu185CysfsTer6 | Indel | | LP | Het | 0/ >2500 | Multiple types of epilepsy, microcephaly, delayed motor and cognitive development, axial hypotonia ad episodes of nystagmus. MRI showed hypomyelination | - | - | - | - | AD | In this study |
| 324 | S-3231 | Ataxia, Microcephaly, ID | *SLC2A1* | Ex 6 | c.844delC | p.Gln282SerfsTer58 | Indel | | LP | Het | 0/ >2500 | Microcephaly, global developmental delay, episodic ataxia, ID, brisk tendon reflexes and coarse facial features. | - | - | - | - | AD | In this study |
| 325 | S-221 | Epilepsy, Microcephaly | *SLC2A1* | Ex 8 | c.997C>T | p.Arg333Trp | MS | | P | Het | 0/ >2500 | Encephalopathy, seizures, global developmental delay, microcephaly, cognitive defect, delayed speech, spasticity and hypoglycorrhachia | - | - | - | - | AD | \*23448551 |
| 326 | S-4302 | Epilepsy, Ataxia | *SLC2A1* | Ex 9 | c.1105\_1112del | p.Ile369HisfsTer9 | Indel | | LP | Het | 0/ >2500 | Episodic ataxia, absence epilepsy, reduced concentration, restlessness and a decline in academic performance | Yes | Ref | Ref | - | AD | In this study |
| 327 | S-3390 | ASD | *SLC6A5* | Int 3 | c.679+2T>C | NA | SS | | LP | Hom | 1θ/ >2500 | Hyperekplexia, global developmental delay and autistic features | - | - | - | - | AR | In this study |
| 328 | S-5036 | Developmental delay | *SMC1A* | Ex 15 | c.2394delA | p.Lys798AsnfsTer31 | Indel | | LP | Het | 0/ >2500 | Global developmental delay. Brain MRI shows semilobar holoprosencephaly with colpocephaly | Yes | Ref | Ref | - | XL | In this study |
| 329 | S-5409 | Epilepsy | *SMC1A* | Ex 22 | c.3305\_3312del | p.Asn1102ArgfsTer53 | Indel | | LP | Het | 0/ >2500 | Inattention, poor eye contact, multiple mongoloid spots, hypotonia, hypertrichosis and epileptic encephalopathy with mild phenotypes of CdLS. | Yes | Ref | Ref | - | XL | In this study |
| 330 | S-5854 | Neuropathy | *SNAP29* | Ex 3 | c.487dupA | p.Ser163LysfsTer6 | Indel | | P | Het | 1θ/ >2500 | Global developmental delay, spastic paraparesis, positive cerebellar signs and peeling of skin. Brain MRI showed hypomyelination | - | - | - | - | AR | \*21073448 |
| Ex 4 | c.586C>T | p.Arg196Ter | NS | | LP | Het | 0/ >2500 | - | - | - | - | AR | \*\* gnomAD |
| 331 | S-4250 | Muscular dystrophy | *SPG11* | Ex 11 | c.2146C>T | p.Gln716Ter | NS | | P | Hom | 1θ/ >2500 | Muscular dystrophy | - | - | - | - | AR | \*20390432 |
| 332 | S-7220 | Spasticity | *SPG11* | Ex16 | c.3018T>G | p.Tyr1006Ter | NS | | P | Hom | 0/ >2500 | Progressive spastic paraplegia and cognitive impairment | - | - | - | - | AR | In this study |
| 333 | S-4523 | Spasticity | *SPG11* | Ex 32 | c.6100C>T | p.Arg2034Ter | NS | | P | Hom | 0/ >2500 | Problems with walking, talking and memory | - | - | - | - | AR | \*18332254 |
| 334 | S-4351 | Spasticity | *SPG11* | Ex 38 | c.6856C>T | p.Arg2286Ter | NS | | P | Hom | 0/ >2500 | Difficulty in walking and learning | - | - | - | - | AR | \*24090761 |
| 335 | S-4130 | Ataxia | *SQSTM1* | Ex 5 | c.712\_713insTCCTCCG  AGTGTGAATTTCCTGA | p.Lys238IlefsTer5 | Indel | | LP | Hom | 0/ >2500 | Ataxia, cognitive decline, oculomotor and eyelid apraxia | - | - | - | - | AR | In this study |
| 336 | S-2368 | Ataxia | *SQSTM1* | Ex 6 | c.823\_824delAG | p.Ser275PhefsTer17 | Indel | | P | Hom | 0/ >2500 | Ataxia, abnormal gait, hypotonia, clumsiness of all four limbs, recurrent falls, opthalmoplegia, abnormal eye movements, fundus albipunctatus, lactic acidemia and pancerebellar signs | - | - | - | - | AR | ΔRCV000519042.1 |
| 337 | S-2673 | Epilepsy | *STXBP1* | Ex 10 | c.841\_845delCTGGA | p.Leu281ArgfsTer31 | Indel | | LP | Het | 0/ >2500 | Seizures since day 5 of her life and infantile spasms | - | - | - | - | AD | In this study |
| 338 | S-3287 | Epilepsy | *STXBP1* | Ex 10 | c.841\_845delCTGGA | p.Leu281ArgfsTer31 | Indel | | LP | Het | 0/ >2500 | Seizures since day 5 of her life and infantile spasms | - | - | - | - | AD | In this study |
| 339 | S-3458 | Epilepsy | *STXBP1* | Ex 16 | c.1439C>T | p.Pro480Leu | MS | | P | Het | 0/ >2500 | Encephalopathy, generalized seizures since day 4of life and EEG showed multifocal spikes with burst suppression | Yes | Ref | Ref | - | AD | \*26514728 |
| 340 | S-3181 | Leigh syndromeo | *SURF1* | Ex 6 | c.535dupA | p.Arg179LysfsTer12 | Indel | | P | Hom | 0/ >2500 | Floppiness and inability to walk | - | - | - | - | AR | In this study |
| 341 | S-3233 | Leigh syndromeo | *SURF1* | Ex 7 | c.751C>T | p.Gln251Ter | NS | | P | Hom | 0/ >2500 | Hypotonia in upper and lower limbs, developmental delay, mild facial dysmorphism and decreased reflexes. MRI showed bilateral cerebellar subcortical white matter with bilateral caudate lentiform | - | - | - | - | AR | \*10636738 |
| 342 | S-776 | Muscular dystrophy | *SYNE1* | Ex 83 | c.16015C>T | p.Arg5339Ter | NS | | LP | Het | 0/ >2500 | Muscle wasting, muscle weakness, tight tendo achilles, restricted elbow movement, difficulty in walking and chronic generalized benign neurogenic disease with mild atrophy and fibrosis. A high level of CPK was detected | - | - | - | - | AD | In this study |
| 343 | S-2171 | Epilepsy, ASD | *SYNGAP1* | Ex 10 | c.1591\_1592delTG | p.Cys531HisfsTer17 | Indel | | LP | Het | 0/ >2500 | Manifested global developmental delay with autistic features, bruxism, depressed nasal bridge, bulbous nose, self harming at night and excessive cry. MRI was normal and EEG was suggestive of multifocal epilepsy. | - | - | - | - | AD | In this study |
| 344 | S-1331 | Epilepsy, ASD | *SYNGAP1* | Ex 15 | c.2474\_2477dupCGGA | p.Ile827GlyfsTer24 | Indel | | LP | Het | 0/ >2500 | Myoclonus seizures, autistic symptoms and global developmental delay | - | - | - | - | AD | In this study |
| 345 | R-0840 | Developmental delay | *TCF4* | Int 8 | c.550-2A>C | NA | SS | | LP | Het | 0/ >2500 | Developmental delay, poor cognition, coarse face, macrostomia, hand flapping movement, divergent squint and prominent upper lip | - | - | - | - | AD | In this study |
| 346 | S-5111 | Muscular dystrophy | *TK2* | Ex 6 | c.416C>T | p.Ala139Val | MS | | P | Hom | 0/ >2500 | Congenital muscular dystrophy | - | - | - | - | AR | \*25446393 |
| 347 | S-3156 | Myopathy | *TNNT1* | Int 2 | c.32+1G>A | NA | SS | | LP | Hom | 1θ/ >2500 | Global developmental delay, hypomyelination, spasticity, hypotonia, flat facies and has a history of old fractures | - | - | - | - | AR | \*\*gnomAD |
| 348 | S-2314 | Myopathy | *TNNT1* | Int 12 | c.750+1G>A | NA | SS | | LP | Hom | 0/ >2500 | Motor delay, proximal muscle weakness, dysmorphism and chest deformity | - | - | - | - | AR | #rs111998831 |
| 349 | S-2587 | NCLe | *TPP1* | Ex 5 | c.496dupC | p.His166ProfsTer22 | Indel | | P | Het | 1θ/ >2500 | Seizures, shaking of hand, shivering of body. MRI showed prominent cerebral cortical sulci and atrophy of ventricles, severe dysfunction of the visual pathway and speech delay | - | - | - | - | AR | In this study |
| Ex 6 | c.622C>T | p.Arg208Ter | NS | | P | Het | 0/ >2500 | - | - | - | - | AR | \*9295267 |
| 350 | S-1079 | Epilepsy, Microcephaly, ID | *TPP1* | Int 9 | c.1146-1G>A | NA | SS | | P | Hom | 0/ >2500 | Neuroregression, seizures, encephalopathy, muscle wasting and weakness, delayed motor development, microcephaly, joint contractures, ID and impaired vision and hearing. MRI of the brain showed cerebral and cerebellar atrophy | - | - | - | - | AR | ΔRCV000494500 |
| 351 | S-2691 | Epilepsy | *TPP1* | Ex 10 | c.1264C>T | p.Gln422Ter | NS | | LP | Hom | 1θ/ >2500 | Seizures, developmental regression and is currently on anti-epileptic medication. MRI showed diffuse cerebellar atrophy and EEG showed bilateral occipital epileptiform abnormalities | - | - | - | - | AR | In this study |
| 352 | S-2684 | Muscular dystrophy | *TTN* | Ex 275 | c.72686dupC | p.Ser24230IlefsTer8 | Indel | | LP | Het | 0/ >2500 | Progressive proximal weakness of lower and upper limbs since last eight years, calf pseudohypertrophy and winging of scapula | - | - | - | - | AR | In this study |
| Ex 311 | c.99683A>C | p.Glu33228Ala | MS | | VUS | Het | 0/ >2500 | - | - | - | - | AR | ΔRCV000642697.1 |
| 353 | S-3342 | Ataxia | *TTPA* | Ex 1 | c.26C>A | p.Ser9Ter | NS | | LP | Hom | 0/ >2500 | Slurred speech and motor difficulties | Yes | Het | Het | Hom (u) | AR | In this study |
| 354 | S-3161 | Ataxia, Microcephaly | *UBE3A* | Ex 3 | c.1480\_1481delCA | p.Gln494AlafsTer11 | Indel | | LP | Het | 0/ >2500 | Delayed motor development, microcephaly, hypotonia, abnormal gait, ataxia and abnormal ECG | - | - | - | - | AD | In this study |
| 355 | S-1732 | ASD | *UBE3A* | Ex 5 | c.1811\_1812delGT | p.Cys604TyrfsTer23 | Indel | | P | Het | 0/ >2500 | Attention deficiency, delayed motor development, cognitive deficit, delayed speech, global developmental delay and autistic features | Yes | - | - | Het (a) | AD | \*25212744 |
| 356 | S-6581 | Developmental delay | *UNC80* | Ex 55 | c.8215\_8227del | p.Leu2739GlnfsTer22 | Indel | | LP | Hom | 0/ >2500 | Muscle weakness, hypotonia, developmental delay, abnormal jerky movements, poor visual fixation, hyperekplexia and poor head control. MRI showed diffused cerebral atrophy and white matter hyperintensity | Yes | Het | Het | - | AR | In this study |
| 357 | S-569 | Neuropathy | *VRK1* | Int 12 | c.1159+1G>A | NA | SS | | LP | Hom | 0/ >2500 | Bilateral foot drop and high stepping gait, difficulty in getting up from squatting position and weak bilateral interosseous nerve, suggesting axonal neuropathy | Yes | Het | Het | Het (a) | AR | In this study |
| 358 | S-96 | Epilepsy | *WDR45* | Ex 12 | c.1007\_1008delAT | p.Tyr336CysfsTer5 | Indel | | P | Het | 1θ/ >2500 | Seizures, slurred speech and progressive abnormal behaviour. MRI was suggestive of NBIA | - | - | - | - | XL | \*23176820 |
| 359 | S-2563 | Joubert syndromeo | *WDR62* | Int15 | c.1959-2A>G | NA | SS | | LP | Hom | 0/ >2500 | Microcephaly, global developmental delay and profound mental retardation | Yes | Het | Het | - | AR | \*\* gnomAD |
| 360 | S-5186 | Epilepsy | *WWOX* | Ex 7 | c.735dupT | p.Val246CysfsTer24 | Indel | | LP | Hom | 0/ >2500 | Refractory epilepsy and developmental delay | - | - | - | - | AR | In this study |
| 361 | S-2908 | MWSo | *ZEB2* | Ex 8 | c.1791\_1813dup | p.Lys605IlefsTer10 | Indel | | P | Het | 0/ >2500 | Developmental delay, dolichocephaly, bilaterally raised ear lobule, broadening of eyelashes and undescended testis | - | - | - | - | AD | In this study |
| 362 | S-2656 | Spasticity | *ZFYVE26* | Ex 36 | c.6692T>A | p.Leu2231Ter | NS | | LP | Hom | 0/ >2500 | Progressive spasticity of lower limbs | - | - | - | - | AR | In this study |

**Abbreviations**:

AAA-Achalasia-Addisonianism-Alacrima syndrome; AD-Autosomal dominant; AGS- Aicardi-goutieres syndrome; AHC-Alternating hemiplegia of childhood; ALS- Amyotrophic lateral sclerosis; AR-Autosomal recessive; ASD-Autism spectrum disorders; CdLS-Cornelia de Lange syndrome; CPK- Creatine Phosphokinase; CS-Clinical significance; Del-Deletion; DMD-Duchenne muscular dystrophy; Dup-Duplication; Ex-Exon; F-Father; Hemi-Hemizygous; Het-Heterozygous; Hom-Homozygous; ID-Intellectual disability; INAD-Infantile neuroaxonal dystrophy; Inc-Inconclusive; Indel- Small deletion/duplication/insertion or insertion/deletion; Int-Intron; LGMD-Limb girdle muscular dystrophy; LP-Likely pathogenic; M-Mother; MD-CMD-Merosin-deficient congenital muscular dystrophy; MOI-Mode of inheritance; MS-Missense; MWS-Mowat-Wilson syndrome; NCL-Neuronal ceroid lipofuscinosis; NS-Nonsense; P-Pathogenic; Sibl-Sibling; SLS-Sjogren-Larsson syndrome; SS-Splice site; SV- Sanger Validation; UCMD-Ullrich muscular dystrophy; VLCFA- Very Long Chain Fatty Acids; VT-Variant type; VUS-Variant of uncertain significance; VUSD-Variant of uncertain significance with probable damaging effect; XL-X-linked; (a)-Affected; (u)-Unaffected; (unk)-Unknown.

Δ: ClinVar database ID, #: dbSNP database ID, \*: Reference from PubMed database (PMID), \*\*: Reported in genome Aggregation Database (gnomAD)

Θ: Identified variant observed in control population in heterzygous state for an autosomal recessive condition

µ: Incomplete penetrance has been reported for this gene

For the disease conditions in clinical subtypes, following abbreviation have been used;

a: Ataxia; asd: Autism spectrum disorders; d: Dystonia; e: Epilepsy; l: Leukodystrophy; m: Microcephaly; md: Muscular dystrophy; n: Neuropathy; o: Others

