Supplementary Material to Bastida Bermeja et al. Application of a molecular diagnostic algorithm for haemophilia A and B using next-generation sequencing of entire $F8$, $F9$ and $VWF$ genes” (Thromb Haemost 2017; 117.1)

Suppl. Figure 1: Ratio chart of comparative MLPA analysis between severe HA patient and reference sample.

Right hand side grey area represents reference regions. Box plots on the blue area represent $F8$ gene MLPA results from exon 1 to exon 26. Blue and red lines delineate confidence interval. Red circles represent deleted regions affecting from exon 2 to exon 13. Additionally exon 19 and exon 22 also show results outside of the confidence interval.
Suppl. Figure 2: Pedigree and co-segregation inheritance of the c.1010-842G>A variant in intron 7 of F8 gene.

All female relatives were symptomatic for FVIII deficiency. While the propositus was hemizygous, females were heterozygous.