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Severe Ankyrin-R deficiency results in impaired surface retention and lysosomal degradation of RhAG in human erythroblasts

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Contributions: TJS and AJB contributed equally to this work. TJS and AJB conceived and designed experiments, performed experiments, wrote and edited the manuscript; BRH and SP performed experiments; KEM performed protein 4.2 knockdowns; RvW diagnosed and performed NGS analysis of HS patients and edited the manuscript; BvdZ performed sequencing, CThBMvD, NHtBraak, GH, MPGL, EO, RYJT, EF, PB diagnosed HS patients and provided blood samples; AMT was the principal investigator, conceived and designed experiments, wrote and edited the manuscript.