**Table S4: List of structural variants identified in the study.**

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Sl. No.** | **Sample**  **ID** | **Disease condition** | **Gene** | **Chr. coordinates** | **Ex/Int** | **Nucleotide** | | **VT** | | **CS** | | **Zygosity** | | **MOI** | | **Reference** | |
| 1 | S-2588 | Ataxia | *ATM* | chr11:108137898?\_108138069+?del | Ex 17 del | c.(2466+1\_2467-1)\_(2638+1\_2639-1)  del | | Large del | | LP | | Het | | AR | | In this study | |
| chr11:108216596C>T | Ex 58 | c.8545C>T | | NS | | P | | Het | | AR | | ΔRCV000407552.4 | |
| 2 | S-3831 | Myopathy | *ATP8A2* | chr13:26273311-?\_26436546+?del | Ex 25-33 del | c.(2211+1\_2212-1)\_(3183+1\_3184-1)  del | | Large del | | LP | | Hom | | AR | | In this study | |
| 3 | S-4670 | ID | *BCL11A* | chr2:60687539-?\_60780405+?del | Ex 1-4 del | c.(?\_-1)\_(\*1\_?)del | | Large del | | P | | Het | | AD | | \*24810580 | |
| 4 | S-246 | Developmental delay | *CAPN3* | chr15:42701501-?\_42703971+?del | Ex 17-24 del | c.(1914+1\_1915-1)\_(\*1\_?)del | | Large del | | LP | | Het | | AR | | In this study | |
| Second variant not detected | | | | | | | | | | | | | |
| 5 | S-4663 | Epilepsy, ID | *CLN3* | chr16:28493426-?\_28493993+?del | Ex 11-14 del | c.(790+1\_791-1)\_(1056+1\_1057- 1)  del | Large del | | P | | Hom | | AR | | \*9932957 | |
| 6 | R-0109 | Spasticity | *CYB5R3* | chr22:43019795-?\_43024287+?del | Ex 5-8 del | c.(333+1\_334-1)\_(733+1\_734-1)  del | Large del | | LP | | Hom | | AR | | In this study | |
| 7 | S-3869 | Muscular dystrophy | *DMD* | chrX:31947713-?\_31986631+?del | Ex 45-47 del | c.(6438+1\_6439-1)\_(6912+1\_6913-1)del | Large del | | P | | Hemi | | XL | | \*20847377 | |
| 8 | S-4073 | Muscular dystrophy | *DMD* | chrX:31645790-?\_31697703+?del | Ex 53-55 del | c.(7660+1\_7661-1)\_(8217+1\_8218-1)del | Large del | | P | | Hemi | | XL | | \*20847377 | |
| 9 | S-5422 | Muscular dystrophy | *DMD* | chrX:31947713-?\_31986631+?del | Ex 45-47 del | c.(6438+1\_6439-1) \_ (6912+1\_6913-1)del | Large del | | P | | Hemi | | XL | | \*20847377 | |
| 10 | S-1083 | Becker muscular dystrophymd | *DMD* | chrX:31947713-?\_31986631+?del | Ex 45-47 del | c.(6438+1\_6439-1) \_(6912+1\_6913-1)del | Large del | | P | | Hemi | | XL | | \*20847377 | |
| 11 | S-1130 | Muscular dystrophy | *DMD* | chrX:31854835-?\_31986631+?del | Ex 45-49 del | c.(6438+1\_6439-1)\_(7200+1\_7201-1)del | Large del | | P | | Hemi | | XL | | \*20847377 | |
| 12 | S-130 | Muscular dystrophy | *DMD* | chrX:31196786-?\_31341775+?del | Ex 62-70 del | c.(9163+1\_9164-1)\_(10223+1\_10224-1)del | Large del | | P | | Hemi | | XL | | In this study | |
| 13 | S-224 | Muscular dystrophy | *DMD* | chrX:31747748-?\_31950344+?del | Ex 46-52 del | c.(6614+1\_6615-1)\_(7660+1\_7661-1)del | Large del | | P | | Hemi | | XL | | \*20847377 | |
| 14 | S-4015 | Muscular dystrophy | *DMD* | chrX:31525398-?\_31525570+?del | Ex 56 del | c.(8217+1\_8218-1)\_(8390+1\_8391-1)del | Large del | | LP | | Hemi | | XL | | In this study | |
| 15 | S-4696 | Muscular dystrophy | *DMD* | chrX:31893305-?\_31986631+?del | Ex 45-48 del | c.(6438+1\_6439-1)\_(7098+1\_7099-1)del | Large del | | P | | Hemi | | XL | | \*27582364 | |
| 16 | S-776 | Muscular dystrophy | *EMD* Ex 2-6 del along with 17 bp insertion and *FLNA* Ex 14-47 inverted dup | chrX:153608050?\_153609557+?delinsTGCAGTTCACTGCAATCinsNC\_000023.10:g.153577217-?\_153592740inv | *EMD* Ex 2-6 del along with  17 bp insertion and *FLNA*  Ex 14-47 inverted dup | c.(82+1\_831)\_(\*1\_?)delinsTGCAGTTCACTGCAATCinsNM\_001456.3:c.(2022+1\_2023-1)\_(\*1\_?)inv | Complex rearrangement | | P | | Hemi | | XL | | \*9140403 | |
| 17 | S-4491 | Epilepsy, Microcephaly | *GALC* | chr14:88401076-?\_88417092+?del | Ex 11-17 del | c.(1161+1\_1162-1)\_(\*31\_?)del | Large del | | P | | Hom | | AR | | \*11151421 | |
| 18 | S-1035 | Epilepsy, Neuropathy | *IGHMBP2* | chr11:68696651-?\_68707199+?del | Ex 8-15 del | c.(1060+1\_1061-1)\_(\*1\_?)del | Large del | | LP | | Hom | | AR | | In this study | |
| 19 | S-5508 | ASD | *MECP2* | chrX:153295818-?\_153298008+?del | Ex 3-4 del | c.(26+1\_27-1)\_(\*1\_?)del | Large del | | P | | Het | | XL | | \*17968969 | |
| 20 | R-0520 | CdLSo | *NIPBL* | chr5:36995724-?\_37038840+?del | Ex 11-34 del | c.(3121+1\_3122-1)\_(6108+1\_6109-1)del | Large del | | LP | | Het | | AD | | In this study | |
| 21 | S-2564 | CdLSo | *NIPBL* | chr5:36958206-?\_36958333+?del | Ex 4 del | c.(230+1\_231-1)\_(358+1\_359-1)del | Large del | | LP | | Het | | AD | | In this study | |
| 22 | S-5367 | Joubert syndromeo | *NPHP1* | chr2:110881368-?\_110962545+?del | Ex 1-20 del | c.(?\_-1)\_(\*1\_?)del | Large del | | P | | Hom | | AR | | \*15689444 | |
| 23 | S-6113 | Epilepsy, ASD | *NRXN1* | chr2:50692580-?\_51149018+?del | Ex 6-18 del | c.(919+1\_920-1)\_(3484+1\_3485- 1)  del | Large del | | P | | Het | | AD/  AR | | \*22617343 | |
| 24 | S-5906 | Sotos syndrome | *NSD1* | chr5:176636637-?\_176639196+?del | Ex 5 del | c.(1236+1\_1237-1)\_(3796+1\_3797-1)del | Large del | | P | | Het | | AD | | In this study | |
| 25 | S-2769 | Spasticity | *PAFAH1B1* | chr17:2541583-?\_2585096+?del | Ex 2-11 del | c.(?\_-1)\_(\*1\_?)del | Large del | | P | | Het | | AD | | \*18285425 | |
| 26 | S-6624 | PMD | *PLP1* | chrX:103031924?\_103045526+?dup | Ex 1-7 dup (whole gene dup) | c.(?\_-1)\_(\*1\_?)dup | Large dup | | P | | Hemi | | XL | | \*21679407 | |
| 27 | R-0536 | Leukodystrophy | *ROGDI* | chr16:4847461?\_4848668+?delinsCAG | Ex 7-11 del | c.432+462\_\*145delinsCAG | Large del | | LP | | Hom | | AR | | In this study | |
| 28 | S-2919 | Epilepsy | *SCN1A* | chr2:166908229-?\_166915198+?del | Ex 2-6 del | c.(264+1\_265-1)\_(964+1\_965-1)del | Large del | | LP | | Het | | AD | | \*19400878 | |
| 29 | S-3157 | ID | *TRIP12* | chr2:230656869?\_230744795+?dup | Ex 2-27 dup | c.(?\_-1)\_(3990+1\_3991-1)dup | Large dup | | LP | | Het | | AD | | In this study | |
| 30 | S-3882 | Microcephaly | *TSEN2* | chr3:12531300-?\_12531488+?del | Ex 2 del | c.(?\_-1)\_(189+1\_190-1)del | Large del | | LP | | Het | | AR | | In this study | |
| chr3:12571274A>T | Ex 10 | c.1150A>T | MS | | VUS | | Het | | AR | | In this study | |
| 31 | S-4436 | ID | *TUSC3* | chr8:15508206-?\_15621757+?del | Ex 3-11 del | c.(308+1\_309-1)\_ (\*1\_?)del | Large del | | LP | | Hom | | AR | | In this study | |
| 32 | S-2889 | Epilepsy, Microcephaly, ASD | chr1p36.31-p36.22 | chr1:5923325-?\_12267077+?del | *NPHP4, KCNAB2, ESPN, TNFRSF25, PLEKHG5, TAS1R1, CAMTA1, PER3, UTS2, PARK7, ENO1, CA6, H6PD, PIK3CD, NMNAT1, KIF1B, PGD, PEX14, TARDBP, MASP2, UBIAD1, MTHFR, NPPA, NPPB, PLOD1, MFN2, MIIP* and *TNFRSF1B* | NA | Large del | | P | | Het | | AD | | In this study | |
| 33 | S-2857 | Epilepsy | chr1p36.33-p36.23 | chr1:955553-?\_8934967+?del | *AGRN, TNFRSF4, B3GALT6, TAS1R3, CDK11A, GABRD, SKI, PEX10, MMEL1, TP73, NPHP4, KCNAB2, ESPN, TNFRSF25, PLEKHG5, TAS1R1, CAMTA1, PER3, UTS2, PARK7* and *ENO1* | NA | Large del | | P | | Het | | AD | | \*16690727 | |
| 34 | S-5116 | Epilepsy | chr1q43-q44 | chr1:243419476-?\_245027609+?del | *SDCCAG8, AKT3, ZBTB18* and *HNRNPU* | NA | Large del | | P | | Het | | AD | | \*26853090 | |
| 35 | S-2836 | Epilepsy | chr2q24.3 | chr2:165946660-?\_169313128+?del | *SCN3A, SCN2A, GALNT3, TTC21B, SCN1A, SCN9A, STK39* and *CERS6* | NA | Large del | | P | | Het | | AD | | \*25524840 | |
| 36 | S-3496 | Epilepsy | chr2q24.3 | chr2:165946660-?\_169038600+?del | *SCN3A, SCN2A, GALNT3, TTC21B, SCN1A, SCN9A* and *STK39* | NA | Large del | | P | | Het | | AD | | \*25524840 | |
| 37 | S-1887 | Epilepsy | chr2q24*.3* | chr2:166605291-?\_167168266+?del | *GALNT3, TTC21B, SCN1A* and *SCN9A* | NA | Large del | | LP | | Het | | AD | | \*25524840 | |
| 38 | S-594 | Developmental delay | chr2q37.1-q37.3 | chr2:232879545-?\_242795132+?del | *DIS3L2, PRSS56, CHRND, CHRNG, GIGYF2, KCNJ13, NEU2, ATG16L1, SAG, DGKD, UGT1A8, UGT1A10, UGT1A9, UGT1A7, UGT1A6, UGT1A5, UGT1A4, UGT1A3, UGT1A1, COL6A3, MLPH, HES6, PER2, TRAF3IP1, TWIST2, HDAC4, NDUFA10, CAPN10, KIF1A, AGXT, PASK, ANO7, HDLBP, D2HGDH* and *PDCD1* | NA | Large del | | LP | | Het | | AD | | \*28690993 | |
| 39 | S-5221 | Macrocephaly | chr4q21.21-q22.3 | chr4:78978724-?\_96256782+?del | *FRAS1, BMP2K, ANTXR2, COQ2, HELQ, WDFY3, ARHGAP24, MAPK10, PTPN13, DSPP, DMP1, IBSP, SPP1, PKD2, ABCG2, SNCA, SMARCAD1, PDLIM5, BMPR1B* and *UNC5C* | NA | Large del | | P | | Het | | AD | | \*20522426 | |
| 40 | S-4861 | Epilepsy, ASD | chr5q14.3 | chr5:86564269-?\_90459717+?del | *RASA1, MEF2C* and *GPR98* | NA | Large del | | P | | Het | | AD | | \*19471318 | |
| 41 | S-5188 | ASD | chr5q14.3-q15 | chr5:89854713-?\_96253309+?del | *GPR98, NR2F1, TTC37, PCSK1, CAST, ERAP1* and *ERAP2* | NA | Large del | | P | | Het | | AD | | \*19471318 | |
| 42 | S-582 | Developmental delay | chr5q35.2-q35.3 | chr5:175815821-?\_177422934+?del | *HIGD2A, SNCB, TSPAN17, UIMC1, FGFR4, NSD1, SLC34A1, F12, B4GALT7* and *PROP1* | NA | Large del | | P | | Het | | AD | | \*25510705 | |
| 43 | S-5411 | Microcephaly | chr6q27 | chr6:170591962-?\_170881353+?del | *DLL1* and *TBP* | NA | Large del | | LP | | Het | | AD | | \*24736736 | |
| 44 | S-3030 | Microcephaly | chr7p14.1 | chr7:40172658-?\_40314255+?del | *MPLKIP* and *SUGCT* | NA | Large del | | P | | Hom | | AR | | \*21296629 | |
| 45 | S-5204 | Developmental delay | chr7q11.23 | chr7:72742578-?\_74016760+?dup | *FKBP6, FZD9, MLXIPL, STX1A, ELN, LIMK1, RFC2* and *GTF2IRD1* | NA | Large dup | | P | | Het | | AD | | \*26333794 | |
| 46 | S-1592 | Attention deficit hyperactivity disordero | chr7q31.2-q31.32 | chr7:116165117-?\_122635688+?del | *CAV1, MET, ST7, CFTR, CTTNBP2, KCND2, TSPAN12, AASS* and *TAS2R16* | NA | Large del | | LP | | Het | | AD | | \*28224041 | |
| 47 | S-4962 | Kabuki syndromeo | chr8p23.3-p23.1 | chr8:1496860-?\_6728348+?del | *DLGAP2, CLN8, ARHGEF10, CSMD1, MCPH1* and *DEFB1* | NA | Large del | | P | | Het | | AD | | \*1619636 | |
| 48 | S-4289 | Joubert syndromeo | chr15q11.2-q13.1 | chr15:23810930-?\_28544662+?del | *MKRN3, MAGEL2, NDN, PWRN1, NPAP1, SNURF, SNRPN, SNORD116-1, SNORD116-10, SNORD115-1, UBE3A, ATP10A, GABRB3, GABRA5, GABRG3, OCA2* and *HERC2* | NA | Large del | | LP | | Het | | AD | | \*18840528 | |
| 49 | S-177 | Leigh syndromeo | chr15q24.1-q25.1 | chr15:74630313-?\_78927929+?del | *CYP11A1, CYP1A1, CYP1A2, MPI, NEIL1, ETFA, PSTPIP1, CIB2, DNAJA4, CHRNA5, CHRNA3* and *CHRNB4* | NA | Large del | | LP | | Het | | AD | | \*17360722 | |
| 50 | S-3969 | Sotos syndromeo | chr16p13.11-p12.3 | chr16:15758636-?\_17564653+?del | *NDE1, MYH11, ABCC1, ABCC6* and *XYLT1* | NA | Large del | | P | | Het | | AR | | \*23704059 | |
| 51 | S-3888 | Microcephaly, ID | chr17p11.2 | chr17:16842861-?\_19871774+?del | *TNFRSF13B, FLCN, PEMT, RAI1, SREBF1, ATPAF2, MYO15A, LLGL1, SHMT1, B9D1, SLC47A1, ALDH3A2, SLC47A2* and *AKAP10* | NA | Large del | | P | | Het | | AD | | \*27075776 | |
| 52 | S-3209 | Neuropathy | chr17p12 | chr17:14110127-?\_15164044+?del | *COX10, PMP22* | NA | Large del | | P | | Het | | AD | | \*9285799 | |
| 53 | S-6580 | Neuropathy | chr17p12 | chr17:14110127-?\_15162510+?dup | *COX10, PMP22* | NA | Large dup | | P | | Het | | AD | | \*23224996 | |
| 54 | S-6503 | Neuropathy | chr17p12 | chr17:14110127-?\_15162510+?dup | *COX10, PMP22* | NA | Large dup | | P | | Het | | AD | | \*23224996 | |
| 55 | S-5648 | Developmental delay | chr17p13.3-p13.2 | chr17:2568666-?\_3422144+?del | *PAFAH1B1, ASPA* and *TRPV3* | NA | Large del | | P | | Het | | AD | | \*20452996 | |
| 56 | S-3191 | Developmental delay | chr18q12.1-q12.2 | chr18:28574436-?\_33848648+?del | *DSC3, DSC2, DSG1, DSG4, DSG2, TTR, MEP1B, DTNA, ZNF24, ELP2* and *MOCOS* | NA | Large del | | P | | Het | | AD | | \*23727450 | |
| 57 | S-6017 | Pitt-Hopkins syndromei | chr18q21.2 | chr18:50278424-?\_53303128+?del | *DCC* and *TCF4* | NA | Large del | | P | | Het | | AD | | \*23165966 | |
| 58 | S-3206 | Developmental delay | chr19p13.3 | chr19:3586493-?\_4517716+?del | *GIPC3, TBXA2R, PIP5K1C, RAX2, ATCAY, MAP2K2, CREB3L3, SH3GL1* and *PLIN4* | NA | Large del | | P | | Het | | AD | | \*27239227 | |
| 59 | S-338 | Sotos syndromeo | chr22q13.31-q13.33 | chr22:44221881-?\_51169740+?del | *SULT4A1, PNPLA3, UPK3A, SMC1B, FBLN1, ATXN10, PPARA, TRMU, CELSR1, BRD1, ALG12, IL17REL, MLC1, TUBGCP6, SCO2, TYMP, CPT1B, CHKB, ARSA* and *SHANK3* | NA | Large del | | P | | Het | | AD | | \*12920066 | |
| 60 | S-4467 | Epilepsy | chrXp22.13 | chrX:18525217-?\_18690188+?del | *CDKL5* and *RS1* | NA | Large del | | P | | Het | | XL | | \*21293276 | |
| 61 | S-4223 | Developmental delay | chrXq22.1-q22.2 | chrX:102471082?\_103268232+?dup | *BEX4, PLP1* and *H2BFWT* | NA | Large dup | | LP | | Hemi | | XL | | \*24646727 | |

**Abbreviations:**

AD-Autosomal dominant; ASD-Autism spectrum disorders; AR-Autosomal recessive; CdLS-Cornelia de Lange syndrome; CS-Clinical Significance; Del-Deletion; Dup-Duplication; Ex-Exon; Hemi-Hemizygous; Het-Heterozygous; Hom-Homozygous; ID-Intellectual disability; Int-Intron; LP-Likely Pathogenic; MOI-Mode of Inheritance; MS-Missense; NS-Nonsense; PMD-Pelizaeus-Merzbacher disease; P-Pathogenic; XL-X-linked.

Δ: ClinVar database ID, \*: Reference from PubMed database (PMID) with overlapping deletion/duplication (exact breakpoints not known)

For the disease conditions in clinical subtypes, following abbreviation have been used

i: Intellectual disability; md: muscular dystrophy; o: others

**Table S5: List of ‘variant of uncertain significance with probable damaging effect’ (VUSD) identified in the study.**

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Sl. No** | **Sample ID** | **Disease condition** | **Gene/**  **Genomic**  **region** | **Ex/**  **Int** | **Nucleotide/chr. coordinate** | **Protein** | **VT** | **Zygosity** | **MOI** | **Reference** | **Public Databases** | | | ***In Silico* Predictors** | | | | | | | | | |
| **dbSNP** | **ClinVar** | **ExAC$** | **FA** | **LRT** | **MT** | **MA** | **PP** | **SIFT** | **NN** | **SP** | **HS** | **Pro** |
| 1 | S-4432 | Spasticity | *ALDH18A1* | Ex 4 | c.382C>T | p.Arg128Cys | MS | Hom | AR | In this study | NA | NA | NA | B | D | D | D | D | D | NA | NA | NA | NA |
| 2 | S-5097 | Epilepsy | *ALDH7A1* | Ex 17 | c.1556G>A | p.Arg519Lys | MS | Hom | AR | \*23925287 | rs561343926 | NA | <0.01 | B | D | D | D | D | D | NA | NA | NA | NA |
| 3 | S-3889 | Epilepsy | *ALDH7A1* | Ex 18 | c.1598C>A | p.Ala533Asp | MS | Hom | AR | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 4 | S-5300 | Epilepsy | *ARHGEF9* | Ex 5 | c.754C>T | p.Gln252Ter | MS | Hemi | XL | In this study | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA |
| 5 | S-4314 | Leukodystrophy | *ARSA* | Ex 1 | c.173G>A | p.Gly58Glu | MS | Hom | AR | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 6 | S-1845 | Leukodystrophy | *ARSA* | Ex 3 | c.577C>T | p.Pro193Ser | MS | Hom | AR | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 7 | S-4021 | Epilepsy | *ATRX* | Ex 14 | c.4317G>A | p.Lys1439= | SS | Hemi | XL | \*8968741 | NA | NA | NA | NA | NA | NA | NA | NA | NA | D | D | D | NA |
| 8 | S-2895 | Joubert syndrome | *C5orf42* | Ex 26 | c.4643A>G | p.Asp1548Gly | MS | Hom | AR | \*25407461 | rs759649053 | RCV00  0605589.1 | NA | B | D | D | B | D | D | NA | NA | NA | NA |
| 9 | R-0691 | Epilepsy, ID | *CLN6* | Ex 7 | c.679G>A | p.Glu227Lys | MS | Hom | AR | NA | rs746753722 | RCV00  0625854.1 | <0.01 | D | D | D | D | D | D | NA | NA | NA | NA |
| 10 | S-4869 | Epilepsy | *CLN8* | Ex 2 | c.208C>T | p.Arg70Cys | MS | Hom | AR | NA | rs765097897 | RCV00  0201947.1 | <0.01 | D | B | D | D | D | D | NA | NA | NA | NA |
| 11 | S-3540 | Muscular dystrophy | *CRADD* | Ex 2 | c.285dupC | p.Asp96ArgfsTer6 | Indel | Hom | AR | NA | NA | RCV00  0502270.1 | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA |
| 12 | S-277 | Neuropathy | *GAN* | Ex 2 | c.278G>A | p.Gly93Glu | MS | Hom | AR | In this study | NA | NA | NA | B | D | D | D | D | B | NA | NA | NA | NA |
| 13 | S-1429 | Dystonia | *GCH1* | Ex 3 | c.458A>T | p.His153Leu | MS | Het | AD | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 14 | S-5772 | Myopathy | *GNE* | Ex 7 | c.1174T>C | p.Tyr392His | MS | Het | AR | NA | rs1192630467 | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| Ex 12 | c.2179G>A | p.Val727Met | MS | Het | AR | \*24005727 | rs121908627 | RCV00  0627754.1 | <0.01 | D | D | D | D | D | D | NA | NA | NA | NA |
| 15 | S-2623 | Ataxia | *ITPR1* | Ex 45 | c.6136G>C | p.Glu2046Gln | MS | Het | AD/  AR | \*27108798 | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 16 | S-345 | SeSAME  syndrome | *KCNJ10* | Ex 2 | c.230\_231delinsAA | p.Gly77Glu | MS | Hom | AR | In this study | NA | NA | NA | D | B | D | D | D | B | NA | NA | NA | NA |
| 17 | S-5207 | Epilepsy | *KCNQ2* | Ex 4 | c.601C>G | p.Arg201Gly | MS | Het | AD | In this study | NA | NA | NA | D | B | D | D | D | D | NA | NA | NA | NA |
| 18 | S-4383 | Hypotonia | *LMNA* | Ex 1 | c.98\_100delAGG | p.Glu33del | Indel | Het | AD | In this study | NA | NA | NA | NA | NA | D | NA | NA | NA | NA | NA | NA | D |
| 19 | R-0563 | Epilepsy | *MCCC2* | Int 3 | c.281+4C>T | NA | SS | Het | AR | In this study | rs758998339 | NA | <0.01 | NA | NA | NA | NA | NA | NA | D | NA | D | NA |
| Ex 9 | c.845A>G | p.His282Arg | MS | Het | AR | \*22642865 | NA | NA | NA | D | D | D | D | D | D | D | NA | NA | NA |
| 20 | S-4506 | Neuropathy | *MFN2* | Ex 4 | c.262A>T | p.Ile88Phe | MS | Het | AD/AR | \*26392352 | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 21 | S-5119 | Leigh syndrome | *NDUFA1* | Ex 2 | c.109A>G | p.Arg37Gly | MS | Hemi | XL | In this study | NA | NA | NA | D | D | D | B | D | D | NA | NA | NA | NA |
| 22 | S-3715 | Sotos syndrome | *NSD1* | Ex 20 | c.6063T>G | p.His2021Gln | MS | Het | AD | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 23 | S-3450 | Neuropathy | *NTRK1* | Ex 8 | c.1030G>C | p.Gly344Arg | MS | Het | AR | In this study | NA | NA | NA | B | B | D | D | D | D | NA | NA | NA | NA |
| Ex 16 | c.2294G>A | p.Arg765His | MS | Het | AR | NA | rs780724170 | NA | <0.01 | D | D | D | D | D | D | NA | NA | NA | NA |
| 24 | S-760 | Epilepsy | *PC* | Ex 8 | c.784G>A | p.Glu262Lys | MS | Hom | AR | NA | rs1324553572 | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 25 | S-2433 | NBIA | *PLA2G6* | Ex 13 | c.1804T>C | p.Tyr602His | MS | Het | AR | In this study | NA | NA | NA | B | D | D | D | D | D | NA | NA | NA | NA |
| Ex 14 | c.1894C>T | p.Arg632Trp | MS | Het | AR | \*16783378 | rs121908683 | RCV00  0006576.2 | <0.01 | B | D | D | D | D | D | NA | NA | NA | NA |
| 26 | S-3240 | Neuropathy | *PMP22* | Ex 4 | c.239T>C | p.Leu80Pro | MS | Het | AD | \*9055797 | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 27 | S-2369 | Muscular dystrophy | *POMT1* | Ex 18 | c.1868G>C | p.Arg623Thr | MS | Hom | AR | \*17878207 | rs779321311 | NA | <0.01 | D | B | D | D | D | D | NA | NA | NA | NA |
| 28 | S-3551 | Ataxia,  Dystonia | *PSEN1* | Ex 5 | c.404A>G | p.Asn135Ser | MS | Het | AD | \*15776278 | rs63751278 | RCV00  0084303.1 | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 29 | S-2573 | AGS | *RNASEH2A* | Ex 2 | c.158C>G | p.Pro53Arg | MS | Hom | AR | \*28387595 | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 30 | S-2211 | Hypotonia | *RYR1* | Ex 33 | c.4934G>A | p.Arg1645Gln | MS | Het | AD/AR | \*18253926 | rs193922778 | RCV00  0119627.1 | NA | D | B | D | D | D | D | D | D | NA | NA |
| Int 68 | c.10348-6C>G | NA | SS | Het | AD/AR | \*18253926 | rs193922837 | RCV00  0119410.1 | <0.01 | NA | NA | NA | NA | NA | NA | D | D | B | NA |
| 31 | S-2854 | Spasticity | *SACS* | Ex 8 | c.1607C>T | p.Pro536Leu | MS | Hom | AR | \*20876471 | rs1440541889 | RCV00  0523535.1 | NA | B | D | D | D | D | D | NA | NA | NA | NA |
| 32 | R-0563 | Epilepsy | *SCN1A* | Ex 6 | c.746A>G | p.Asp249Gly | MS | Het | AD | NA | rs762927460 | NA | <0.01 | D | D | D | D | D | D | NA | NA | NA | NA |
| 33 | S-3323 | Epilepsy | *SCN1A* | Ex 8 | c.1145A>G | p.Asp382Gly | MS | Het | AD | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 34 | S-1140 | Epilepsy | *SCN1A* | Ex 16 | c.2972T>G | p.Leu991Arg | MS | Het | AD | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 35 | S-4617 | Epilepsy | *SCN1A* | Ex 23 | c.4339G>T | p.Val1447Leu | MS | Het | AD | In this study | NA | NA | NA | D | B | D | D | D | B | D | D | NA | NA |
| 36 | S-334 | Epilepsy, ID | *SCN2A* | Ex 15 | c.2558G>A | p.Arg853Gln | MS | Het | AD | \*23935176 | rs794727152 | RCV00  0197677 | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 37 | S-4317 | Epilepsy,  Ataxia | *SCN9A* | Ex 4 | c.446C>G | p.Pro149Arg | MS | Het | AD | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 38 | S-4510 | Ataxia | *SETX* | Ex 15 | c.6029A>G | p.Asn2010Ser | MS | Hom | AR | \*19696032 | rs759806045 | NA | <0.01 | D | D | D | D | D | D | NA | NA | NA | NA |
| 39 | S-5518 | Muscular dystrophy | *SGCA* | Ex 3 | c.241C>T | p.Arg81Cys | MS | Hom | AR | \*17994539 | rs398123098 | RCV00  0077938 | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 40 | S-4320 | Neuropathy | *SIGMAR1* | Ex 4 | c.446G>A | p.Gly149Glu | MS | Hom | AR | NA | rs546522200 | NA | NA | D | D | D | D | D | D | D | NA | NA | NA |
| 41 | S-3676 | Neuropathy | *SIGMAR1* | Ex 4 | c.446G>A | p.Gly149Glu | MS | Hom | AR | NA | rs546522200 | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 42 | S-5011 | ALS | *SOD1* | Ex 5 | c.436G>A | p.Ala146Thr | MS | Het | AD | \*7496169 | rs121912447 | RCV00  0015892.26 | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 43 | S-3365 | ASD | *TCF4* | Ex 12 | c.990G>A | p.Ser330= | SS | Het | AD | \*25693842 | rs587784469 | RCV00  0147730.1 | NA | NA | NA | NA | NA | NA | NA | D | B | D | NA |
| 44 | R-0490 | Neuropathy | *TTR* | Ex 3 | c.218G>C | p.Gly73Ala | MS | Het | AD | \*16971399 | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 45 | S-200 | Epilepsy | *CHRNA7* | Ex 1-4 dup | c.(?-12)\_(350+1\_?) dup | NA | Large dup | Inc | AD | In this study | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA |
| 46 | S-5954 | Epilepsy,  Ataxia | *KCTD7* | Ex 3-4 dup | c.(314+1\_3151)\_(\*3997\_?)dup | NA | Large dup | Hom | AR | In this study | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA |
| 47 | S-4263 | Developmental delay | chr4q21.  21-q21.23 | NA | chr4:80828583?\_86491874+?del | NA | Large del | Het | AD | In this study | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA |

