

| GENE | GENE NAME | OMIM | MOI | COMMENT | ENSEMBL TRANSCRIPT | CHR | POS [HG19] | VARIANT | LOCALIZATION OF THE VARIANT | COVERAGE [fold] | VARIANT FREQUENCY [%] | dbSNP | MINOR ALLELE HET * | MINOR ALLELE HOMO * | ExAC SERVER: VARIANTS |
|------------------|--------------------------------------|---|-----|--|--------------------|-----|------------|------------|-----------------------------|-----------------|-----------------------|-------|--------------------|---------------------|-----------------------|
| REEP1 | Receptor accessory protein 1 | OMIM #610250 SPASTIC PARAPLEGIA 31, AUTOSOMAL DOMINANT; SPG31 | AD | | ENST00000165698 | 2 | 86481810 | GTAATAT>AC | intron, donor splice site | 141 | 100 | . | 0 | 0 | 0 |
| CCDC6 | Coiled-coil domain containing 6 | OMIM #188550 THYROID CARCINOMA, PAPILLARY | | phenotype does not fit | ENST00000263102 | 10 | 61666198 | G>A | 5'UTR | 19 | 100 | . | 0 | 0 | 0 |
| C10orf107 | Chromosome 10 open reading frame 107 | No known disease associated with <i>C10orf107</i> mutations | | potentially associated with hypertension | ENST00000330194 | 10 | 63440922 | T>C | 5'UTR | 251 | 100 | . | 0 | 0 | 0 |

Table e-1: Homozygous variants in the autozygous region that were predicted to be disease causing by the MutationTaster2 software. **MOI**, mode of inheritance; **AD**, autosomal dominant; **UTR**, untranslated region; * Frequencies refer to the genotypes of the 1000 Genome