

Haematologica
HAEMATOL/2018/194555
Version 4

Molecular mechanisms of bleeding disorder-associated GFI1BQ287* mutation and its affected pathways in megakaryocytes and platelets.

Rinske van Oorschot, Marten Hansen, Johanna M. Koornneef, Anna E. Marneth, Saskia M. Bergevoet, Maaike G.J.M van Bergen, Floris P.J. van Alphen, Carmen van der Zwaan, Joost H.A. Martens, Michiel Vermeulen, Pascal W.T.C. Jansen, Marijke P.A. Baltissen, Britta A.P. Laros-van Gorkom, Hans Janssen, Joop H. Jansen, Marieke von Lindern, Alexander B. Meijer, Emile van den Akker, and Bert A. van der Reijden

Disclosures: none

Contributions: RO, MH, JMK, ABM, EA, and BAR coordinated the research and wrote the majority of the manuscript. RO, MH, JMK, AEM, SMB, MGJMB, and JHAM conducted and analyzed the majority of the experiments. CZ and FPJA helped in the proteomics mass spectrometry analysis. MV, PWTCJ, and MPAB performed the mass spectrometry analysis of GFI1B interacting proteins. BAPLG provided patient samples. HJ performed electron microscopy analysis. JHJ, ML, ABM, EA and BAR provided funding. All authors critically revised the paper and approved the final version.