**Abbreviations:**

AD-Autosomal dominant; AGS-Aicardi-Goutieres syndrome; ALS-Amyotrophic lateral sclerosis; AR-Autosomal recessive; ASD-Autism spectrum disorders; B-Benign; CS-Clinical significance; D-Damaging; Del-Deletion; Dup-Duplication; ExAC-Exome Aggregation Consortium; FA-FATHMM (Functional analysis through Hidden Markov models); Hemi-Hemizygous; Het-Heterozygous; Hom-Homozygous; HSF-Human splicing finder; Inc-Inconclusive; ID-Intellectual disability; Indel- Small deletion/duplication/insertion or insertion/deletion ; Int-Intron; LRT-Likelihood-ratio test; MA-Mutation assessor; MOI-Mode of inheritance; MS-Missense; MT-Mutation taster ; NBIA-Neurodegeneration with brain iron accumulation; NN-NNSPLICE; NS-Nonsense; PP-PolyPhen; Pro-Provean; SIFT-Sorting intolerant from tolerant ; SS-Splice site; VT-Variant type; XL-X-linked

\*: Reference from PubMed database (PMID), \*\*: Reported in genome Aggregation Database (gnomAD), $: ExAC allelic frequency in percentage

**Table S6: List of ‘variant of uncertain significance’ (VUS) identified in the study.**

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **SL No** | **Sample**  **ID** | **Disease**  **condition** | **Gene** | **Ex/Int** | **Nucleotide** | **Protein** | **VT** | **Zygosity** | **MOI** | **Reference** | **Public Databases** | | | ***In silico* Predictors** | | | | | | | | | |
| **dbSNP** | **ClinVar** | **ExAC$** | **FA** | **LRT** | **MT** | **MA** | **PP** | **SIFT** | **NN** | **SP** | **HSF** | **Pro** |
| 1 | S-1334 | Myopathy, Neuropathy | *AARS* | Int 12 | c.1672-4T>A | NA | SS | Het | AD | NA | rs187509039 | RCV000  197414.7 | <0.01 | NA | NA | NA | NA | NA | NA | NA | NA | B | NA |
| 2 | S-3974 | Poor feeding, reduced activity | *ABCC8* | Ex 12 | c.1785T>A | p.Ser595Arg | MS | Het | AD/AR | NA | rs983621605 | NA | NA | D | NA | D | B | B | B | NA | NA | NA | NA |
| 3 | S-5811 | Epilepsy | *ACADS* | Ex 6 | c.778G>T | p.Gly260Cys | MS | Hom | AR | NA | rs759856061 | NA | <0.01 | D | NA | D | D | D | D | NA | NA | NA | NA |
| 4 | S-2740 | ASD, ID | *ACSL4* | Ex 11 | c.1181A>G | p.Asn394Ser | MS | Hemi | XL | In this study | NA | NA | NA | B | D | D | B | B | D | NA | NA | NA | NA |
| 5 | S-1452 | Myopathy | *ACTA1* | Ex 5 | c.630C>G | p.Ile210Met | MS | Hom | AR | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 6 | S-5766 | Myopathy | *ACTA1* | Ex 7 | c.1106C>A | p.Pro369His | MS | Het | AD/AR | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 7 | S-4896 | Leukodystrophy | *ADAR* | Ex 2 | c.524T>A | p.Val175Asp | MS | Het | AD | In this study | NA | NA | NA | B | B | B | B | D | D | NA | NA | NA | NA |
| 8 | S-1710 | Epilepsy, ASD | *ADSL* | Ex 5 | c.502G>A | p.Val168Ile | MS | Hom | AR | NA | rs1385675650 | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 9 | S-2544 | Ataxia, Dystonia, Neuropathy | *AFG3L2* | Ex 8 | c.874G>A | p.Gly292Arg | MS | Het | AD | NA | rs532179680 | NA | <0.01 | B | D | D | D | B | D | NA | NA | NA | NA |
| 10 | S-2397 | Muscle weakness | *AGRN* | Ex 23 | c.4061C>A | p.Thr1354Asn | MS | Het | AR | NA | rs766470739 | NA | <0.01 | D | B | D | B | D | B | NA | NA | NA | NA |
| Ex 34 | c.5866G>A | p.Val1956Met | MS | Het | AR | NA | rs773243053 | NA | <0.01 | B | B | B | B | D | B | NA | NA | NA | NA |
| 11 | S-4871 | Joubert syndrome | *AHI1* | Ex 6 | c.370A>G | p.Lys124Glu | MS | Hom | AR | In this study | NA | NA | NA | B | B | D | B | B | B | NA | NA | NA | NA |
| 12 | S-4176 | Joubert syndrome | *AHI1* | Ex 14 | c.1979T>A | p.Leu660His | MS | Hom | AR | NA | rs772560701 | NA | <0.01 | B | D | D | D | D | D | NA | NA | NA | NA |
| 13 | S-2634 | Atrophy of corpus callosum | *ALDH18A1* | Ex 18 | c.2351A>G | p.His784Arg | MS | Hom | AR | NA | rs765966176 | NA | <0.01 | D | D | D | D | D | D | NA | NA | NA | NA |
| 14 | S-3202 | Epilepsy | *ALDH5A1* | Ex 3 | c.608C>T | p.Pro203Leu | MS | Hom | AR | NA | rs906284769 | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 15 | S-3650 | Epilepsy | *ALDH7A1* | Ex 18 | c.1598C>A | p.Ala533Asp | MS | Hom | AR | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 16 | S-3466 | Muscle weakness | *ALG2* | Ex 1 | c.338T>A | p.Val113Glu | MS | Hom | AR | In this study | NA | NA | NA | B | B | D | D | D | D | NA | NA | NA | NA |
| 17 | S-5889 | ALS | *ALS2* | Ex 20 | c.3326T>C | p.Met1109Thr | MS | Het | AR | NA | rs372595317 | NA | <0.01 | B | D | D | D | D | B | NA | NA | NA | NA |
| Ex 20 | c.3345C>T | p.Tyr1115= | SS | Het | AR | NA | rs557709223 | NA | <0.01 | NA | NA | NA | NA | NA | NA | D | D | D | NA |
| 18 | S-424 | Ataxia | *ANO10* | Ex 8 | c.1232A>G | p.Asn411Ser | MS | Hom | AR | NA | rs762428946 | NA | <0.01 | B | D | D | D | D | D | NA | NA | NA | NA |
| 19 | S-4755 | Microcephaly, Leukodystrophy | *AP4B1* | Ex 11 | c.2096T>C | p.Leu699Ser | MS | Hom | AR | In this study | NA | NA | NA | B | B | D | B | D | D | NA | NA | NA | NA |
| 20 | S-2888 | Brain malformations | *ARFGEF2* | Int 37 | c.5064-2A>G | NA | SS | Hom | AR | In this study | NA | NA | NA | NA | NA | NA | NA | NA | NA | D | NA | D | NA |
| 21 | S-2954 | Neuropathy | *ARHGEF10* | Ex 4 | c.218A>C | p.Glu73Ala | MS | Het | AD | In this study | NA | NA | NA | B | B | B | B | B | D | NA | NA | NA | NA |
| 22 | S-592 | Neuropathy | *ARHGEF10* | Ex 28 | c.3439G>A | p.Gly1147Arg | MS | Het | AD | NA | rs1212009927 | NA | NA | B | D | D | B | D | D | NA | NA | NA | NA |
| 23 | S-3146 | Epilepsy, Microcephaly | *ARID1A* | Ex 18 | c.4946C>G | p.Thr1649Arg | MS | Het | AD | In this study | NA | NA | NA | B | D | D | D | D | D | NA | NA | NA | NA |
| 24 | S-4776 | ID | *ARID1B* | Ex 19 | c.4969A>G | p.Thr1657Ala | MS | Het | AD | In this study | NA | NA | NA | B | D | D | B | D | B | NA | NA | NA | NA |
| 25 | S-1850 | Leukodystrophy | *ARSA* | Ex 2 | c.385G>A | p.Gly129Arg | MS | Hom | AR | NA | rs753872402 | NA | <0.01 | D | D | D | D | D | D | NA | NA | NA | NA |
| 26 | S-1020 | Leukodystrophy, Neuropathy | *ARSA* | Ex 7 | c.1130\_1132delTCT | p.Phe377del | Indel | Hom | AR | NA | rs1021236473 | NA | NA | NA | NA | D | NA | NA | NA | NA | NA | NA | D |
| 27 | S-1845 | Leukodystrophy | *ARSA* | Ex 8 | c.1400A>G | p.Gln467Arg | MS | Hom | AR | In this study | NA | NA | NA | D | B | D | B | B | B | NA | NA | NA | NA |
| 28 | S-2334 | Epilepsy | *ASNS* | Ex 3 | c.146G>A | p.Arg49Gln | MS | Hom | AR | NA | rs769236847 | RCV000  432896.1 | <0.01 | B | D | D | D | D | D | D | NA | NA | NA |
| 29 | S-1003 | Ataxia | *ATP13A2* | Ex 16 | c.1556C>T | p.Thr519Ile | MS | Het | AR | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| Ex 22 | c.2440G>A | p.Val814Met | MS | Het | AR | NA | NA | NA | <0.01 | D | B | B | B | B | B | NA | NA | NA | NA |
| 30 | S-3679 | Obsessive disease, auditory hallucination | *ATP8A2* | Ex 19 | c.1711A>G | p.Ser571Gly | MS | Het | AR | NA | rs555217046 | NA | <0.01 | B | D | D | D | D | D | NA | NA | NA | NA |
| Ex26 | c.2431C>T | p.Arg811Trp | MS | Het | AR | NA | rs560280973 | NA | <0.01 | D | B | D | B | D | B | NA | NA | NA | NA |
| 31 | S-2252 | ASD | *ATRX* | Ex 9 | c.2095G>C | p.Asp699His | MS | Hemi | XL | In this study | NA | NA | NA | D | B | D | B | D | D | NA | NA | NA | NA |
| 32 | S-2432 | Developmental delay | *ATRX* | Ex 32 | c.6914A>G | p.Gln2305Arg | MS | Hemi | XL | In this study | NA | NA | NA | D | B | D | B | D | B | NA | NA | NA | NA |
| 33 | S-5618 | Dystonia | *C19orf12* | Ex 2 | c.77T>C | p.Leu26Pro | MS | Hom | AR | NA | rs773824674 | NA | <0.01 | D | B | D | B | D | D | NA | NA | NA | NA |
| 34 | S-2918 | Epilepsy | *CACNA1A* | Ex 17 | c.2137G>C | p.Ala713Pro | MS | Het | AD | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 35 | S-834 | Epilepsy | *CACNA1A* | Ex 35 | c.5398T>A | p.Phe1800Ile | MS | Het | AD | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 36 | S-1300 | Developmental delay | *CACNA1G* | Ex 8 | c.1633\_1634delinsTT | p.Ala545Phe | Indel | Het | AD | In this study | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | B |
| 37 | S-5907 | Epilepsy | *CACNA1G* | Ex 17 | c.3386G>A | p.Arg1129Gln | MS | Het | AD | NA | rs1180552980 | NA | NA | D | B | D | D | D | D | NA | NA | NA | NA |
| 38 | S-5549 | Epilepsy | *CACNA1H* | Ex 5 | c.567C>G | p.Asp189Glu | MS | Het | AD | NA | rs1175587489 | NA | NA | D | B | B | B | D | B | NA | NA | NA | NA |
| 39 | S-4738 | Epilepsy | *CACNA1H* | Ex9 | c.1528C>A | p.His510Asn | MS | Het | AD | In this study | NA | NA | NA | D | B | B | B | B | B | NA | NA | NA | NA |
| 40 | S-4738 | Epilepsy | *CACNA1H* | Ex 10 | c.2258G>T | p.Gly753Val | MS | Het | AD | NA | rs767816214 | NA | <0.01 | D | B | B | B | B | B | NA | NA | NA | NA |
| 41 | S-5457 | Epilepsy | *CACNA1H* | Ex 15 | c.3067G>A | p.Asp1023Asn | MS | Het | AD | NA | rs977157885 | NA | NA | D | B | D | D | D | D | NA | NA | NA | NA |
| 42 | S-2377 | Abnormal head and limb movements | *CACNA1H* | Ex 16 | c.3313C>T | p.Arg1105Cys | MS | Het | AD | NA | rs370833327 | NA | <0.01 | D | B | B | D | D | D | NA | NA | NA | NA |
| 43 | S-487 | Epilepsy | *CACNA1H* | Ex 20 | c.4018G>A | p.Val1340Met | MS | Het | AD | NA | rs747265858 | NA | <0.01 | D | D | B | D | D | D | NA | NA | NA | NA |
| 44 | S-4256 | Epilepsy, Microcephaly | *CACNA1H* | Ex 27 | c.4907T>C | p.Met1636Thr | MS | Het | AD | NA | rs577258290 | NA | <0.01 | D | D | D | B | B | D | NA | NA | NA | NA |
| 45 | S-2989 | Dystonia | *CASK* | Ex 27 | c.2675A>G | p.Asn892Ser | MS | Het | XL | NA | rs762230615 | NA | <0.01 | B | D | D | B | D | B | NA | NA | NA | NA |
| 46 | S-3407 | Muscular dystrophy | *CAV3* | Ex 2 | c.244G>T | p.Val82Phe | MS | Het | AD | In this study | NA | NA | NA | D | B | D | B | D | D | NA | NA | NA | NA |
| 47 | S-452 | Developmental delay | *CC2D2A* | Ex 15 | c.1484G>A | p.Arg495His | MS | Het | AR | NA | rs373906628 | RCV000  478789 | <0.01 | D | D | D | D | D | B | NA | NA | NA | NA |
| Ex 15 | c.1598T>C | p.Val533Ala | MS | Het | AR | NA | rs777351655 | RCV000  299384 | <0.01 | B | B | D | D | B | B | NA | NA | NA | NA |
| 48 | S-4871 | Joubert syndrome | *CC2D2A* | Ex 35 | c.4431T>G | p.Ser1477Arg | MS | Hom | AR | In this study | NA | NA | NA | B | D | D | D | D | D | NA | NA | NA | NA |
| 49 | S-5225 | ID | *CDH15* | Ex 5 | c.613G>C | p.Asp205His | MS | Het | AD | NA | rs138870237 | NA | NA | B | B | D | B | D | B | B | NA | NA | NA |
| 50 | S-2964 | Epilepsy | *CDKL5* | Ex 8 | c.517G>C | p.Ala173Pro | MS | Het | XL | In this study | NA | NA | NA | B | D | D | B | D | D | NA | NA | NA | NA |
| 51 | S-3975 | Epilepsy | *CDKL5* | Ex 12 | c.1837A>C | p.Met613Leu | MS | Hemi | XL | In this study | NA | NA | NA | B | D | D | B | B | D | NA | NA | NA | NA |
| 52 | S-5296 | Epilepsy | *CDKL5* | Ex 15 | c.2269G>A | p.Asp757Asn | MS | Hemi | XL | NA | rs758383464 | NA | <0.01 | B | D | D | B | D | D | NA | NA | NA | NA |
| 53 | S-2427 | Epilepsy, ASD | *CDKL5* | Ex 20 | c.2896G>A | p.Val966Ile | MS | Hemi | XL | NA | rs747799506 | RCV000  194778.1 | <0.01 | B | B | B | B | B | D | NA | NA | NA | NA |
| 54 | S-2466 | Epilepsy | *CDKL5* | Ex 20 | c.2896G>A | p.Val966Ile | MS | Hemi | XL | NA | rs747799506 | RCV000  194778.1 | <0.01 | B | B | B | B | B | D | NA | NA | NA | NA |
| 55 | S-5453 | Joubert syndrome | *CEP290* | Ex 17 | c.1711G>A | p.Gly571Arg | MS | Hom | AR | In this study | NA | NA | NA | B | B | D | B | D | B | D | NA | NA | NA |
| 56 | S-5774 | Meckel-Gruber syndrome | *CEP290* | Ex 17 | c.1711G>A | p.Gly571Arg | MS | Hom | AR | In this study | NA | NA | NA | B | B | D | B | B | B | D | NA | D | NA |
| 57 | S-5347 | Joubert syndrome | *CEP290* | Ex 17 | c.1711G>A | p.Gly571Arg | MS | Hom | AR | In this study | NA | NA | NA | B | B | D | B | D | B | D | NA | NA | NA |
| 58 | S-4228 | Epilepsy, ID | *CHD2* | Ex 14 | c.1709G>A | p.Ser570Asn | MS | Het | AD | In this study | NA | NA | NA | D | B | D | B | D | D | NA | NA | NA | NA |
| 59 | S-3306 | Developmental delay | *CHD7* | Ex 13 | c.3295T>C | p.Trp1099Arg | MS | Het | AD | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 60 | S-1284 | Epilepsy | *CHRNA4* | Ex 5 | c.1079G>A | p.Arg360Gln | MS | Het | AD | NA | rs1379329180 | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 61 | S-1300 | Developmental delay | *CHRNB1* | Ex 6 | c.480C>A | p.Phe160Leu | MS | Het | AD | In this study | NA | NA | NA | B | D | D | D | D | D | NA | NA | NA | NA |
| 62 | S-4247 | Myasthenic syndrome | *CHRNE* | Ex 8 | c.893C>G | p.Ser298Cys | MS | Het | AD | In this study | NA | NA | NA | D | D | D | D | D | B | NA | NA | NA | NA |
| 63 | S-3540 | Muscular dystrophy | *CLCN1* | Ex 5 | c.677G>A | p.Gly226Asp | MS | Het | AD/AR | NA | rs1304529237 | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 64 | S-225 | Epilepsy | *CLCN2* | Ex 3 | c.246C>G | p.Phe82Leu | MS | Het | AD | \*21703448 | rs140463309 | RCV000  201829.1 | <0.01 | D | D | D | B | D | B | NA | NA | NA | NA |
| 65 | S-3146 | Epilepsy, Microcephaly | *CLN8* | Ex 2 | c.303C>G | p.Asn101Lys | MS | Hom | AR | NA | rs755831055 | NA | <0.01 | D | B | D | B | B | B | NA | NA | NA | NA |
| 66 | S-4685 | Epilepsy, Microcephaly, ID | *CNNM2* | Ex 1 | c.1372A>G | p.Met458Val | MS | Het | AD/AR | NA | rs1359308930 | NA | NA | B | D | D | D | D | D | NA | NA | NA | NA |
| 67 | S-2937 | Developmental delay | *CNTNAP2* | Ex 8 | c.1157T>C | p.Val386Ala | MS | Het | AR | NA | NA | NA | NA | B | B | D | B | B | D | NA | NA | NA | NA |
| Ex 23 | c.3781T>G | p.Ser1261Ala | MS | Het | AR | NA | rs754580386 | RCV000  187239.1 | <0.01 | B | D | D | B | B | B | NA | NA | NA | NA |
| 68 | S-3155 | Leukodystrophy | *COL11A2* | Ex 12 | c.1334C>T | p.Pro445Leu | MS | Het | AD/AR | In this study | NA | NA | NA | D | D | D | B | D | B | NA | NA | NA | NA |
| 69 | S-929 | Muscular dystrophy | *COL12A1* | Ex 52 | c.7951G>T | p.Val2651Phe | MS | Het | AD | In this study | NA | NA | NA | B | D | D | D | B | D | D | D | NA | NA |
| 70 | R-0517 | Leukodystrophy | *COL4A1* | Ex 23 | c.1426C>T | p.Arg476Trp | MS | Het | AD | NA | rs369960952 | NA | <0.01 | D | B | D | D | D | D | NA | NA | NA | NA |
| 71 | R-0100 | Microcephaly | *COL4A1* | Ex 33 | c.2705C>T | p.Pro902Leu | MS | Het | AD | NA | rs146134172 | NA | NA | B | D | D | B | D | B | NA | NA | NA | NA |
| 72 | S-498 | Microcephaly, Spasticity, Dystonia | *COL4A1* | Ex 42 | c.3704A>G | p.Lys1235Arg | MS | Het | AD | NA | rs781655700 | NA | <0.01 | D | D | D | B | D | B | NA | NA | NA | NA |
| 73 | S-2298 | Ataxia | *COL6A1* | Ex 2 | c.225C>T | p.Asp75= | SS | Het | AD/AR | In this study | NA | NA | NA | NA | NA | NA | NA | NA | NA | D | D | D | NA |
| 74 | S-4133 | Myopathy | *COL6A1* | Ex 3 | c.262G>A | p.Ala88Thr | MS | Het | AD/AR | NA | rs540429266 | NA | <0.01 | D | D | D | B | D | B | NA | NA | NA | NA |
| 75 | S-0297 | Muscular dystrophy | *COL6A2* | Ex 18 | c.1489C>T | p.Pro497Ser | MS | Het | AD/AR | In this study | NA | NA | NA | D | B | D | D | B | D | NA | NA | NA | NA |
