Supplementary Appendix

Integration of molecular and clinical data of 40 unrelated von Willebrand Disease families in a Spanish locus-specific mutation database: first release including 58 mutations


Online Supplementary Figure S1. Schematic scale representing the genomic region of VWF with the locations of the 41 different putative mutations included in the database. Exons are represented by vertical black lines. Mutations identified for the first time in our laboratory are shown in bold. Mutation c.6187C>T has also been found in a type 2 VWD patient in compound heterozygous status in trans with mutation c.4225G>T. Mutation c.7730-1G>C has also been found in a homozygous or heterozygous state in type 3 VWD patients. The mutation c.3931C>T in exon 28, involved in a gene conversion event, was also found alone in other type 3 patients.