

Haematologica
HAEMATOL/2019/217539
Version 3

Next-generation sequencing and recombinant expression characterized aberrant splicing mechanisms and provided correction strategies in factor VII deficiency

Paolo Ferraresi, Dario Balestra, Caroline Guittard, Delphine Buthiau, Brigitte Pan-Petesh, Iva Maestri, Roula Farah, Mirko Pinotti, and Muriel Giansily-Blaizot

Disclosures: The authors declare no conflict of interest.

Contributions: C.Guittard performed the NGS. D.Buthiau recorded and analysed the clinical data. Brigitte Pan-Petesh and Roula Farah were in charge of many patients for pedigree and haplotype analysis respectively. M.Giansily-Blaizot designed the genetic study and analysed the NGS data. M.Pinotti and P.Ferraresi planned the expression studies. P.Ferraresi performed the expression studies and computational analysis. I.Maestri performed the denaturing capillary electrophoresis. P.Ferraresi, M.Giansily-Blaizot and D.Balestra wrote the manuscript. D.Balestra and M.Pinotti revised the manuscript.