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GATA1 erythroid-specific regulation of SEC23B expression and its implication in the pathogenesis of Congenital Dyserythropoietic Anemia type II

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Contributions: RR, IA and AI designed and conducted the study, and prepared the manuscript; AG, performed clinical evaluation of the patients and contributed to sequencing analysis; RR performed in silico studies and ex vivo analyses; IA performed in vitro functional studies; AG and GDR established vector cloning and mutagenesis; FM and AA performed sequencing analysis; FW and MGB provided family samples and cared for the patients.