Title: Homozygous mutations in a predicted endonuclease cause Congenital Dyserythropoietic Anaemia Type I

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Specific contributions of each author are listed on page 2 of this document.

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Please describe the contributions of each author, indicating who was responsible for each part of the study and the preparation of the manuscript (collection of data, experiments, data analysis, generation of figures, collection of images, interpretation of data, preparation of the text, etc.)

C.B. conceived the project and wrote the manuscript, N.A.R. and J.M.B. performed segregation analysis and validation of sequence variants, C.P.P. and L.S-P. performed detailed sequence similarity searches, protein modelling and wrote the manuscript, S.J.McG. performed bioinformatics analysis of whole genome data and SNP analysis, M.R.A conducted patient clinical assessments, M.A.S conducted initial genetic analysis of Family 1, WGS500 consortium, G.A.McV, D.R.B, and P.D. generated whole genome sequence data, O.G. performed protein expression and biochemical analysis, D.R.H. conceived and supervised the project, V.J.B supervised the project and wrote the manuscript.