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Severe bleeding and absent ADP-induced platelet aggregation associated with inherited combined CalDAG-GEFI and P2Y12 deficiencies

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Contributions: B. Lunghi and R. Santacroce performed the genetic studies, interpreted the data and revised the manuscript; A. Lecchi, M. Scavone and R. Paniccia performed the platelet function studies, interpreted the data and revised the manuscript; A. Artoni and G. Castaman evaluated the clinical phenotypes of the patients, interpreted the data and revised the manuscript; C. Gachet contributed to the performance of binding studies, interpreted the data and revised the manuscript; M. Margaglione coordinated and planned the WES investigation, interpreted the data and revised the manuscript; F.Bernardi planned the molecular genetic studies, interpreted data and contributed to write the manuscript; M. Cattaneo coordinated the study, interpreted the data and wrote the manuscript.