| 76 | S-4960 | Myopathy | *COL6A2* | Ex 26 | c.2065G>A | p.Glu689Lys | MS | Het | AD/AR | NA | rs560146338 | NA | <0.01 | B | D | D | B | D | B | NA | NA | NA | NA |
| 77 | S-4383 | Hypotonia | *COL6A2* | Ex 26 | c.2395G>A | p.Asp799Asn | MS | Het | AD/AR | NA | rs372936386 | NA | <0.01 | B | D | D | D | D | B | NA | NA | NA | NA |
| 78 | S-3614 | Muscular dystrophy | *COL6A3* | Ex 9 | c.3700G>A | p.Val1234Met | MS | Het | AD/AR | NA | rs747082651 | NA | <0.01 | D | B | D | D | D | D | NA | NA | NA | NA |
| 79 | S-2208 | Muscular dystrophy | *COL6A3* | Ex 17 | c.6224C>A | p.Pro2075Gln | MS | Het | AD/AR | In this study | NA | NA | NA | D | D | D | B | D | B | NA | NA | NA | NA |
| 80 | S-4466 | Muscular dystrophy, Myopathy | *COL6A3* | Ex 36 | c.7375C>T | p.Arg2459Trp | MS | Het | AD/AR | NA | rs371066956 | RCV000  653572.1 | <0.01 | B | D | D | D | D | D | NA | NA | NA | NA |
| 81 | S-3730 | Epilepsy, ASD | *CPA6* | Ex 11 | c.1283T>C | p.Ile428Thr | MS | Het | AD/AR | NA | rs759260101 | NA | <0.01 | B | D | D | D | D | D | NA | NA | NA | NA |
| 82 | S-2264 | Microcephaly, ID | *CTNNB1* | Ex 11 | c.1690G>C | p.Val564Leu | MS | Het | AD | In this study | NA | NA | NA | B | D | D | D | D | B | NA | NA | NA | NA |
| 83 | S-2834 | ID | *CUL4B* | Ex 21 | c.2507C>T | p.Ala836Val | MS | Hemi | XL | In this study | NA | NA | NA | B | D | D | B | B | B | NA | NA | NA | NA |
| 84 | S-5901 | Epilepsy | *D2HGDH* | Ex 7 | c.995A>G | p.Gln332Arg | MS | Het | AR | NA | rs779915637 | NA | <0.01 | D | B | D | B | B | B | B | NA | NA | NA |
| Ex 10 | c.1411A>G | p.Ser471Gly | MS | Het | AR | In this study | NA | NA | NA | D | D | D | D | D | D | D | NA | NA | NA |
| 85 | S-4386 | Neuropathy | *DCTN1* | Ex 12 | c.1211T>C | p.Val404Ala | MS | Het | AD | NA | rs760745970 | NA | <0.01 | D | D | D | D | D | D | NA | NA | NA | NA |
| 86 | R-0100 | Microcephaly | *DDX11* | Ex 10 | c.1221C>A | p.Ser407Arg | MS | Het | AR | NA | rs138190731 | NA | <0.01 | B | D | D | B | D | D | NA | NA | NA | NA |
| *DDX11* | Ex 18 | c.1774C>G | p.Gln592Glu | MS | Het | AR | NA | rs2911826 | RCV000  203006.1 | NA | B | B | D | B | B | B | NA | NA | NA | NA |
| 87 | S-1593 | ID | *DEAF1* | Ex 9 | c.1187G>T | p.Gly396Val | MS | Het | AD | In this study | NA | NA | NA | B | B | D | B | D | D | NA | NA | NA | NA |
| 88 | S-4151 | Neuropathy | *DIAPH3* | Ex 24 | c.2971A>T | p.Lys991Ter | NS | Het | AD | NA | rs531346416 | NA | <0.01 | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA |
| 89 | S-4327 | Muscular dystrophy | *DMD* | Ex 68 | c.9955T>C | p.Cys3319Arg | MS | Hemi | XL | NA | rs886044217 | RCV000  370541.1 | NA | D | B | D | B | D | B | NA | NA | NA | NA |
| 90 | S-2546 | Muscular dystrophy, Myopathy | *DNAJB6* | Ex 9 | c.706G>A | p.Asp236Asn | MS | Het | AD | NA | rs556999563 | RCV000  648072.1 | <0.01 | B | B | B | B | D | B | NA | NA | NA | NA |
| 91 | S-3761 | Epilepsy | *DNM1* | Ex 20 | c.2170C>T | p.Arg724Trp | MS | Het | AD | NA | rs1386035686 | NA | NA | B | B | D | D | D | D | NA | NA | NA | NA |
| 92 | S-5743 | Epilepsy | *DNM1L* | Int 16 | c.1707+1G>A | NA | SS | Het | AD/AR | In this study | NA | NA | NA | NA | NA | NA | NA | NA | NA | D | D | B | NA |
| 93 | S-5507 | Epilepsy | *DNM1L* | Ex 17 | c.1849C>T | p.Pro617Ser | MS | Het | AD | NA | rs753979883 | NA | <0.01 | D | D | D | B | D | B | NA | NA | NA | NA |
| 94 | S-3158 | ASD, ID | *DPP6* | Ex 16 | c.1442A>G | p.His481Arg | MS | Het | AD | In this study | NA | NA | NA | B | D | D | B | D | B | NA | NA | NA | NA |
| 95 | S-2001 | Lissencephaly | *DYNC1H1* | Ex 8 | c.1505C>T | p.Pro502Leu | MS | Het | AD | NA | rs1435555404 | NA | NA | B | D | D | B | B | B | NA | NA | NA | NA |
| 96 | S-2058 | Muscular dystrophy | *DYNC1H1* | Ex 19 | c.4105C>A | p.Gln1369Lys | MS | Het | AD | In this study | NA | NA | NA | B | D | D | B | B | B | NA | NA | NA | NA |
| 97 | S-846 | Neuropathy | *DYNC1H1* | Ex 48 | c.9332G>C | p.Ser3111Thr | MS | Het | AD | NA | rs1395404099 | NA | NA | B | D | D | B | B | B | NA | NA | NA | NA |
| 98 | S-4324 | Neuropathy | *DYNC1H1* | Ex 77 | c.13719\_13721delCAA | p.Asn4573del | Indel | Het | AD | NA | rs1013209915 | RCV000  649562.1 | NA | NA | NA | D | NA | NA | NA | NA | NA | NA | D |
| 99 | S-2546 | Muscular dystrophy, Myopathy | *DYSF* | Int 10 | c.938-6\_938-3delTCTT | NA | SS | Het | AR | In this study | NA | NA | NA | NA | NA | NA | NA | NA | NA | D | NA | D | NA |
| Ex 46 | c.5078G>C | p.Arg1693Pro | MS | Het | AR | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 100 | S-6015 | Muscular dystrophy | *DYSF* | Ex 52 | c.5827T>C | p.Ser1943Pro | MS | Hom | AR | In this study | NA | NA | NA | D | B | D | D | D | D | NA | NA | NA | NA |
| 101 | S-3051 | Epilepsy | *EFHC1* | Ex 2 | c.202\_203insAGGAAC  CAGTC | p.Ala69GlufsTer8 | Indel | Het | AD | In this study | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA |
| 102 | S-5230 | Epilepsy, Ataxia | *EFHC1* | Ex 5 | c.800A>G | p.Tyr267Cys | MS | Het | AD | NA | rs886061629 | RCV000  260926 | NA | B | D | D | D | D | D | D | NA | NA | NA |
| 103 | S-3158 | ASD, ID | *EFHC2* | Ex 12 | c.1901A>G | p.Glu634Gly | MS | Hemi | XL | NA | rs1209679571 | NA | NA | B | D | D | D | D | B | NA | NA | NA | NA |
| 104 | S-2613 | Leukodystrophy | *EIF2B5* | Ex 5 | c.745A>T | p.Ile249Phe | MS | Hom | AR | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 105 | S-5748 | Neuropathy | *ELAC2* | Ex 1 | c.82C>T | p.Arg28Cys | MS | Hom | AR | NA | rs763864314 | RCV000530273.1 | <0.01 | D | D | D | D | B | B | NA | NA | NA | NA |
| 106 | S-5331 | Muscle weakness | *EMD* | Ex 6 | c.548C>G | p.Pro183Arg | MS | Hemi | XL | In this study | NA | NA | NA | D | B | B | B | D | D | NA | NA | NA | NA |
| 107 | S-1713 | Microcephaly | *EP300* | Ex 19 | c.3559C>T | p.Arg1187Cys | MS | Het | AD | NA | rs745492786 | NA | <0.01 | D | D | D | D | D | D | NA | NA | NA | NA |
| 108 | S-2073 | Epilepsy, ASD, ID | *EPB41L1* | Ex 15 | c.1717C>A | p.Pro573Thr | MS | Het | AD | In this study | NA | NA | NA | D | B | D | B | D | B | NA | NA | NA | NA |
| 109 | S-5839 | Spasticity | *FA2H* | Ex 5 | c.664G>A | p.Gly222Arg | MS | Hom | AR | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 110 | S-4435 | Epilepsy | *FASN* | Ex 17 | c.2644G>A | p.Gly882Ser | MS | Het | AD | NA | rs748305734 | NA | <0.01 | D | B | D | D | D | D | NA | NA | NA | NA |
| 111 | S-2493 | Myopathy | *FKBP14* | Ex 1 | c.143T>A | p.Met48Lys | MS | Hom | AR | In this study | NA | NA | NA | B | D | D | B | D | D | NA | NA | NA | NA |
| 112 | S-1537 | Muscular dystrophy | *FKRP* | Ex 4 | c.671T>C | p.Leu224Pro | MS | Hom | AR | In this study | NA | NA | NA | D | B | D | B | D | D | NA | NA | NA | NA |
| 113 | S-3407 | Muscular dystrophy | *FKRP* | Ex 4 | c.869A>T | p.Asn290Ile | MS | Hom | AR | In this study | NA | NA | NA | D | B | B | B | B | D | NA | NA | NA | NA |
| 114 | S-1963 | Muscular dystrophy | *FKRP* | Ex 4 | c.935G>T | p.Arg312Leu | MS | Hom | AR | \*11741828 | rs868138875 | NA | NA | D | D | D | B | B | B | NA | NA | NA | NA |
| 115 | R-0605 | Brain malformations | *FKTN* | Ex 11 | c.1337A>T | p.Asn446Ile | MS | Het | AR | NA | rs374912618 | RCV000634077.1 | <0.01 | B | D | D | D | D | D | NA | NA | NA | NA |
| 116 | R-0109 | Spasticity | *FLNA* | Ex 12 | c.1807G>A | p.Gly603Arg | MS | Hemi | XL | NA | rs200116438 | NA | <0.01 | B | B | D | B | D | D | NA | NA | NA | NA |
| 117 | S-4003 | Epilepsy | *FLNA* | Ex 15 | c.2179G>A | p.Gly727Ser | MS | Hemi | XL | In this study | NA | NA | NA | D | D | D | D | D | B | NA | NA | NA | NA |
| 118 | S-2971 | Epilepsy, Dystonia | *FLNA* | Ex 32 | c.5308G>C | p.Val1770Leu | MS | Hemi | XL | In this study | NA | NA | NA | D | B | D | B | B | B | NA | NA | NA | NA |
| 119 | S-3407 | Muscular dystrophy | *FLNC* | Ex 12 | c.1936G>A | p.Asp646Asn | MS | Het | AD | NA | rs372668691 | NA | <0.01 | D | D | D | B | D | B | NA | NA | NA | NA |
| 120 | S-5249 | Muscular dystrophy | *FLNC* | Ex 19 | c.2848C>G | p.Pro950Ala | MS | Het | AD | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 121 | S-1933 | Epilepsy | *FOLR1* | Int 4 | c.493+2\_493+6delTGAGG | NA | SS | Hom | AR | In this study | NA | NA | NA | NA | NA | NA | NA | NA | NA | D | D | D | NA |
| 122 | S-2710 | ASD | *FOXP1* | Ex 20 | c.1768A>G | p.Met590Val | MS | Het | AD | NA | rs201446635 | NA | <0.01 | D | D | D | B | B | B | NA | NA | NA | NA |
| 123 | S-4325 | Leukodystrophy | *GALC* | Ex 8 | c.776C>T | p.Ser259Leu | MS | Hom | AR | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 124 | S-2248 | Leukodystrophy | *GALC* | Ex 14 | c.1660G>C | p.Asp554His | MS | Hom | AR | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 125 | S-6075 | Leukodystrophy | *GALC* | Ex 15 | c.1685T>A | p.Ile562Lys | MS | Hom | AR | In this study | NA | NA | NA | D | B | B | D | D | D | NA | NA | NA | NA |
| 126 | S-3679 | Obsessive disease, auditory hallucination | *GARS* | Ex 15 | c.1855C>T | p.Leu619Phe | MS | Het | AD | NA | rs751239315 | NA | <0.01 | D | D | D | B | D | B | NA | NA | NA | NA |
| 127 | S-4324 | Neuropathy | *GDAP1* | Ex 5 | c.685G>A | p.Glu229Lys | MS | Het | AD/AR | In this study | NA | NA | NA | D | D | D | B | D | B | NA | NA | NA | NA |
| 128 | S-2346 | Leukodystrophy | *GFAP* | Ex 1 | c.229A>G | p.Asn77Asp | MS | Het | AD | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 129 | S-5688 | Epilepsy | *GFAP* | Ex 1 | c.362G>A | p.Arg121Gln | MS | Het | AD | NA | rs769619261 | NA | <0.01 | D | D | D | B | D | D | NA | NA | NA | NA |
| 130 | S-3669 | Spasticity | *GFAP* | Ex 5 | c.901G>C | p.Gly301Arg | MS | Het | AD | NA | rs1265636628 | NA | NA | D | D | D | B | D | D | NA | NA | NA | NA |
| 131 | S-2660 | Epilepsy | *GLB1* | Ex 8 | c.899G>T | p.Gly300Val | MS | Hom | AR | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 132 | S-1918 | Epilepsy | *GRIN2A* | Ex 4 | c.605A>G | p.Asn202Ser | MS | Het | AD | In this study | NA | NA | NA | D | D | D | B | B | B | NA | NA | NA | NA |
| 133 | S-3118 | Epilepsy | *GRIN2A* | Ex 7 | c.1351A>T | p.Asn451Tyr | MS | Het | AD | In this study | NA | NA | NA | B | D | D | B | D | B | NA | NA | NA | NA |
| 134 | S-2328 | Epilepsy | *GRIN2A* | Ex 7 | c.1351A>T | p.Asn451Tyr | MS | Het | AD | In this study | NA | NA | NA | B | D | D | B | D | B | NA | NA | NA | NA |
| 135 | R-0529 | Epilepsy, Microcephaly | *GRIN2A* | Ex 13 | c.2482A>G | p.Met828Val | MS | Het | AD | In this study | NA | NA | NA | B | D | D | B | D | B | NA | NA | NA | NA |
| 136 | S-4545 | Epilepsy, ASD | *GRIN2A* | Ex 14 | c.4090G>A | p.Asp1364Asn | MS | Het | AD | NA | rs1245573153 | NA | NA | B | B | D | B | B | B | NA | NA | NA | NA |
| 137 | S-1242 | ASD | *GRIN2A* | Ex 14 | c.4139G>A | p.Gly1380Glu | MS | Het | AD | In this study | NA | NA | NA | B | D | D | D | D | B | NA | NA | NA | NA |
| 138 | S-3202 | Epilepsy | *HCN1* | Ex 4 | c.1141G>A | p.Val381Ile | MS | Het | AD | NA | rs750838439 | NA | <0.01 | D | B | D | B | D | B | NA | NA | NA | NA |
| 139 | S-3270 | Epilepsy, ASD | *HCN1* | Ex 8 | c.2317G>C | p.Ala773Pro | MS | Het | AD | In this study | NA | NA | NA | B | D | D | B | B | B | NA | NA | NA | NA |
| 140 | S-5488 | Epilepsy | *HCN1* | Ex 8 | c.2564G>T | p.Gly855Val | MS | Het | AD | NA | rs755932633 | NA | <0.01 | D | D | D | B | B | B | NA | NA | NA | NA |
| 141 | R-0690 | Epilepsy | *HGSNAT* | Ex 9 | c.836A>C | p.Asp279Ala | MS | Hom | AR | NA | rs1085307112 | RCV000  488421.1 | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 142 | S-5913 | Epilepsy | *HNRNPU* | Ex 6 | c.1172G>A | p.Cys391Tyr | MS | Het | AD | In this study | NA | NA | NA | B | D | D | B | D | B | NA | NA | NA | NA |
| 143 | S-1196 | Neuropathy | *HSPB1* | Ex 2 | c.383A>G | p.Gln128Arg | MS | Het | AD | NA | rs558882005 | NA | <0.01 | D | D | D | B | D | D | NA | NA | NA | NA |
| 144 | S-5303 | Neuropathy | *HSPB1* | Ex 3 | c.440G>A | p.Gly147Asp | MS | Het | AD | In this study | NA | NA | NA | D | B | D | B | D | B | NA | NA | NA | NA |
| 145 | S-2554 | Epilepsy, Leukodystrophy | *HTRA1* | Ex 6 | c.1022G>A | p.Gly341Glu | MS | Hom | AR | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 146 | R-0687 | Epilepsy | *IDUA* | Ex 3 | c.382C>T | p.Pro128Ser | MS | Het | AR | NA | rs754674352 | NA | <0.01 | D | D | D | D | D | D | NA | NA | NA | NA |
| Ex 9 | c.1401G>A | p.Pro467= | SS | Het | AR | NA | rs1005671672 | NA | NA | NA | NA | NA | NA | NA | NA | B | NA | D | NA |
| 147 | S-2047 | Microcephaly, Dystonia | *IFIH1* | Ex 1 | c.281A>G | p.Tyr94Cys | MS | Het | AD | NA | rs1325840950 | NA | NA | B | D | D | D | D | D | NA | NA | NA | NA |
| 148 | S-4086 | Epilepsy, Microcephaly | *IFIH1* | Ex 7 | c.1433T>C | p.Val478Ala | MS | Het | AD | In this study | NA | NA | NA | B | B | B | B | B | B | NA | NA | NA | NA |
| 149 | S-84 | Epilepsy, Microcephaly, ID | *IFIH1* | Ex 9 | c.1652A>C | p.Lys551Thr | MS | Het | AD | NA | rs577764827 | NA | <0.01 | B | D | D | D | D | B | NA | NA | NA | NA |
| 150 | S-4716 | Leukodystrophy | *IFIH1* | Ex 14 | c.2642T>A | p.Ile881Lys | MS | Het | AD | In this study | NA | NA | NA | B | D | D | D | B | B | NA | NA | NA | NA |
| 151 | S-3729 | Epilepsy, Microcephaly | *IFIH1* | Ex 14 | c.2784C>G | p.His928Gln | MS | Het | AD | NA | rs145187664 | NA | NA | B | D | D | D | D | D | NA | NA | NA | NA |
| 152 | S-1531 | Neuropathy | *IGHMBP2* | Ex 8 | c.1198G>A | p.Asp400Asn | MS | Hom | AR | NA | rs779654686 | RCV000  642636.1 | <0.01 | D | D | D | D | D | D | NA | NA | NA | NA |
| 153 | S-151 | ID | *IL1RAPL1* | Ex 3 | c.194C>A | p.Ser65Tyr | MS | Hemi | XL | In this study | NA | NA | NA | B | D | D | B | D | D | NA | NA | NA | NA |
| 154 | S-2691 | ASD | *IQSEC2* | Ex 9 | c.2774C>G | p.Pro925Arg | MS | Hemi | XL | In this study | NA | NA | NA | B | D | D | D | D | D | NA | NA | NA | NA |
| 155 | S-4042 | Epilepsy, Microcephaly | *IQSEC2* | Ex 10 | c.2983C>T | p.Arg995Trp | MS | Hemi | XL | NA | rs1057521657 | RCV000  425290.1 | NA | B | D | D | D | D | D | NA | NA | NA | NA |
| 156 | S-3730 | Epilepsy, ASD | *IQSEC2* | Ex12 | c.3227T>C | p.Leu1076Pro | MS | Het | XL | In this study | NA | NA | NA | B | D | D | D | D | D | NA | NA | NA | NA |
| 157 | S-2972 | Ataxia, ID | *ITPR1* | Ex 10 | c.755C>T | p.Thr252Ile | MS | Het | AD | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 158 | S-5822 | Epilepsy, Ataxia | *ITPR1* | Ex 15 | c.1447G>A | p.Val483Ile | MS | Het | AD | NA | rs756745652 | NA | NA | D | D | D | D | B | B | NA | NA | NA | NA |
| 159 | S-4134 | Ataxia | *ITPR1* | Ex 36 | c.4712G>A | p.Arg1571His | MS | Het | AD | NA | rs754960125 | NA | <0.01 | B | D | D | D | D | D | NA | NA | NA | NA |
| 160 | S-2849 | ID | *ITPR1* | Ex 47 | c.6349A>G | p.Ser2117Gly | MS | Het | AD/AR | NA | rs1426471157 | NA | NA | D | B | D | B | B | B | NA | NA | NA | NA |
| 161 | S-3139 | Leukodystrophy | *KARS* | Ex 7 | c.764A>G | p.Tyr255Cys | MS | Hom | AR | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 162 | S-3677 | ASD | *KATNAL2* | Ex 2 | c.42C>G | p.Tyr14Ter | NS | Het | AD | In this study | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA |
