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**Supplemental Data**

# Brown-Vialetto-Van Laere Syndrome, a Ponto-Bulbar Palsy with Deafness, Is Caused by Mutations in *C20orf54*

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C20orf54_wt                  MAFLMHLLVCVFGMGSWVTINGLWVELPLLVMELPEGWYLPSYLTVVIQL 50
C20orf54_c.1325_1326delTC    MAFLMHLLVCVFGMGSWVTINGLWVELPLLVMELPEGWYLPSYLTVVIQL 50

C20orf54_wt                  ANIGPLLVTLLHHFRPSCLSEVPIIFTLLGVGTVTCIIFAFLWNMTSWVL 100
C20orf54_c.1325_1326delTC    ANIGPLLVTLLHHFRPSCLSEVPIIFTLLGVGTVTCIIFAFLWNMTSWVL 100

C20orf54_wt                  DGHHSIAFLVLTFFLALVDCTSSVTFLPFMSRLPTYYLTTFFVGEGLSGL 150
C20orf54_c.1325_1326delTC    DGHHSIAFLVLTFFLALVDCTSSVTFLPFMSRLPTYYLTTFFVGEGLSGL 150

C20orf54_wt                  LPALVALAQGSGLTTCVNVTEISDSVPSPVPTRETDIAQGVPRALVSALP 200
C20orf54_c.1325_1326delTC    LPALVALAQGSGLTTCVNVTEISDSVPSPVPTRETDIAQGVPRALVSALP 200

C20orf54_wt                  GMEAPLSHLESRYLPAHFSPLVFFLLLSIMMACCLVAFFVLQRQPRCWEA 250
C20orf54_c.1325_1326delTC    GMEAPLSHLESRYLPAHFSPLVFFLLLSIMMACCLVAFFVLQRQPRCWEA 250

C20orf54_wt                  SVEDLLNDQVTLHSIRPREENDLGPAGTVDSSQGQGYLEEKAAPCCPAHL 300
C20orf54_c.1325_1326delTC    SVEDLLNDQVTLHSIRPREENDLGPAGTVDSSQGQGYLEEKAAPCCPAHL 300

C20orf54_wt                  AFIYTLVAFVNALTNGMLPSVQTYSCLSYGPVAYHLAATLSIVANPLASL 350
C20orf54_c.1325_1326delTC    AFIYTLVAFVNALTNGMLPSVQTYSCLSYGPVAYHLAATLSIVANPLASL 350

C20orf54_wt                  VSMFLPNRSLLFLGVLSVLGTCFGGYNMAMAVMSPCPLLQGHWGGEVLIV 400
C20orf54_c.1325_1326delTC    VSMFLPNRSLLFLGVLSVLGTCFGGYNMAMAVMSPCPLLQGHWGGEVLIV 400

C20orf54_wt                  ASWVLFSGCLSYVKVMLGVVLRDLSRSALLWCGAAVQLGSLLGALLMFPL 450
C20orf54_c.1325_1326delTC    ASWVLFSGCLSYVKVMLGVVLRDLSRSALLWCGAAVQLGSLRSAAHVPSG 450

C20orf54_wt                  VNVLRLFSSADFCNLHCPA------------------------------ 469
C20orf54_c.1325_1326delTC    QRAAALLVRGLLQSALSSLGRPPTPPPSLTDGTGVQRGQVTEQGAGTERQ 500

C20orf54_wt                  ----
C20orf54_c.1325_1326delTC    SLSN 504
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**Figure S1. Wild Type *C20orf54* Protein and *C20orf54* Frameshift Mutant Protein**

Protein sequences of wild type *C20orf54* and the c.1325_1326delTC (cases 1 & 2) frameshift mutant *C20orf54* are shown. The mutant is predicted to be 35 residues longer than the wild type and is mutated from residue L441. The Genbank accession number for wild type *C20orf54* protein sequence is NP_21234. The mutant protein sequence was predicted using the Translate tool from the ExPASy website.

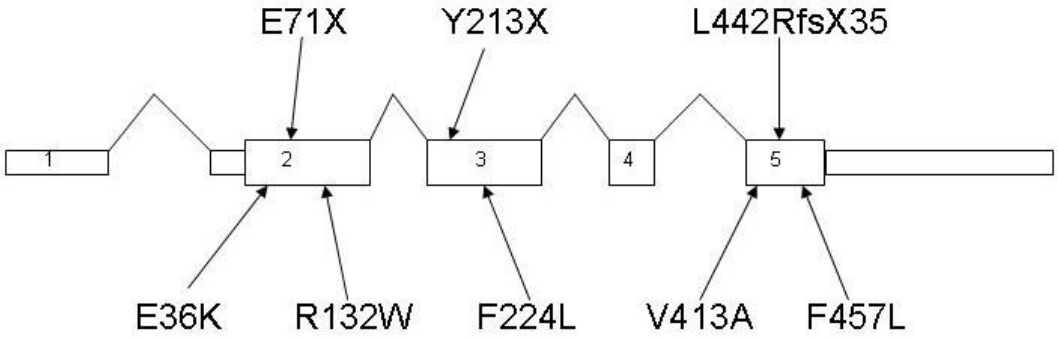


**Figure S2. BVVLS Mutations in *C20orf54***

The 5 exons of *C20orf54* are depicted. Smaller rectangles represent the 5' and 3' untranslated regions. The positions of the mutations described within the text are arrowed

## Table S1. Genotypes of Polymorphisms in Exons of *C20orf54* for Each Mutation

| | dbSNP Reference and Genotype | | | | | | | | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | Exon 1 | | Exon 2 | | | | | | | Exon 3 | | | | | | | | | | Exon 5 |
| Case | rs1884637 | rs57012410 | rs6117517 | rs11467076 | rs35655964 | rs34376836 | rs3746808 | rs3746807 | rs6054614 | rs16992990 | rs6054605 | rs3746806 | rs3746805 | rs3746804 | rs3746803 | rs3746802 | rs6054602 | rs62641669 | rs910857 |
| 1, 2 | G | C | G | GGGCAGATA | C | C | T | C | A | C | C | C | T | C | C | A | G | G | T |
| 3 | G | C | G | GGGCAGATA | C | C | T | C | A | C | C | C | T | C | C | A | G | G | C |
| 4 | A/G | C | G | GGGCAGATA | C | C | C | C | A | C | C | C | C/T | C | C | A | G | G | C |
| 5, 6 | G | C | G | GGGCAGATA | C | C | T | C | A | C | C | C | T | C | C | A | G | G | C |
| 7 | A | C | G | - | G | C | C | T | A | C | C | C | C | T | C | A | G | G | C |
| 8 | G | C | ND | -/ GGGCAGATA | G/C | C | C/T | C/T | A | C | C | C | C | T | C/T | A/G | G | G | C |
| 9 | A | C | G | - | G/C | C | C | C/T | A | C | C | C | C | C/T | C | A | G | G | C/T |

SNPs identified during sequencing of the exons are shown for each BVVLS case descibed in the manuscript. One base shown indicates homozygous; rs11467076 is an indel of 9bp which are shown as homozygous for the insertion while the deletion is represented by - ;ND = Undetermined