

Haematologica  
HAEMATOL/2016/144063  
Version 3

Gene panel sequencing improves the diagnostic work-up of patients  
with idiopathic erythrocytosis and identifies new mutations

Carme Camps, Nayia Petousi, Celeste Bento, Holger Cario, Richard R.  
Copley, Mary Frances McMullin, Richard vanWijk, Peter J. Ratcliffe,  
Peter A. Robbins, and Jenny C. Taylor

Disclosures: This work was supported by the National Institute for Health Research (NIHR) Biomedical Research Centre Oxford with funding from the Department of Health NIHR Biomedical Research Centre funding scheme. The WGS500 study was funded by the Wellcome Trust Core Award (090532/Z/09/Z) and a Medical Research Council Hub grant (G0900747 91070) to Peter Donnelly (director of the Wellcome Trust Centre of Human Genetics), the NIHR Biomedical Research Centre Oxford, the UK Department of Health NIHR Biomedical Research Centres funding scheme and Illumina. Some members of the WGS500 consortium are employees of Illumina, Inc., a public company that develops and markets systems for genetic analysis. The other contributors to the WGS500 study, as well as all the authors involved in the work presented here, declare no competing financial interests.

Contributions: Contribution: N.P., P.A.R., P.J.R. and J.C.T. conceived and designed the study; C.B., H.C., R.W. and M.F.M. provided patient samples; the WGS500 consortium generated the WGS data; R.R.C. analyzed the WGS data; C.C. performed the Ion Torrent sequencing using the erythrocytosis gene panel and analyzed the resulting data; C.C., N.P., C.B., H.C., R.R.C., M.F.M., R.W., P.A.R., P.J.R. and J.C.T. interpreted the data; C.C. and N.P. wrote the manuscript; and all authors approved the manuscript.