| 163 | S-1480 | Epilepsy | *KBTBD13* | Ex 1 | c.172\_191del | p.Phe58AlafsTer80 | Indel | Het | AD | NA | rs778782105 | NA | <0.01 | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA |
| 164 | S-4102 | Epilepsy | *KCNMA1* | Ex 19 | c.2108C>T | p.Pro703Leu | MS | Het | AD | NA | rs771363378 | NA | <0.01 | D | D | D | B | B | D | B | NA | NA | NA |
| 165 | S-4152 | Epilepsy | *KCNQ2* | Ex 1 | c.62A>C | p.Lys21Thr | MS | Het | AD | In this study | NA | NA | NA | D | B | D | B | B | B | NA | NA | NA | NA |
| 166 | S-5296 | Epilepsy | *KCNQ2* | Ex 5 | c.760G>A | p.Glu254Lys | MS | Het | AD | In this study | NA | NA | NA | D | B | D | D | D | D | NA | NA | NA | NA |
| 167 | S-4055 | Epilepsy | *KCNQ2* | Ex 17 | c.2108C>T | p.Ala703Val | MS | Het | AD | NA | rs779430808 | NA | <0.01 | D | D | D | B | D | B | NA | NA | NA | NA |
| 168 | S-2377 | Abnormal head and limb movements | *KCTD7* | Ex 3 | c.458G>C | p.Arg153Pro | MS | Hom | AR | NA | rs765235486 | NA | NA | D | B | D | D | D | B | NA | NA | NA | NA |
| 169 | S-4962 | Kabuki syndrome | *KDM6A* | Ex 16 | c.1861C>T | p.Arg621Cys | MS | Het | XL | NA | rs139486036 | NA | <0.01 | B | D | D | B | D | B | NA | NA | NA | NA |
| 170 | S-4478 | Ataxia, ID | *KIF1A* | Int 35 | c.3705-3C>A | NA | SS | Het | AD | NA | rs879129733 | NA | NA | NA | NA | NA | NA | NA | NA | D | NA | NA | NA |
| 171 | S-4060 | ASD | *KIF1A* | Ex 36 | c.3728C>T | p.Ala1243Val | MS | Het | AD | NA | rs876661146 | RCV000  213368.1 | NA | B | B | D | B | D | B | NA | NA | NA | NA |
| 172 | S-6030 | Muscular dystrophy | *KRAS* | Ex 5 | c.491G>A | p.Arg164Gln | MS | Het | AD | NA | rs758575947 | NA | <0.01 | B | D | D | D | D | D | NA | NA | NA | NA |
| 173 | S-0297 | Muscular dystrophy | *LAMA2* | Ex 21 | c.2916T>G | p.Phe972Leu | MS | Het | AR | NA | rs763840955 | ΔRCV000  654716.1 | <0.01 | B | D | D | B | D | B | NA | NA | NA | NA |
| Ex 29 | c.4223G>A | p.Arg1408His | MS | Het | AR | NA | rs751858884 | NA | <0.01 | B | B | D | B | B | B | NA | NA | NA | NA |
| 174 | S-2250 | Muscular dystrophy | *LMNA* | Ex 7 | c.1325T>C | p.Val442Ala | MS | Het | AD | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 175 | S-1738 | Neuropathy | *LRSAM1* | Ex 6 | c.284C>T | p.Ala95Val | MS | Hom | AR | NA | rs570248730 | RCV000  649924.1 | <0.01 | B | B | B | B | D | D | NA | NA | NA | NA |
| 176 | S-661 | Neuropathy | *LRSAM1* | Ex 16 | c.1198C>T | p.Arg400Trp | MS | Het | AD | NA | rs749575647 | NA | <0.01 | B | B | D | B | D | D | NA | NA | NA | NA |
| 177 | S-1781 | Dementia | *MAPT* | Ex 6 | c.919A>G | p.Ile307Val | MS | Het | AD | In this study | NA | NA | NA | B | D | D | D | D | B | NA | NA | NA | NA |
| 178 | S-1788 | ASD, ID | *MBD5* | Ex 9 | c.845G>A | p.Gly282Asp | MS | Het | AD | NA | rs759201974 | NA | <0.01 | B | D | D | B | D | D | NA | NA | NA | NA |
| 179 | S-4058 | ASD | *MBD5* | Ex 9 | c.2035G>C | p.Ala679Pro | MS | Het | AD | In this study | NA | NA | NA | B | B | B | B | B | B | NA | NA | NA | NA |
| 180 | S-2585 | Epilepsy | *MECP2* | Ex 4 | c.1246\_1248delGAG | p.Glu416del | Indel | Het | XL | NA | rs781918838 | NA | <0.01 | NA | NA | D | NA | NA | NA | NA | NA | NA | D |
| 181 | S-2857 | Epilepsy | *MECP2* | Ex 4 | c.1261G>C | p.Gly421Arg | MS | Het | XL | In this study | NA | NA | NA | D | B | D | B | D | B | NA | NA | NA | NA |
| 182 | S-1595 | Van der Knaap disease | *MLC1* | Ex 10 | c.798C>G | p.Ser266Arg | MS | Hom | AR | NA | rs777790290 | NA | <0.01 | D | D | D | D | D | D | NA | NA | NA | NA |
| 183 | S-3957 | Myotonia | *MTM1* | Ex 10 | c.974A>G | p.Lys325Arg | MS | Hom | XL | In this study | NA | NA | NA | D | B | D | B | B | B | NA | NA | NA | NA |
| 184 | S-3956 | Muscular dystrophy | *MYH2* | Ex 19 | c.2075A>G | p.His692Arg | MS | Het | AD/AR | In this study | NA | NA | NA | D | B | D | B | D | B | NA | NA | NA | NA |
| 185 | S-4250 | Muscular dystrophy | *MYH2* | Ex 30 | c.4027C>G | p.Arg1343Gly | MS | Hom | AR | NA | rs145911509 | NA | <0.01 | D | B | D | D | D | D | NA | NA | NA | NA |
| 186 | S-4289 | Myopathy | *MYH7* | Ex 7 | c.610C>T | p.Arg204Cys | MS | Het | AD | NA | rs397516259 | RCV000  628868.1 | <0.01 | D | B | D | B | D | D | NA | NA | NA | NA |
| 187 | S-4476 | Myopathy | *MYH7* | Ex 27 | c.3430G>A | p.Glu1144Lys | MS | Het | AD | In this study | NA | NA | NA | B | B | D | D | D | D | NA | NA | NA | NA |
| 188 | S-1593 | ID | *MYO1A* | Ex 8 | c.559C>T | p.Arg187Ter | NS | Het | AD | NA | rs146269737 | NA | <0.01 | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA |
| 189 | R-0536 | Leukodystrophy | *NAGLU* | Ex 6 | c.1124G>A | p.Arg375His | MS | Hom | AR | NA | rs768600049 | NA | <0.01 | D | B | B | D | D | B | NA | NA | NA | NA |
| 190 | S-1386 | Epilepsy | *NEDD4L* | Ex 15 | c.1342C>T | p.Arg448Trp | MS | Het | AD | NA | rs1199827684 | NA | NA | B | B | D | B | D | D | NA | NA | NA | NA |
| 191 | S-1100 | Epilepsy, Ataxia, ASD | *NHS* | Ex 6 | c.1438C>T | p.Arg480Cys | MS | Hemi | XL | NA | rs770144581 | NA | <0.01 | B | D | D | D | D | D | NA | NA | NA | NA |
| 192 | S-5934 | Microcephaly, ID | *NIPBL* | Ex 4 | c.256G>A | p.Asp86Asn | MS | Het | AD | NA | rs1188531884 | NA | NA | D | D | D | D | D | B | NA | NA | NA | NA |
| 193 | S-5811 | Epilepsy | *NIPBL* | Ex 10 | c.2423G>C | p.Arg808Pro | MS | Het | AD | NA | rs142574933 | NA | <0.01 | D | D | D | B | D | B | NA | NA | NA | NA |
| 194 | S-1581 | CdLS | *NIPBL* | Int 20 | c.4422-3A>G | NA | SS | Het | AD | In this study | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | D | D | NA |
| 195 | S-4474 | CdLS | *NIPBL* | Int 34 | c.6108+7A>G | NA | SS | Het | AD | NA | rs587783995 | RCV000  146668.1 | NA | NA | NA | NA | NA | NA | NA | D | NA | B | NA |
| 196 | S-1274 | Epilepsy | *NIPBL* | Int 34 | c.6109-3T>C | NA | SS | Het | AD | NA | rs145778995 | RCV000  082498.9 | <0.01 | NA | NA | NA | NA | NA | NA | B | NA | B | NA |
| 197 | S-2652 | Microcephaly | *NIPBL* | Ex 44 | c.7639C>T | p.Leu2547Phe | MS | Het | AD | NA | rs774934420 | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 198 | S-4545 | Epilepsy, ASD | *NLGN3* | Ex 7 | c.1955C>T | p.Pro652Leu | MS | Hemi | XL | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 199 | S-1536 | ASD | *NLGN3* | Ex 7 | c.2165G>A | p.Arg722Gln | MS | Hemi | XL | NA | rs779740957 | NA | <0.01 | B | D | D | B | B | B | NA | NA | NA | NA |
| 200 | S-2416 | Leukodystrophy | *NOTCH3* | Ex 17 | c.2771A>T | p.Asp924Val | MS | Het | AD | In this study | NA | NA | NA | D | B | D | D | D | D | NA | NA | NA | NA |
| 201 | R-0532 | Dystonia | *NPC1* | Ex 4 | c.445G>A | p.Gly149Arg | MS | Het | AR | NA | rs143205855 | RCV000  594495.1 | <0.01 | D | D | D | D | B | B | NA | NA | NA | NA |
| 202 | S-3225 | ASD, ID | *NSD1* | Ex 12 | c.4754A>C | p.Glu1585Ala | MS | Het | AD | In this study | NA | NA | NA | D | D | D | B | D | B | NA | NA | NA | NA |
| 203 | S-1274 | Epilepsy | *OCRL* | Int 6 | c.439+3A>G | NA | SS | Hemi | XL | NA | rs61752971 | RCV000  632787.1 | <0.01 | NA | NA | NA | NA | NA | NA | B | NA | D | NA |
| 204 | S-5390 | Ataxia | *OPA1* | Ex 8 | c.860A>G | p.His287Arg | MS | Het | AD | In this study | NA | NA | NA | D | D | D | B | D | B | NA | NA | NA | NA |
| 205 | R-0518 | ID | *OPHN1* | Ex 19 | c.1682G>A | p.Gly561Asp | MS | Hemi | XL | NA | rs751031568 | NA | <0.01 | D | D | D | D | D | D | NA | NA | NA | NA |
| 206 | S-3928 | Epilepsy | *PCDH19* | Ex 3 | c.2294C>A | p.Ala765Asp | MS | Hemi | XL | NA | rs1165334173 | NA | NA | B | D | D | D | D | D | NA | NA | NA | NA |
| 207 | S-4924 | ASD | *PCDH19* | Ex 6 | c.3019G>T | p.Asp1007Tyr | MS | Het | XL | In this study | NA | NA | NA | B | D | D | B | D | D | NA | NA | NA | NA |
| 208 | S-3955 | Developmental delay | *PDHA1* | Int 10 | c.1009-31\_1009-8del | NA | SS | Hemi | XL | In this study | NA | NA | NA | NA | NA | NA | NA | NA | NA | D | D | D | NA |
| 209 | S-4325 | Leukodystrophy | *PEX1* | Ex 21 | c.3283G>A | p.Asp1095Asn | MS | Hom | AR | NA | rs753699011 | RCV000  597351.1 | <0.01 | D | D | D | B | B | B | NA | NA | NA | NA |
| 210 | S-2707 | Leukodystrophy, Spasticity | *PEX16* | Ex 9 | c.829C>T | p.Arg277Trp | MS | Hom | AR | NA | rs1381800210 | NA | NA | B | D | D | D | D | D | NA | NA | NA | NA |
| 211 | S-5295 | Ataxia, Spasticity | *PEX6* | Ex 1 | c.62T>A | p.Val21Glu | MS | Hom | AR | In this study | NA | NA | NA | D | B | D | B | D | D | NA | NA | NA | NA |
| 212 | S-1590 | Microcephaly | *PHF6* | Ex 8 | c.800C>T | p.Thr267Ile | MS | Hemi | XL | In this study | NA | NA | NA | D | D | D | B | D | D | NA | NA | NA | NA |
| 213 | S-2534 | Epilepsy, ASD | *PIGN* | Ex 5 | c.329G>A | p.Ser110Asn | MS | Het | AR | NA | rs746450459 | NA | <0.01 | B | D | D | D | D | B | NA | NA | NA | NA |
| Ex 26 | c.2371G>A | p.Val791Ile | MS | Het | AR | NA | rs1204147408 | NA | NA | B | D | D | B | D | B | NA | NA | NA | NA |
| 214 | S-4894 | Lissencephaly | *PIK3R2* | Ex 5 | c.586C>T | p.Arg196Trp | MS | Het | AD | NA | rs770193876 | NA | NA | B | B | B | D | D | D | NA | NA | NA | NA |
| 215 | S-2072 | Microcephaly | *PLA2G6* | Ex 8 | c.1097T>A | p.Ile366Asn | MS | Hom | AR | NA | rs778225931 | NA | <0.01 | B | D | D | D | D | D | NA | NA | NA | NA |
| 216 | R-0862 | Infantile neuroaxonal dystrophy | *PLA2G6* | Ex 11 | c.1580C>A | p.Ala527Asp | MS | Hom | AR | NA | rs1422865730 | NA | NA | B | D | D | D | D | D | NA | NA | NA | NA |
| 217 | R-0612 | Neuropathy | *PLEKHG5* | Ex 14 | c.1405C>T | p.His469Tyr | MS | Het | AR | In this study | NA | NA | NA | B | D | D | D | D | D | NA | NA | NA | NA |
| 218 | S-4602 | Epilepsy, Microcephaly, Leukodystrophy | *PNPLA6* | Ex 16 | c.1472T>C | p.Ile491Thr | MS | Hom | AR | NA | rs1246639539 | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 219 | S-5822 | Epilepsy, Ataxia | *POLG* | Ex 16 | c.2509T>C | p.Tyr837His | MS | Hom | AR | NA | rs544828395 | RCV000  518238.1 | <0.01 | D | D | D | D | D | B | NA | NA | NA | NA |
| 220 | S-1334 | Myopathy, Neuropathy | *POLG2* | Ex 5 | c.1105A>G | p.Arg369Gly | MS | Het | AD | \*21555342 | rs201936720 | RCV000  198981.2 | <0.01 | D | D | D | D | D | D | NA | NA | NA | NA |
| 221 | S-4503 | Leukodystrophy | *POLR1C* | Ex 2 | c.118T>G | p.Trp40Gly | MS | Hom | AR | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 222 | R-1349 | Developmental delay | *POLR3A* | Ex 23 | c.2993G>A | p.Arg998His | MS | Hom | AR | NA | rs139665002 | NA | <0.01 | B | D | D | B | B | B | NA | NA | NA | NA |
| 223 | S-3015 | Lissencephaly | *POMGNT2* | Ex 2 | c.239G>A | p.Arg80His | MS | Hom | AR | NA | rs146511234 | RCV000  650546.1 | <0.01 | B | D | D | B | D | B | NA | NA | NA | NA |
| 224 | S-2393 | Leukodystrophy | *POMGNT2* | Ex 2 | c.629G>T | p.Ser210Ile | MS | Hom | AR | NA | rs561770941 | NA | <0.01 | D | D | D | D | D | D | NA | NA | NA | NA |
| 225 | S-5896 | Epilepsy | *PPT1* | Ex 6 | c.603\_605dupCTT | p.Phe201dup | Indel | Hom | AR | In this study | NA | NA | NA | NA | NA | D | NA | NA | NA | NA | NA | NA | D |
| 226 | S-3351 | Epilepsy | *PPT1* | Ex 7 | c.713C>T | p.Pro238Leu | MS | Hom | AR | NA | rs878853322 | RCV0002  25555.1 | NA | D | D | D | D | D | D | D | NA | NA | NA |
| 227 | S-1931 | Epilepsy, ASD | *PRICKLE2* | Ex 7 | c.883C>T | p.Arg295Trp | MS | Het | AD | NA | rs776167467 | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 228 | S-1403 | Epilepsy | *PRRT2* | Ex 3 | c.954C>G | p.Ile318Met | MS | Het | AD | In this study | NA | NA | NA | D | B | D | D | D | B | NA | NA | NA | NA |
| 229 | S-2289 | Spasticity | *PSAP* | Ex 4 | c.257T>A | p.Ile86Asn | MS | Hom | AR | NA | NA | RCV000  624741.1 | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 230 | S-4683 | Leukodystrophy, Neuropathy | *PSAP* | Ex 4 | c.257T>A | p.Ile86Asn | MS | Hom | AR | NA | NA | RCV000624741.1 | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 231 | S-1063 | Epilepsy | *RAB27A* | Ex 4 | c.340A>G | p.Ile114Val | MS | Hom | AR | In this study | NA | NA | NA | B | D | D | B | B | B | NA | NA | NA | NA |
| 232 | R-0556 | Warburg micro syndrome | *RAB3GAP1* | Ex 24 | c.2894\_2895delAA | p.Lys965ArgfsTer4 | Indel | Hom | AR | In this study | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA |
| 233 | R-0527 | Periventricular white matter cystic changes | *RAB3GAP2* | Ex 18 | c.1853A>G | p.Asp618Gly | MS | Het | AR | NA | rs1369418378 | NA | NA | B | D | D | B | D | B | NA | NA | NA | NA |
| 234 | S-3843 | Epilepsy | *RANBP2* | Ex 20 | c.4433\_4434insAAAGGAAAAAGCTTTGTCCTTTCCTTCTAA | p.Glu1480\_Gly1481  insLysAlaLeuSerPhe  Pro SerLysLysGlu | Indel | Het | AD | In this study | NA | NA | NA | NA | NA | D | NA | NA | NA | NA | NA | NA | D |
| 235 | S-132 | Epilepsy | *RELN* | Ex 37 | c.5594C>T | p.Ser1865Leu | MS | Het | AD | NA | rs1386230430 | NA | NA | B | D | D | B | D | B | NA | NA | NA | NA |
| 236 | S-1713 | Microcephaly | *RELN* | Ex 46 | c.7283G>A | p.Arg2428Gln | MS | Hom | AR | NA | rs948422815 | NA | NA | B | D | D | B | D | D | NA | NA | NA | NA |
| 237 | S-2073 | Epilepsy, ASD, ID | *RELN* | Ex 48 | c.7651G>A | p.Ala2551Thr | MS | Het | AD | NA | rs1275052727 | NA | NA | B | D | D | D | D | B | NA | NA | NA | NA |
| 238 | S-4014 | Epilepsy, Leukodystrophy, ID | *RPIA* | Ex 6 | c.592T>C | p.Phe198Leu | MS | Hom | AR | In this study | NA | NA | NA | B | D | D | D | B | B | NA | NA | NA | NA |
| 239 | S-6023 | Epilepsy, ID | *RPS6KA3* | Ex 22 | c.2135G>A | p.Arg712His | MS | Het | XL | NA | rs1017565774 | NA | NA | B | D | D | B | B | D | NA | NA | NA | NA |
| 240 | S-2950 | Myopathy | *RYR1* | Ex 12 | c.1198G>A | p.Ala400Thr | MS | Het | AR | NA | rs777016690 | NA | NA | D | B | D | D | D | D | NA | NA | NA | NA |
| Ex 17 | c.1817G>A | p.Cys606Tyr | MS | Het | AR | In this study | NA | NA | NA | D | B | D | D | D | D | NA | NA | NA | NA |
| 241 | S-1799 | Muscular dystrophy | *RYR1* | Ex 24 | c.3175C>A | p.Pro1059Thr | MS | Het | AD/AR | In this study | NA | NA | NA | D | B | B | B | B | B | NA | NA | NA | NA |
| 242 | S-4525 | Myasthenic syndrome | *RYR1* | Ex 89 | c.12238G>A | p.Val4080Ile | MS | Hom | AR | In this study | NA | NA | NA | D | D | B | B | B | D | NA | NA | NA | NA |
| 243 | S-3377 | Epilepsy, Spasticity, Neuropathy | *SACS* | Int 7 | c.605-3C>G | NA | SS | Hom | AR | In this study | NA | NA | NA | NA | NA | NA | NA | NA | NA | D | D | D | NA |
| 244 | S-3405 | Ataxia | *SACS* | Ex 10 | c.7016A>G | p.Asp2339Gly | MS | Hom | AR | NA | rs557648891 | NA | <0.01 | D | D | D | B | B | D | NA | NA | NA | NA |
| 245 | S-2313 | Epilepsy | *SCN1A* | Ex 2 | c.316\_319delinsC | p.Ser106\_Ala107  delinsPro | Indel | Het | AD | In this study | NA | NA | NA | NA | NA | D | NA | NA | NA | NA | NA | NA | D |
| 246 | S-4761 | Epilepsy | *SCN1A* | Int 8 | c.1170+5G>A | NA | SS | Het | AD | In this study | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | D | D | NA |
