**Supplemental Table 1. Post-filtering candidate heterozygous variants shared by exomes of two affected subjects in Family A.**

|  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Gene** | **Chr** | **Position (gr37)** | **cDNA** | **Protein** | **Freq. in gnomAD** | **Impact** | **Known Disease****Assoc** | **Co-segreg** |
| STUB1 | 16 | 730683 | c.158T>C | p.Ile53Thr | absent | deleterious | SCAR16 | yes |
| RASA4 | 7 | 102236221 | c.859\_865delCCAGGGC | p.Pro287fs | absent | frame-shift | **–** | no |
| CRIPAK | 4 | 1388350 | c.78\_79insCACATGCCCATGTGGAGTGCCCGCCTGCTCA | p.Cys27fs | absent | frame-shift | **–** | no |
| CRB2 | 9 | 126135887 | c.3089\_3104dupGGCCCGGCGCGGCCCC | p.Gly1036fs | absent | frame-shift | ventriculo-megaly | no |
| ATXN3 | 14 | 92537354 | c.915\_916insCAG13 | p.Gln293-Gln305dup | 0.0000198 | in-frame | SCA3 | no |