

Haematologica  
HAEMATOL/2017/178327  
Version 3

Clustered F8 missense mutations cause hemophilia A by combined alteration of splicing and protein biosynthesis/activity

Irving Donadon, John H. McVey, Isabella Garagiola, Alessio Branchini, Mimosa Mortarino, Flora Peyvandi, Francesco Bernardi, and Mirko Pinotti

Disclosures: The authors declare no competing financial interests.

Contributions: I.D expressed and characterized recombinant protein and minigene variants; J.H.M created the LV vectors and expressed rFVIII proteins; I.G and M.M conducted studies on ectopic F8 mRNA and analysed results; A.B created expression plasmids for rFVIII variants; M.P., F.B., J.H.M and F.P conceived the study and designed research, analysed and interpreted data and wrote the manuscript. All authors revised and approved the final version of the manuscript.