| 247 | S-5698 | Epilepsy | *SCN1A* | Ex 16 | c.2972T>G | p.Leu991Arg | MS | Het | AD | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 248 | S-518 | Epilepsy, Microcephaly | *SCN1A* | Ex 18 | c.3638G>A | p.Arg1213Gln | MS | Het | AD | NA | rs566081370 | RCV0006600699.1 | <0.01 | D | B | B | B | B | B | NA | NA | NA | NA |
| 249 | S-1665 | Epilepsy | *SCN1A* | Ex 26 | c.5030T>C | p.Leu1677Pro | MS | Het | AD | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 250 | S-2607 | Epilepsy | *SCN1B* | Ex 2 | c.55G>A | p.Gly19Arg | MS | Het | AD | NA | rs771386831 | NA | <0.01 | D | D | D | B | D | B | NA | NA | NA | NA |
| 251 | S-987 | Epilepsy | *SCN2A* | Ex 10 | c.1220T>C | p.Leu407Pro | MS | Het | AD | In this study | NA | NA | NA | D | D | D | B | D | D | NA | NA | NA | NA |
| 252 | S-4029 | Epilepsy | *SCN2A* | Ex 27 | c.4952T>A | p.Phe1651Tyr | MS | Het | AD | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 253 | S-926 | Epilepsy | *SCN2A* | Ex 27 | c.5131A>G | p.Thr1711Ala | MS | Het | AD | In this study | NA | NA | NA | D | D | D | B | D | D | NA | NA | NA | NA |
| 254 | S-4029 | Epilepsy | *SCN3A* | Ex 13 | c.1966T>G | p.Leu656Val | MS | Het | AD | NA | rs764003738 | NA | <0.01 | D | D | D | D | D | B | NA | NA | NA | NA |
| 255 | S-5239 | Epilepsy, Neuropathy | *SCN3A* | Ex 19 | c.3494C>A | p.Pro1165Gln | MS | Het | AD | NA | rs563038965 | NA | <0.01 | D | D | D | D | D | D | B | NA | NA | NA |
| 256 | S-3901 | Microcephaly, Spasticity | *SCN4A* | Ex 16 | c.3004T>C | p.Trp1002Arg | MS | Hom | AR | NA | rs544082594 | NA | <0.01 | D | B | B | B | B | D | NA | NA | NA | NA |
| 257 | S-3957 | Myotonia | *SCN4A* | Ex 24 | c.4720G>A | p.Gly1574Ser | MS | Het | AD | NA | rs369437369 | NA | NA | D | D | D | B | D | D | NA | NA | NA | NA |
| 258 | S-5387 | Epilepsy | *SCN5A* | Ex 12 | c.1558A>G | p.Met520Val | MS | Het | AD | NA | rs769799503 | NA | <0.01 | D | B | B | B | B | B | B | NA | NA | NA |
| 259 | S-2313 | Epilepsy | *SCN8A* | Ex 2 | c.220C>A | p.Gln74Lys | MS | Het | AD | In this study | NA | NA | NA | D | B | B | B | B | B | NA | NA | NA | NA |
| 260 | S-4211 | Epilepsy | *SCN8A* | Ex 5 | c.572G>A | p.Arg191Gln | MS | Het | AD | NA | rs1345271275 | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 261 | S-752 | Epilepsy | *SCN8A* | Ex 7 | c.875A>G | p.Tyr292Cys | MS | Het | AD | NA | rs760924400 | NA | <0.01 | D | B | D | B | D | B | NA | NA | NA | NA |
| 262 | S-5997 | Epilepsy | *SCN8A* | Ex 12 | c.1648A>C | p.Ile550Leu | MS | Het | AD | In this study | NA | NA | NA | D | D | D | D | D | B | NA | NA | NA | NA |
| 263 | S-4199 | Epilepsy | *SCN8A* | Ex 13 | c.2074G>A | p.Gly692Arg | MS | Het | AD | NA | rs1037521613 | NA | NA | D | D | D | B | D | B | NA | NA | NA | NA |
| 264 | S-2534 | Epilepsy, ASD | *SCN8A* | Ex 27 | c.4951C>G | p.Leu1651Val | MS | Het | AD | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 265 | S-3192 | Epilepsy, Neuropathy | *SCN9A* | Ex 5 | c.554G>A | p.Arg185His | MS | Het | AD | \*22826602 | rs73969684 | RCV000459981.3 | <0.01 | D | B | D | D | D | D | NA | NA | NA | NA |
| 266 | S-5836 | Epilepsy, Ataxia | *SCN9A* | Ex 27 | c.4962C>A | p.Asn1654Lys | MS | Het | AD | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 267 | R-0564 | Myopathy | *SDHA* | Ex 3 | c.205G>A | p.Ala69Thr | MS | Het | AR | NA | rs370481102 | RCV000234463.4 | <0.01 | B | B | D | D | D | D | NA | NA | NA | NA |
| Ex 9 | c.1177G>T | p.Val393Leu | MS | Het | AR | NA | rs372989971 | RCV000548710.1 | <0.01 | B | B | D | D | D | D | NA | NA | NA | NA |
| 268 | S-4839 | Microcephaly, ID | *SETBP1* | Ex 4 | c.2935G>A | p.Glu979Lys | MS | Het | AD | NA | rs373493544 | NA | <0.01 | D | D | D | B | D | B | NA | NA | NA | NA |
| 269 | S-1738 | Neuropathy | *SETX* | Ex 10 | c.1504C>T | p.Arg502Trp | MS | Hom | AR | NA | rs534723946 | RCV000143813.1 | <0.01 | B | B | B | B | D | D | NA | NA | NA | NA |
| 270 | S-2102 | ALS | *SETX* | Ex 10 | c.3347A>G | p.Asn1116Ser | MS | Het | AD | NA | rs148550755 | NA | <0.01 | D | B | B | B | B | B | NA | NA | NA | NA |
| 271 | S-3330 | Ataxia | *SETX* | Ex 10 | c.5102T>C | p.Phe1701Ser | MS | Hom | AR | NA | rs139309754 | NA | NA | D | B | D | D | D | D | NA | NA | NA | NA |
| 272 | S-3722 | Ataxia | *SETX* | Ex 19 | c.6530G>A | p.Cys2177Tyr | MS | Het | AR | In this study | NA | NA | NA | B | D | D | B | D | D | NA | NA | NA | NA |
| EX 26 | c.7361A>T | p.Asn2454Ile | MS | Het | AR | In this study | NA | NA | NA | D | B | D | D | D | B | NA | NA | NA | NA |
| 273 | S-1778 | Muscular dystrophy | *SGCA* | Ex 3 | c.197T>A | p.Leu66His | MS | Hom | AR | NA | rs767928766 | RCV000  384383 | <0.01 | D | B | D | D | D | D | NA | NA | NA | NA |
| 274 | S-4750 | Muscular dystrophy | *SGCB* | Ex3 | c.346A>G | p.Met116Val | MS | Hom | AR | NA | rs752168132 | RCV000  407437.1 | <0.01 | D | D | D | B | D | B | NA | NA | NA | NA |
| 275 | S-2583 | Muscular dystrophy | *SGCB* | Ex 3 | c.286G>C | p.Gly96Arg | MS | Het | AR | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| Ex 4 | c.544A>C | p.Thr182Pro | MS | Het | AR | NA | rs751427686 | RCV000  544134.1 | <0.01 | D | D | D | D | D | D | NA | NA | NA | NA |
| 276 | S-4766 | Muscular dystrophy | *SGCB* | Ex 4 | c.544A>C | p.Thr182Pro | MS | Hom | AR | NA | rs751427686 | RCV000  544134.1 | <0.01 | D | D | D | D | D | D | NA | NA | NA | NA |
| 277 | S-1704 | Muscular dystrophy | *SGCB* | Ex 5 | c.683G>A | p.Gly228Glu | MS | Hom | AR | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 278 | S-2985 | Dystonia | *SGCE* | Int 4 | c.463+5G>A | NA | SS | Het | AD | NA | rs1210401434 | NA | NA | NA | NA | NA | NA | NA | NA | D | D | D | NA |
| 279 | S-3198 | Muscular dystrophy | *SGCG* | Ex 3 | c.297G>A | p.Val99= | SS | Hom | AR | In this study | NA | NA | NA | NA | NA | NA | NA | NA | NA | D | D | D | NA |
| 280 | S-846 | Neuropathy | *SIGMAR1* | Ex 4 | c.446G>A | p.Gly149Glu | MS | Hom | AR | NA | rs546522200 | NA | NA | D | D | D | D | D | D | D | NA | D | NA |
| 281 | S-5807 | Leigh syndrome | *SLC25A19* | Ex 8 | c.869T>A | p.Leu290Gln | MS | Hom | AR | NA | rs750590533 | NA | <0.01 | D | B | D | D | D | D | NA | NA | NA | NA |
| 282 | S-4145 | Epilepsy | *SLC25A22* | Ex 8 | c.718G>A | p.Ala240Thr | MS | Hom | AR | NA | rs746087902 | NA | <0.01 | B | D | D | B | D | B | NA | NA | NA | NA |
| 283 | S-4971 | Myopathy | *SLC2A1* | Ex 2 | c.31C>T | p.Arg11Cys | MS | Het | AD | NA | rs1333609390 | NA | NA | D | B | D | B | B | B | NA | NA | NA | NA |
| 284 | S-993 | Epilepsy | *SLC2A1* | Ex 4 | c.338C>T | p.Ser113Leu | MS | Het | AD/AR | NA | rs774348625 | NA | <0.01 | B | D | D | D | B | D | NA | NA | NA | NA |
| 285 | R-1079 | Epilepsy | *SLC2A1* | Ex 7 | c.907G>T | p.Val303Leu | MS | Het | AD | \*20129935 | rs1205631854 | RCV000  520390.1 | NA | B | D | D | B | B | B | NA | NA | NA | NA |
| 286 | S-3016 | ASD | *SLC9A9* | Ex 6 | c.707T>C | p.Leu236Ser | MS | Het | Not known | NA | rs113649536 | NA | <0.01 | B | D | D | D | D | D | NA | NA | NA | NA |
| 287 | S-4850 | Epilepsy, Spasticity | *SOS1* | Ex 13 | c.2105A>G | p.Tyr702Cys | MS | Het | AD | NA | rs757094189 | RCV000  171288.1 | <0.01 | B | D | D | D | D | D | NA | NA | NA | NA |
| 288 | S-5230 | Epilepsy, Ataxia | *SOX10* | Ex 4 | c.698G>C | p.Gly233Ala | MS | Het | AD | In this study | NA | NA | NA | D | D | D | D | D | D | D | D | D | NA |
| 289 | S-4151 | Neuropathy | *SPG11* | Ex 30 | c.5398C>G | p.Gln1800Glu | MS | Het | AR | In this study | NA | NA | NA | B | B | D | D | B | B | NA | NA | NA | NA |
| Ex 30 | c.5582C>T | p.Pro1861Leu | MS | Het | AR | In this study | NA | NA | NA | B | D | D | D | D | D | NA | NA | NA | NA |
| 290 | S-4359 | Spasticity | *SPG7* | Ex 4 | c.614G>A | p.Arg205Gln | MS | Het | AR | NA | rs760639086 | NA | <0.01 | B | D | D | B | D | B | NA | NA | NA | NA |
| Ex 6 | c.853A>G | p.Ser285Gly | MS | Het | AR | NA | rs763745195 | NA | <0.01 | B | B | D | B | B | B | NA | NA | NA | NA |
| 291 | S-1058 | Neuropathy | *SPG7* | Ex 9 | c.1264T>C | p.Ser422Pro | MS | Het | AD/AR | In this study | NA | NA | NA | D | D | D | D | D | B | NA | NA | NA | NA |
| 292 | S-4405 | Epilepsy, Ataxia | *SPTAN1* | Ex 41 | c.5185C>G | p.Leu1729Val | MS | Het | AD | NA | rs764203560 | NA | <0.01 | B | D | D | D | D | D | NA | NA | NA | NA |
| 293 | S-3079 | Epilepsy | *SPTAN1* | Ex 44 | c.5701G>A | p.Glu1901Lys | MS | Het | AD | NA | rs748450616 | NA | <0.01 | B | D | D | B | D | B | NA | NA | NA | NA |
| 294 | S-2607 | Epilepsy | *SPTAN1* | Ex 57 | c.7415G>A | p.Arg2472His | MS | Het | AD | NA | rs375016371 | NA | NA | B | D | D | B | D | B | NA | NA | NA | NA |
| 295 | S-5649 | Ataxia | *SPTBN2* | Ex 17 | c.3591C>A | p.His1197Gln | MS | Het | AD | In this study | NA | NA | NA | B | D | D | D | D | B | NA | NA | NA | NA |
| 296 | S-5660 | Myopathy | *SPTLC1* | Ex 11 | c.1074G>C | p.Glu358Asp | MS | Het | AD | NA | NA | NA | <0.01 | D | D | D | B | B | B | NA | NA | NA | NA |
| 297 | S-3208 | Developmental delay | *SRCAP* | Ex 34 | c.9224G>A | p.Gly3075Glu | MS | Het | AD | NA | rs757045475 | RCV000  261862.1 | <0.01 | D | D | B | B | D | D | NA | NA | NA | NA |
| 298 | S-4228 | Epilepsy, ID | *ST3GAL3* | Ex 12 | c.1046C>T | p.Thr349Met | MS | Hom | AR | NA | rs1201878175 | NA | NA | B | D | D | B | D | D | NA | NA | NA | NA |
| 299 | S-5350 | Muscular dystrophy | *SYNE1* | Ex 47 | c.6889G>A | p.Glu2297Lys | MS | Het | AD | In this study | NA | NA | NA | B | D | D | B | D | B | NA | NA | NA | NA |
| 300 | S-5129 | Ataxia | *SYNE1* | Ex 59 | c.9455C>T | p.Ala3152Val | MS | Het | AR | In this study | NA | NA | NA | B | D | D | D | D | D | NA | NA | NA | NA |
| *SYNE1* | Ex 106 | c.19559A>G | p.Tyr6520Cys | MS | Het | AR | NA | rs759071467 | NA | <0.01 | D | D | D | D | D | D | NA | NA | NA | NA |
| 301 | S-5660 | Myopathy | *SYNE1* | Ex 110 | c.877C>T | p.Glu6748Lys | MS | Het | AD | NA | rs773788715 | NA | <0.01 | B | D | D | B | D | B | NA | NA | NA | NA |
| 302 | S-4296 | Muscular dystrophy, Myopathy | *SYNE2* | Ex 43 | c.6503G>A | p.Gly2168Asp | MS | Het | AD | In this study | NA | NA | NA | B | D | D | B | D | B | NA | NA | NA | NA |
| 303 | R-0109 | Spasticity | *TCF4* | Ex 11 | c.826A>T | p.Ser276Cys | MS | Het | AD | In this study | NA | NA | NA | B | D | D | B | D | D | NA | NA | NA | NA |
| 304 | S-2740 | ASD, ID | *TCF4* | Ex 12 | c.929G>A | p.Arg310Lys | MS | Het | AD | NA | rs1166273670 | NA | NA | B | D | D | D | D | D | NA | NA | NA | NA |
| 305 | S-2399 | Ataxia | *TGM6* | Ex 3 | c.332G>T | p.Arg111Leu | MS | Het | AD | In this study | NA | NA | NA | D | B | D | B | D | B | NA | NA | NA | NA |
| 306 | S-4400 | Ataxia, Dystonia | *TGM6* | Ex 7 | c.974C>T | p.Thr325Ile | MS | Het | AD | NA | rs141990961 | NA | NA | D | D | B | D | D | D | NA | NA | NA | NA |
| 307 | S-2482 | Dystonia | *TH* | Int 3 | c.406-3A>G | NA | SS | Het | AR | NA | rs747848885 | NA | <0.01 | NA | NA | NA | NA | NA | NA | D | D | D | NA |
| Ex 14 | c.1492G>A | p.Asp498Asn | MS | Het | AR | NA | rs200751977 | NA | <0.01 | D | D | D | B | D | D | NA | NA | NA | NA |
| 308 | S-5861 | Dystonia | *TH* | Ex 8 | c.919T>G | p.Ser307Ala | MS | Hom | AR | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 309 | S-3955 | Developmental delay | *TPM3* | Ex 1 | c.65G>A | p.Arg22Gln | MS | Het | AD/AR | NA | NA | RCV000  551766.1 | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 310 | S-5502 | Ataxia | *TPP1* | Int 2 | c.90-8C>G | NA | SS | Hom | AR | In this study | NA | NA | NA | NA | NA | NA | NA | NA | NA | D | D | D | NA |
| 311 | S-1904 | Epilepsy | *TPP1* | Ex10 | c.1222\_1224delAGT | p.Ser408del | Indel | Hom | AR | NA | rs1474804613 | NA | NA | NA | NA | D | NA | NA | NA | NA | NA | NA | D |
| 312 | S-5501 | Epilepsy, Leukodystrophy | *TREX1* | Ex 2 | c.137G>C | p.Ser46Thr | MS | Het | AD/AR | In this study | NA | NA | NA | D | B | D | B | B | B | NA | NA | NA | NA |
| 313 | S-4698 | Neuropathy | *TRPV4* | Ex 13 | c.1981C>T | p.Arg661Cys | MS | Hom | AD | NA | rs772074281 | NA | <0.01 | D | D | D | D | D | D | NA | NA | NA | NA |
| 314 | S-5831 | ASD | *TSC2* | Ex 34 | c.4351C>T | p.Arg1451Cys | MS | Het | AD | NA | rs369553241 | RCV000  465620 | <0.01 | D | B | D | B | D | D | NA | NA | NA | NA |
| 315 | S-4641 | ID | *TTC19* | Ex 10 | c.1104\_1108delAAAGA | p.Lys370Terfs | Indel | Hom | AR | NA | rs539447756. | NA | <0.01 | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA |
| 316 | R-0689 | Epilepsy | *TUBGCP6* | Ex 16 | c.3139C>T | p.Arg1047Trp | MS | Het | AR | NA | rs538652140 | NA | <0.01 | B | B | B | B | D | D | NA | NA | NA | NA |
| Ex 23 | c.5140G>A | p.Ala1714Thr | MS | Het | AR | NA | rs748135189 | NA | <0.01 | B | B | D | D | D | B | NA | NA | NA | NA |
| 317 | S-4323 | Epilepsy | *TUBGCP6* | Ex 21 | c.4697T>G | p.Leu1566Arg | MS | Het | AR | In this study | NA | NA | NA | B | B | D | D | D | D | NA | NA | NA | NA |
| Ex 22 | c.4890C>A | p.Phe1630Leu | MS | Het | AR | In this study | NA | NA | NA | B | D | D | D | D | B | NA | NA | NA | NA |
| 318 | S-193 | Developmental delay | *WDR62* | Ex 18 | c.2210G>C | p.Ser737Thr | MS | Hom | AR | In this study | NA | NA | NA | B | D | D | B | D | B | D | NA | D | NA |
| 319 | S-495 | Epilepsy | *ZEB2* | Ex 8 | c.2099A>G | p.Tyr700Cys | MS | Het | AD | In this study | NA | NA | NA | D | D | D | B | D | B | NA | NA | NA | NA |
| 320 | S-1668 | Spasticity | *ZFYVE26* | Ex 21 | c.3970T>A | p.Ser1324Thr | MS | Hom | AR | NA | rs776708469 | RCV000  380893.1 | <0.01 | B | B | D | B | B | B | NA | NA | NA | NA |
| 321 | S-1039 | Epilepsy, Microcephaly | *ZNF335* | Ex 9 | c.1459C>T | p.His487Tyr | MS | Hom | AR | In this study | NA | NA | NA | D | D | D | D | D | D | NA | NA | NA | NA |
| 322 | S-2953 | Epilepsy | *CHRNA7* | Ex 2-4 dup | c.(55+1\_561)\_(350+1\_351-1)dup | NA | Large dup | Het | AD | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA |
| 323 | S-3922 | ASD | *KIF1A* | Ex 16-42 dup | c.(1470+1\_14711)\_(4565+1\_4566-1)dup | NA | Large dup | Het | AD | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA |
| 324 | S-3739 | Epilepsy | *PRICKLE1* | Ex 2-8 del | c.(?\_-1)\_(\*1\_?)del | NA | Large del | Het | AR | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA |
| 325 | S-222 | Epilepsy | *WWOX* | Ex 6-8 dup | c.(516+1\_5171)\_(1056+1\_1057-1)dup | NA | Large dup | Inc | AR | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA |
| 326 | S-1887 | Epilepsy | chr2q24.3 | NA | NA | NA | Large del | Het | AD | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA |
| 327 | S-4476 | Myopathy | chr10q22.3 | NA | NA | NA | Large del | Het | AD | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA |
| 328 | S-3677 | ASD | chr16p13.11 | NA | NA | NA | Large del | Het | AD | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA |
| 329 | S-4388 | Dystonia | chr18p11.21 | NA | NA | NA | Large dup | Het | AD/AR | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA |
| 330 | S-5945 | Microcephaly | chr22q11.21 | NA | NA | NA | Large del | Het | AD | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA | NA |

