**Supplementary Table S1:** List of whole exome sequencing derived candidate variants remaining after filtering

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| Gene | Genotype | Consequence | HGVSc | HGVSp | dbSNP ID |
| *VPS13D* | het | synonymous\_variant | NM\_015378.2:c.2583A>G | NM\_015378.2:c.2583A>G(p.=) |  |
| *NPHP3-ACAD11, UBA5* | het | upstream\_gene\_variant |  |  | rs770749604 |
| *PEX6* | het | downstream\_gene\_variant |  |  | rs368299692 |
| *SPAST* | het | intron\_variant | NM\_014946.3:c.415+48delG |  |  |
| *MTPAP* | het | splice\_region\_variant,intron\_variant | NM\_018109.3:c.1313-3C>T |  | rs201064853 |
| *MACROD1, FLRT1* | het | intron\_variant | NM\_014067.3:c.517+34934G>C |  | rs139768227 |
| *C12ORF65* | het | synonymous\_variant | NM\_152269.4:c.243C>T | NM\_152269.4:c.243C>T(p.=) | rs140411575 |
| *C12ORF65* | het | missense\_variant | NM\_152269.4:c.413A>G | NP\_689482.1:p.Lys138Arg | rs147328685 |
| *SPG7* | **hom** | **missense\_variant** | **NM\_003119.2:c.1763C>T** | **NP\_003110.1:p.Thr588Met** |  |