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Macrothrombocytopenia and dense granule deficiency associated with
FLI1 variants: ultrastructural and pathogenic features

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Contributions: PS designed and performed the experiments, analyzed data and wrote the paper; LV, DB, VB and SF performed the experiments and analyzed data; MC, FP, MF and PEM analyzed data; CF and CP collected clinical data; JCB, CG and AE performed the ultrastructural study and analyzed data; NS, AR, JFD, RO, AB, PL, and DAT performed the genetic study and analyzed data; FM provided critical advice in the setup of the whole-blood-based flow-cytometric quantification of MYH10 expression; MP and MCA designed and supervised the research and participated in writing the manuscript and contributed equally to this work. All authors participated sufficiently in the work to take public responsibility for appropriate portions of the content, and gave final approval of the manuscript to be published.