**Abbreviations:**

AD-Autosomal dominant; ALS-Amyotrophic lateral sclerosis; AR-Autosomal recessive; ASD-Autism spectrum disorders; B-Benign; CdLS-Cornelia de Lange syndrome; CS-Clinical significance; D-Damaging; Del-Deletion; Dup-Duplication; Ex-Exon; ExAC- Exome Aggregation Consortium; FA-FATHMM (Functional analysis through Hidden Markov models); Hemi-Hemizygous; Het-Heterozygous; Hom-Homozygous; HSF-Human splicing finder; ID-Intellectual disability; Inc-Inconclusive; Indel-Small deletion/duplication/insertion or insertion/deletion; Int-Intron; LRT-Likelihood-ratio test; MA-Mutation assessor; MOI-Mode of Inheritance; MS-Missense; MT-Mutation taster; NN-NNSPLICE; NS-Nonsense; PP-PolyPhen; Pro-Provean; SIFT-Sorting intolerant from tolerant; SS-Splice site; VT-Variant type; XL-X-linked

\*: Reference from PubMed database (PMID), \*\*: Reported in genome Aggregation Database (gnomAD), $: ExAC allelic frequency in percentage

**Table S7: List of ‘pathogenic’/’likely pathogenic’ variants identified in the** **ACMG (American College of Medical Genetics and Genomics) recommended genes as secondary or incidental findings**: 12 of the 1012 patients had secondary findings in a total of 11 genes; *ACTC1, ATP7B, BRCA2, DSG2, KCNQ1, LDLR, MEN1, MYBPC3, NF2, RYR1,* and *TMEM43.*

|  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Sl.No.** | **Sample id** | **Gene** | **Disease associated** | **Nucleotide** | **Protein** | **VT** | **Zygosity** | **MOI** | **Reference** |
| 1 | S-80 | *ACTC1* | Familial HCM 11 | c.107\_120delinsC | p.Ile36ThrfsTer17 | Indel | Het | AD | In this study |
| 2 | S-4266 | *ATP7B* | Wilson disease | c.813C>A | p.Cys271Ter | NS | Hom | AR | ΔRCV000169298.1 |
| 3 | S-4320 | *BRCA2* | Breast-ovarian cancer, familial 2 | c.4695\_4698dupGACC | p.Leu1567AspfsTer9 | Indel | Het | AD | \*17503080 |
| 4 | S-2397 | *DSG2* | ARVC, type 10 | c.1021delG | p.Asp341IlefsTer5 | Indel | Het | AD | In this study |
| 5 | S-4014 | *KCNQ1* | Long QT syndrome 1 | c.613G>A | p.Val205Met | MS | Het | AD | \*18580685 |
| 6 | S-4698 | *KCNQ1* | Long QT syndrome 1 | c.1552C>T | p.Arg518Ter | NS | Het | AD | \*10482963 |
| 7 | S-1553 | *LDLR* | Familial hypercholesterolemia | c.1060+2T>G | NA | SS | Het | AD | \*27765764 |
| 8 | S-5116 | *MEN1* | Multiple endocrine neoplasia, type 1 | c.1350+1G>A | NA | SS | Het | AD | \*15292357 |
| 9 | S-5257 | *MYBPC3* | DCM 1A and Familial HCM 4 | c.383\_384insGCCCCAAGCTG | p.Glu129ProfsTer34 | Indel | Het | AD | In this study |
| 10 | S-1143 | *NF2* | Neurofibromatosis, type 2 | c.1021C>T | p.Arg341Ter | NS | Het | AD | \*7913580 |
| 11 | S-334 | *RYR1* | Malignant hyperthermia | c.10825-2A>T | NA | SS | Het | AD | In this study |
| 12 | S-3945 | *TMEM43* | ARVC, type 5 | c.316\_317delTA | p.Tyr106TrpfsTer22 | Indel | Het | AD | In this study |

**Abbreviations:**

AD-Autosomal dominant; AR-Autosomal recessive; ARVC-Arrhythmogenic right ventricular cardiomyopathy; DCM-Dilated cardiomyopathy; HCM-Hypertrophic cardiomyopathy; Het-Heterozygous; Hom-Homozygous; Indel-Small deletion/duplication/insertion or insertion/deletion; MOI-Mode of Inheritance; MS- Missense; NS-Nonsense; SS-Splice site; VT- Variant type

Δ: ClinVar database ID, \*: Reference from PubMed database (PMID)

**Table S8: List of cases with multiple phenotypes, wherein a confirmed diagnosis was arrived at with multi-gene panel testing.**

