

Haematologica
HAEMATOL/2014/120980
Version 3

Combined immunodeficiency with CD4 lymphopenia and sclerosing
cholangitis caused by a novel loss-of-function mutation affecting IL21R

Disclosures: Authors declare to have no financial or other potential conflicts of interest.

Contributions: B.E., I.B. and T.H. performed experiments. E.S., Z.A., D.A.C., O.Z. and I.T. provided clinical care of the patients and provided clinical data. K.B. conceived this study, provided laboratory resources and together with B.E., I.B., and E.S. planned, designed and interpreted experiments. B.E., I.B., E.S. and K.B. wrote the manuscript. All authors critically reviewed the manuscript and agreed to its publication.