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PIEZO1-R1864H rare variant accounts for a genetic phenotype-modifier role in dehydrated hereditary stomatocytosis

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Contributions: I.A., R.R.; and A.I. designed and conducted the study, and prepared the manuscript; I.A. and L.D.F. performed the ionic flux assays; R.R. performed the sequencing analysis; F.M. performed the ionic flux assays and real time PCR assays; G.D.R. performed the western blotting analysis; B.E.R. performed the Immunofluorescence assays. A.G. take care of patients. G.T. and A.C. performed the ektacytometry analyses; R.M. performed the mutational screening; L.D.F. also provided critical evaluation of the study.