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **Sl. No.** | **Sample ID** | **Clinical indications provided** | **Gene/**  **Genomic region** | **Mutation identified** | **Disease associated** |
| 1 | S- 2593 | Progressive dystonia, difficulty in swallowing, slurring of speech, difficulty in walking, bilateral hypertonia, brisk tendon reflexes and bilateral ankle clonus. | *ALS2* | p.Leu294CysfsTer4 | Amyotrophic lateral sclerosis 2, juvenile |
| 2 | S- 3469 | Global developmental delay, microcephaly, intellectual disability, intrauterine growth retardation, prominent eye, preauricular skin tag in right ear and seizure disorder. | *AP4M1* | p.Ser301LeufsTer15;  c.59-1G>C | Spastic paraplegia 50 |
| 3 | S- 6032 | Microcephaly, generalized seizures, hypomyelination, global developmental delay, hyperglycemia, hypoinsulinema, respiratory distress and succumbed on the 65th day. | *ASNS* | c.1321-2A>G | Asparagine synthetase deficiency |
| 4 | S- 2993 | Encephalopathy, global developmental delay, hypertonia, delayed speech, cerebral palsy, lethargy, round facies and seizures. | *ATP1A3* | p.Gly947Arg | Alternating hemiplegia of childhood |
| 5 | S- 590 | Refractory left focal seizures and global developmental delay. | *ATP1A3* | p.Asp801Asn | Alternating hemiplegia of childhood |
| 6 | S- 3386 | Global developmental delay and dystonia. | *ATP8A2* | c.3019-1G>A | Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4 (CAMRQ4) |
| 7 | S- 3353 | Visual disturbance, frequent falls, progressive gait difficulty, oculomotor apraxia, spasticity, cognitive deficit and peripheral neuropathy. | *C12orf65* | p.Val116Ter | Spastic paraplegia 55 and combined oxidative phosphorylation deficiency 7 |
| 8 | S- 3597 | Generalised dystonia, behavioural issues, anger outbursts and progressive pancerebellar features. | *CAPN3* | p.Arg748Gln | Limb-girdle muscular dystrophy, type 2A |
| 9 | S- 2936 | Developmental delay, microcephaly and Dandy-Walker malformations | *CASK* | p.Arg27Ter | Microcephaly with pontine and cerebellar hypoplasia (MICPCH) |
| 10 | S- 4099 | Thalamic hyperintensity, hypotonia, recurrent flaccid weakness triggered by fever, inability to move limbs, areflexia, gaze palsy and recurrent neuropathy. | *CD59* | p.Val8SerfsTer72; p.Asp74del | Hemolytic anemia with or without immune-mediated polyneuropathy |
| 11 | S- 705 | Muscle weakness, abnormal eye movements, drooping eyelids and thinning of facial muscles. | *CHRNE* | p.Glu443Lysfs | Congenital myasthenic syndrome |
| 12 | S- 4681 | Pseudohypertrophy with recent onset weakness, muscle hypertrophy, loss of fat pads and short stature. | *CLCN1* | p.Arg377Ter | Myotonia congenita |
| 13 | S- 4390 | Regression of milestones, myoclonic jerk, progressive visual blurring and vision loss. | *CLN5* | p.Arg199Ter | Neuronal ceroid lipofuscinoses |
| 14 | S- 5622 | Developmental delay, spasticity, speech and cognitive deficit and periventricular white matter loss with intact subcortical U-fibres suggestive of hypoxic-ischemic encephalopathy (HIE). | *COL4A1* | p.Gln113Ter | *COL4A1*-related disorders |
| 15 | S- 4466 | Motor delay and muscle weakness. | *COL6A1* | p.Gly293Arg | Ullrich congenital muscular dystrophy and Bethlem myopathy |
| 16 | S- 1176 | Myopathic face, high arched groove palate, flexon deformity, hyperextensibility of fingers, prominent fetal finger pads, restriction of shoulder joints, narrow chest, proximal contracture. | *COL6A2* | p.Asp428AlafsTer120;  p.Asp955GlufsTer37 | Ullrich congenital muscular dystrophy |
| 17 | S- 5445 | Muscle weakness and periodic paralysis. | *COQ2* | p.Leu395PhefsTer24 | Susceptibility to multiple system atrophy |
| 18 | S- 6075 | Krabbe disease and leukodystrophy. | *CTC1* | p.Ser353ProfsTer10; p.Glu558Gly | Cerebroretinal microangiopathy with calcifications and cysts |
| 19 | S- 2427 | Encephalopathy, seizures with neuroregression, global developmental delay, autistic features and mild dysmorphism. | *CUL4B* | p.Ile336LysfsTer2 | X-linked mental retardation |
| 20 | S- 2300 | Developmental delay, seizures, ptosis, posteriorly rotated ears, hypotonia, agyria, pachygyria and lissencephaly. | *DCX* | p.Gly223Arg | Lissencephaly |
| 21 | S- 4863 | Mild motor delay, toe walking and calf muscle hypertrophy. | *DMD* | p.Thr565AsnfsTer2 | Duchenne muscular dystrophy |
| 22 | S- 1126 | Global developmental delay, neuroregression, spasticity with bipyramidal and extrapyramidal involvement, microcephaly and subtle dysmorphism. | *DNAJC6* | p.Arg250Ter | Parkinson disease 19A |
| 23 | S- 4952 | Failure to thrive, developmental delay, clinodactyly, macroglossia, hypotonia, seizures, dolichocephaly, microcephaly, low set ears and high arched palate. | *DOK7* | p.Ala378SerfsTer30 | Congenital myasthenic syndrome |
| 24 | R- 1349 | Neuroregression, frequent falls, slurred speech, cognitive decline, drooling and mild cerebral atrophy. | *ENAM* | p.Pro422ValfsTer27 | Amelogenesis imperfecta |
| 25 | S- 4707 | Abnormal gait, spasticity, cognitive deficit, intellectual disability, microcephaly and short stature. | *ERCC6* | p.Thr513AspfsTer4;  c.4063-1G>C | Cockayne syndrome |
| 26 | S- 3631 | Developmental delay, cerebral atrophy and pigmentation changes in the retinal fundus. | *ERCC8* | p.Val318PhefsTer10; c.173+3\_173+6delAAGT | Cockayne syndrome A |
| 27 | S- 1614 | Drooping neck, upper limb hypotonia, brisk reflex in lower limb, delay in walking, motor and language delay, recurrent respiratory tract infection and white matter volume loss. | *EXOSC3* | p.Asp132Ala | Pontocerebellar hypoplasia, type 1B |
| 28 | S- 3714 | Floppiness, global developmental delay, brisk deep tendon reflexes, head lag, plagiocephaly, bilateral subdural hydroma and hypoxic ischemic encephalopathy. | *GAA* | p.Gly643Arg;  p.Thr286Met | Glycogen storage disease II |
| 29 | S- 5083 | Autism spectrum disorder and neuropathies. | *GDAP1* | p.Trp268GlyfsTer22 | Charcot-Marie-Tooth disease |
| 30 | S- 4657 | Developmental arrest and regression, exaggerated startle response, hypotonia, spasticity, and cherry red spot. | *GM2A* | c.244-2A>G | GM2-gangliosidosis |
| 31 | S- 1191 | Weakness in right leg since 1st pregnancy, writing problem, difficulty breathing while walking and talking, sleep difficulties, weakness around finger tips, clawing of toes, pes cavus, thinning of forearms since 10 years. | *GNE* | p.Ile587Thr | Inclusion body myopathy |
| 32 | S- 1675 | Recurrent falls, foot drop and leg weakness that moved from the ankles up to the hips, hands, shoulders and biceps, difficulty in walking and lifting things. | *GNE* | p.Val696Met;  p.His552Arg | Inclusion body myopathy |
| 33 | S- 5444 | Macrocephaly, gait imbalance and Arnold-Chiari malformation with skeletal features of mucopolysaccharidoses. | *GNPTG* | p.Phe65SerfsTer19 | Mucolipidosis III gamma |
| 34 | S- 760 | Global developmental delay, seizures, shock sepsis, metabolic aciduria, hypoglycemia, anemia, thrombocytopenia, nephrocalcinosis and hypervitaminosis. | *GRIN2A* | p.Ala922LeufsTer20 | *GRIN2A*-related speech disorders and epilepsy |
| 35 | S- 5250 | Febrile encephalopathy, neuroregression, seizures, cherry red spot, hepatomegaly, macrocephaly and coarse facies. | *HEXA* | p.Asp322Tyr | Tay-Sachs disease |
| 36 | S- 5286 | Febrile seizures, delayed motor development, failure to thrive, milestone regression. | *HEXA* | p.Glu482Lys | Tay-Sachs disease |
| 37 | S- 37 | Global developmental delay, hypertonia, cerebral atrophy, encephalopthy, coarse facies, behavioral abnormalities, deaf mutism and stereotypic movements. | *HGSNAT* | p.Trp340Ter | Mucopolysaccharidosis, type IIIC |
| 38 | S- 1643 | Epileptic encephalopathy, seizures since 2nd day of birth and delayed milestones. | *HPD* | p.Ile335Met;  p.Phe133Ser | Tyrosinemia, type III |
| 39 | S- 2482 | *TOR1A*-related dystonia. | *HPRT1* | c.532+5G>A | Lesch-Nyhan syndrome |
| 40 | S- 1375 | Rett syndrome and autism spectrum disorders. | *KIAA2022* | p.Gly917Ter | X-linked intellectual disability |
| 41 | S- 3449 | Regression since early infancy, intellectual disability, impaired vision, myoclonic epilepsy, spastic quadriparesis and cerebellar atrophy. | *KIF1A* | p.Thr99Met | Mental retardation, autosomal dominant 9 |
| 42 | S- 4873 | Plagiocephaly, hypertelorism, depressed nasal bridge, midfacial hypoplasia and adducted thumb. | *L1CAM* | p.Arg760Ter | L1 syndrome |
| 43 | S- 5585 | Global developmental delay with regression, epilepsy and dystonic movements. | *MECP2* | p.Arg270Ter | Rett syndrome and neonatal encephalopathy. |
| 44 | S- 2801 | Autistic regression, epilepsy and microcephaly. | *MECP2* | p.Pro217TrpfsTer18 | Rett syndrome |
| 45 | S- 4299 | Microcephaly, limb length discrepancy, ataxia, spasticity, flappy hands, delayed walking, cognitive delay, delayed and bisyllabic speech, laughter spells, inward strabismus, no eye contact, hyperactivity, repetitive hand clapping and absence seizures. | *MECP2* | p.Pro152Arg | Rett syndrome |
| 46 | S- 4628 | Infantile spasms, global developmental delay, focal myoclonic jerks and MRI showed cerebellar polymicrogyria. | *MFN2* | p.Arg364Trp | Charcot-Marie-Tooth disease, type 2 |
| 47 | S- 5406 | Global developmental delay, seizures, spasticity, tremors, pancytopenia, optic atrophy, alopecia, hyperpigmentation and recurrent respiratory tract infection. | *MMACHC* | p.Arg132Ter | Methylmalonic acidemia with homocystinuria. |
| 48 | S- 6030 | Broad facies, nose abnormality, downslanting palpebral fissure, breathing and swallowing difficulties, large ears and failure to thrive. | *MTHFR* | p.Trp421Ser | Homocystinuria |
| 49 | S- 3769 | Generalised hypotonia, frog leg position, pooling of secretions, myoclonic dystrophy, myopathy and myasthenia gravis. | *MTM1* | p.Arg474Ter | X-linked myotubular myopathy |
| 50 | S- 329 | Retinitis pigmentosa, progressive cerebellar and sensory ataxia, tremors and malabsorption with recurrent diarrhea. | *MTTP* | p.Ser365LeufsTer8 | Abetalipoproteinemia |
| 51 | S- 4525 | Bilateral lower and upper limb weakness, slow extraocular movement and bilateral restricted abduction. | *MUSK* | p.Asn103Ser | Congenital myasthenic syndrome |
| 52 | S- 4875 | Congenital myasthenia, myopathy, mitochondrial disorders and alternating esotropia. | *MYH2* | p.Arg793Ter | Inclusion body myopathy 3 |
| 53 | S- 1310 | Episodic vomiting, headache, irritability, global developmental delay and ataxia. | *NDUFV2* | p.Arg143Ter;  p.Thr232Pro | Mitochondrial complex I deficiency |
| 54 | S- 3171 | Complex movement disorder involving motor delay, hypotonia, ataxia, myoclonus and choreoathetosis. | *NKX2-1* | p.Ser217Ter | Benign hereditary chorea |
| 55 | S- 2000 | Difficulty in walking, slow response, ataxia, poor scholastic performance, extensor plantar reflex along with increased tone and reflex. | *NPC1* | c.2604+1G>A;  p.Ile690Phe | Niemann-Pick disease type C |
| 56 | S- 5459 | Recurrent fracture of long bones, decreased or absent sweating and insensitivity to pain. | *NTRK1* | p.Val135AlafsTer37; p.Arg596Gln | Congenital insensitivity to pain with anhidrosis |
| 57 | S- 3192 | Global developmental delay, congenital cataract, hypotonia, umbilical hernia, failure to thrive, vitamin D deficiency and seizures. | *OCRL* | p.Phe276Ser | Lowe syndrome |
| 58 | S- 5835 | Encephalopathy, seizures, spasticity, motor delay, global developmental delay, failure to thrive, microcephaly, septicemia, lactic acidemia and oligohydramnios at 7th month of gestation. | *OSTM1* | p.Asn173IlefsTer2 | Osteopetrosis, autosomal recessive 5 |
| 59 | S- 4691 | Cerebellar atrophy, generalised seizures, global developmental delay, microcephaly, tremors, cognitive deficit, delayed speech and intellectual disability. | *PCDH19* | p.Glu32Ter | Epileptic encephalopathy, early infantile, 9 |
| 60 | S- 4592 | Cerebellar ataxia, repetitive eye movements, developmental delay, and had trouble eating and performing fine motor movements. | *PDHA1* | p.Tyr161Tyr | Pyruvate dehydrogenase deficiency |
| 61 | S- 4155 | Developmental delay, motor regression, ataxia, nystagmus, cerebellar atrophy and generalised motor neuronal axonopathy. | *PLA2G6* | p.Glu667Ter | Infantile neuroaxonal dystrophy |
| 62 | S- 2562 | Microcephaly, global developmental delay, seizures and dysmorphism. | *PNKP* | c.936+1G>T;  p.Leu12Pro | Microcephaly, seizures and developmental delay |
| 63 | S- 1439 | Mitochondrial cytopathy, basal ganglia abnormality, cerebral atrophy, encephalopathy, generalized seizures, epilepsy, motor delay, failure to thrive, regression of milestones since 15 months of age, hypotonia, attention and cognitive deficit. | *POLG* | p.Leu304Arg | POLG deficiency |
| 64 | S- 2690 | Bilateral congenital cataract, stiffness on both legs and toe walking. | *RAB3GAP2* | p.Tyr646Terfs | Warburg micro syndrome 2 |
| 65 | S- 4788 | Global developmental delay and regression. | *RNASEH2A* | p.Arg108Trp | Aicardi-Goutieres syndrome, type 4 |
| 66 | S- 1722 | Global developmental delay, intermittent ataxia, seizures and autistic features. | *SCN2A* | p.Thr1491GlnfsTer11 | Early infantile epileptic encephalopathy 11 and benign familial infantile seizures 3 |
| 67 | S- 1372 | Mild motor delay, kyphoscoliosis, hypotonia, hyperreflexia and proximal muscle weakness. | *SEPN1* | c.1282-2A>C;  p.Ala276\_Cys277insSer | *SEPN1*-related myopathy |
| 68 | S- 722 | Neuropathy, sensory ataxia, proximal muscle weakness, tremors, delayed speech and intellectual disability. | *SLC12A6* | p.Ala886CysfsTer9 | Hereditary motor and sensory neuropathy with agenesis of the corpus callosum (Andermann syndrome) |
| 69 | S- 2432 | Cerebral atrophy and global developmental delay. | *SLC16A2* | p.Trp324Ter | Allan-Herndon-Dudley syndrome |
| 70 | S- 3475 | Encephalopathy, seizures, microcephaly, lethargy and elevated levels of methylmalonic acid (MMA) and hyperintensities in putamen and swollen thalamus. | *SLC19A3* | p.Asp2IlefsTer7 | Biotin-thiamine-responsive basal ganglia disease |
| 71 | S- 3778 | Congenital myopathy, hypotonia, motor development delay and floppy infant. | *SLC22A5* | p.Arg399Trp;  p.Arg336Gln | Systemic primary carnitine deficiency |
| 72 | S- 3173 | Global developmental delay, seizures, hypoplasia of corpus callosum, coarse features, hirsuitism, dysmorphism, rocker bottom feet and hypospadias. | *SLC25A22* | c.202+1dupG | Epileptic encephalopathy, early infantile, 3 |
| 73 | S- 4145 | Generalized seizures, global developmental delay, hypotonia, dyskinesia, cognitive deficit and speech delay. | *SLC25A22* | c.818+1G>C;  p.Ala240Thr | Epileptic encephalopathy, early infantile, 3 |
| 74 | S- 4302 | Episodic ataxia, absence epilepsy, reduced concentration, restlessness and a decline in academic performance. | *SLC2A1* | p.Ile369HisfsTer9 | GLUT1 deficiency syndrome |
| 75 | S- 1891 | Epilepsy, microcephaly, delayed motor and cognitive development, axial hypotonia, episodes of nystagmus and hypomyelination. | *SLC2A1* | p.Leu185CysfsTer6 | GLUT1 deficiency syndrome |
| 76 | S- 3231 | Microcephaly, global developmental delay, episodic ataxia, intellectual disability, brisk tendon reflexes and coarse facial features. | *SLC2A1* | p.Gln282SerfsTer58 | GLUT1 deficiency syndrome and dystonia |
| 77 | S- 5628 | Seizures, spastic quadriplegia, walking and speech difficulties, vacant and unresponsive stare, episodes of excessive sleep and delayed developmental milestones. | *SLC2A1* | p.Trp65Ter | GLUT1 deficiency syndrome |
| 78 | S- 5036 | Global developmental delay, semilobar holoprosencephaly with colpocephaly. | *SMC1A* | p.Lys798AsnfsTer31 | Cornelia de Lange syndrome, type 2 |
| 79 | S- 5854 | Global developmental delay, spastic paraparesis, positive cerebellar signs, peeling of skin and hypomyelination. | *SNAP29* | p.Ser163LysfsTer6; p.Arg196Ter | Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome |
| 80 | S- 4250 | Dysferlinopathy and muscular dystrophy. | *SPG11* | p.Gln716Ter | Spastic paraplegia type 11 |
| 81 | S- 2368 | Manifested ataxia, abnormal gait, hypotonia, clumsiness of all four limbs, recurrent falls, opthalmoplegia, abnormal eye movements, fundus albipunctatus, lactic acidemia and pancerebellar signs. | *SQSTM1* | p.Ser275PhefsTer17 | Neurodegeneration with ataxia, dystonia, and gaze palsy |
| 82 | S- 3458 | Encephalopathy, generalized seizures since day4 of life. | *STXBP1* | p.Pro480Leu | Epileptic encephalopathy |
| 83 | S- 3181 | Floppiness and inability to walk. | *SURF1* | p.Arg179LysfsTer12 | Leigh syndrome |
| 84 | S- 3233 | Hypotonia, developmental delay, facial dysmorphism, decreased reflexes and bilateral cerebellar subcortical white matter with bilateral caudate lentiform. | *SURF1* | p.Gln251Ter | Leigh syndrome |
| 85 | S- 1331 | Myoclonus seizures, autistic symptoms and global developmental delay. | *SYNGAP1* | p.Ile827GlyfsTer24 | Mental retardation, autosomal dominant 5 |
| 86 | S- 2171 | Global developmental delay with autistic features, bruxism, depressed nasal bridge, bulbous nose, self harming at night and excessive cry. | *SYNGAP1* | p.Cys531HisfsTer17 | Mental retardation, autosomal dominant 5 |
| 87 | R- 0840 | Developmental delay, poor cognition, coarse face, macrostomia, hand flapping movement, divergent squint and prominent upper lip. | *TCF4* | c.550-2A>C | Pitt-Hopkins syndrome |
| 88 | S- 5111 | Congenital muscular dystrophy. | *TK2* | p.Ala139Val | Mitochondrial DNA depletion syndrome 2, myopathic type |
| 89 | S- 2314 | Motor delay, proximal muscle weakness, dysmorphism and chest deformity. | *TNNT1* | c.750+1G>A | Nemaline myopathy type 5 |
| 90 | S- 3156 | Global developmental delay, hypomyelination, spasticity, hypotonia, flat facies and has a history of old fractures. | *TNNT1* | c.32+1G>A | Nemaline myopathy type 5 |
| 91 | S- 3161 | Delayed motor development, microcephaly, hypotonia, abnormal gait, and ataxia. | *UBE3A* | p.Gln494AlafsTer11 | Angelman syndrome |
| 92 | S- 569 | Bilateral foot drop, high stepping gait, difficulty in getting up from squatting position and weak bilateral interosseous nerve, suggesting axonal neuropathy. | *VRK1* | c.1159+1G>A | Hereditary motor and sensory neuropathy |
| 93 | S- 96 | Seizures, slurred speech and progressive abnormal behavior suggestive of NBIA. | *WDR45* | p.Tyr336CysfsTer5 | Neurodegeneration with brain iron accumulation 5 |
| 94 | S- 2563 | Microcephaly, global developmental delay and profound mental retardation. | *WDR62* | c.1959-2A>G | Microcephaly 2, with or without cortical malformations (MCPH2) |
| 95 | S- 6581 | Muscle weakness, hypotonia, developmental delay, abnormal jerky movements, poor visual fixation, hyperekplexia, poor head control, diffused cerebral atrophy and white matter hyperintensity. | *UNC80* | p.Leu2739GlnfsTer22 | Infantile hypotonia with psychomotor retardation and characteristic facies-2 |
| 96 | S- 6113 | Autistic behaviour and epilepsy. | *NRXN1* | c.(919+1\_9201)\_(3484+1\_3485-1)del | Autism spectrum disorders and epilepsy |
| 97 | S- 3831 | Delayed motor development, hypotonia and muscle weakness. | *ATP8A2* | c.(2211+1\_2212-1)\_(3183+1\_3184-1)del | Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4 |
| 98 | S- 4670 | Cognitive deficiency, global developmental delay, delayed speech, intellectual disability, broad facies and obesity. | *BCL11A* | c.(?\_-30)\_(\*28\_?)del | Intellectual developmental disorder with persistence of fetal hemoglobin |
| 99 | S- 4663 | Seizures, progressive visual blurring and cognitive impairment. | *CLN3* | c.(790+1\_7911)\_(1056+1\_1057-1)del | Neuronal ceroid lipofuscinosis |
| 100 | R- 0109 | Spastic diplegia, global developmental delay, gastro-esophageal reflux, esotropia and adducted thumbs. | *CYB5R3* | c.334-733del | Methemoglobinemia |
| 101 | S- 4491 | Microcephaly, myoclonic seizures, spastic quadraplegia and tonic posturing. | *GALC* | c.(1161+1\_1162-1)\_(\*31\_?)  del | Krabbe disease |
| 102 | S- 1035 | Encephalopathy, seizures at two months of age, developed pneumonia and sensory neuropathy. | *IGHMBP2* | c.(1060+1\_1061-1)\_(\*1\_?)del | Charcot-Marie-Tooth neuropathy, type 2 (CMT2) and spinal muscular atrophy with respiratory distress, type 1 |
| 103 | R- 0536 | Recurrent seizures, lethargy, choreic movements and regression in milestones. | *ROGDI* | c.432+462\_\*145delinsCAG | Mucopolysaccharidosis, type IIIB |
| 104 | S- 3157 | Attention deficit, intellectual disability, global development delay, broad facies, myopia, macrocephaly and macrostomia. | *TRIP12* | c.(?\_-1)\_(3990+1\_3991-1)dup | Mental retardation, autosomal dominant 49 |
| 105 | S- 3882 | Cerebral and cerebellar atrophy, cerebral palsy, spasticity, global developmental delay, hypotonia, microcephaly, long eye lashes and plagiocephaly. | *TSEN2* | c.(?\_-1)\_(189+1\_190-1)del;  p.Ile384Phe | Pontocerebellar hypoplasia, type 2 |
| 106 | S- 4436 | Attention difficulty, head hitting when angry, strabismus, difficulty in walking, no eye contact and does not recognize family members. | *TUSC3* | c.(308+1\_309-1)\_(\*1\_?)del | Mental retardation, autosomal recessive 7 |
| 107 | S- 2889 | Febrile seizures, global developmental delay, microcephaly, short stature, spasticity, attention deficit, autism, delayed speech, hyperpigmentation and curly hair with abnormality of the hairline. | chr1p36.31-p36.22 | Large deletion | 1p36 deletion syndrome |
| 108 | S- 2857 | Refractory epilepsy and had deep set eyes, no head control and straight eyebrows. | chr1p36.33-p36.23 | Large deletion | 1p36 deletion syndrome |
| 109 | S- 5116 | Epilepsy. | chr1q43-q44 | Large deletion | 1q43-q44 deletion syndrome |
| 110 | S- 594 | Intestinal malrotation, feeding difficulties, failure to thrive, global developmental delay, unclear speech, subnormal intelligence, seizure, hearing loss, obesity, hypertelorism, thin lips and hyperextensible phalangeal joints. | chr2q37.1-q37.3 | Large deletion | 2q37 microdeletion syndrome |
| 111 | S- 5221 | Macrocephaly, speech delay, hypomyelination, hypertelorism, hypotonia and intermittent involuntary eye movements. | chr4q21.21-q22.3 | Large deletion | 4q21-q22 deletion syndrome |
| 112 | S- 4861 | Global developmental delay, infantile spasms, reduced visual acuity, mild autistic features and undescended testis. | chr5q14.3 | Large deletion | 5q14.3-q15 microdeletion syndrome |
| 113 | S- 5188 | Global developmental delay, dysmorphism, vision and hearing difficulty, axial hypotonia, macrocephaly and autistic features. | chr5q14.3-q15 | Large deletion | 5q14.3 microdeletion syndrome |
| 114 | S- 582 | Global developmental delay, hypotonia with exaggerated reflexes, macrocephaly, dysmorphic facies and hyperpigmentation. | chr5q35.2-q35.3 | Large deletion | Sotos syndrome |
| 115 | S- 5411 | Microcephaly and global developmental delay. | chr6q27 | Large deletion | 6q27 deletion syndrome |
| 116 | S- 3030 | Mild developmental delay, trichorrhexis nodosa, facial features and microcephaly. | chr7p14.1 | Large deletion | Trichothiodystrophy |
| 117 | S- 5204 | Developmental delay, dysmorphism, hyperactivity, brachycephaly, macrocephaly, synophrys, long eye lashes, hirsuitism, triangular philtrum and thin upper lip. | chr7q11.23 | Large duplication | 7q11.23 duplication syndrome |
| 118 | S- 1592 | Attention-deficit/hyperactivity disorderwith subnormal social functioning, large ears, broad facies, prominent philtrum, hypotonia and macrostomia. | chr7q31.2-q31.32 | Large deletion | chr7q31 deletions have been shown to be associated with speech and language impairment, oromotor dyspraxia, dysmorphism, developmental delay and behavioural problems |
| 119 | S- 4962 | Kabuki syndrome. | chr8p23.3-p23.1 | Large deletion | 8p23 deletion syndrome |
| 120 | S- 4289 | Hypotonia, bitemporal hollowing, open mouth appearance, frog leg position, metopic ridging, low set ears, microretrognathia and absence of deep tendon reflexes. | chr15q11.2-q13.1 | Large deletion | Angelman syndrome and Prader-Willi syndrome |
| 121 | S- 177 | Global developmental delay, failure to thrive, seizures, hypotonia, agenesis of corpus callosum, microcephaly, choanal atresia, hypothyroidism, anemia, facial dysmorphism, anocutaneous fistula and semilobular holoprosencephaly. | chr15q24.1-q25.1 | Large deletion | 15q24 microdeletion syndrome |
| 122 | S- 3888 | Delayed motor development, global developmental delay, microcephaly, flat facies, frontal bossing, hypertelorism, intellectual disability and hyperactivity. | chr17p11.2 | Large deletion | Smith-Magenis syndrome |
| 123 | S- 5648 | Global developmental delay, infantile spasms and agyria-pachygyria complex | chr17p13.3-p13.2 | Large deletion | Miller-Dieker syndrome (chr17p13.3 del syndrome) |
| 124 | S- 3191 | Delayed milestones, delayed speech, hypotonia, mild dysmorphism and polyminimyoclonus. | chr18q12.1-q12.2 | Large deletion | 18q deletion syndrome |
| 125 | S- 3206 | Global developmental delay, hypertelorism, broad frontal region, short nose and protruding tongue. | chr19p13.3 | Large deletion | 19p13.3 microdeletion syndrome |
| 126 | S- 338 | Facial dysmorphism, agenesis of corpus callosum, seizures, hypotonia, developmental delay, macrocephaly, limb malformation, abnormal gait and congenital heart defect, hypospadias. | chr22q13.31-q13.33 | Large deletion | 22q13 deletion syndrome, also known as Phelan–McDermid syndrome |
| 127 | S- 4467 | Focal seizures, tonic stiffening and global developmental delay. | chrXp22.13 | Large deletion | X-linked infantile spasm syndrome |
| 128 | S- 4223 | Developmental delay and nystagmus. | chrXq22.1-q22.2 | Large duplication | Xq22.1 duplication syndrome |