SUPPLEMENTAL MATERIAL

Kotlarz et al., http://www.jem.org/cgi/content/full/jem.20121229/DC1

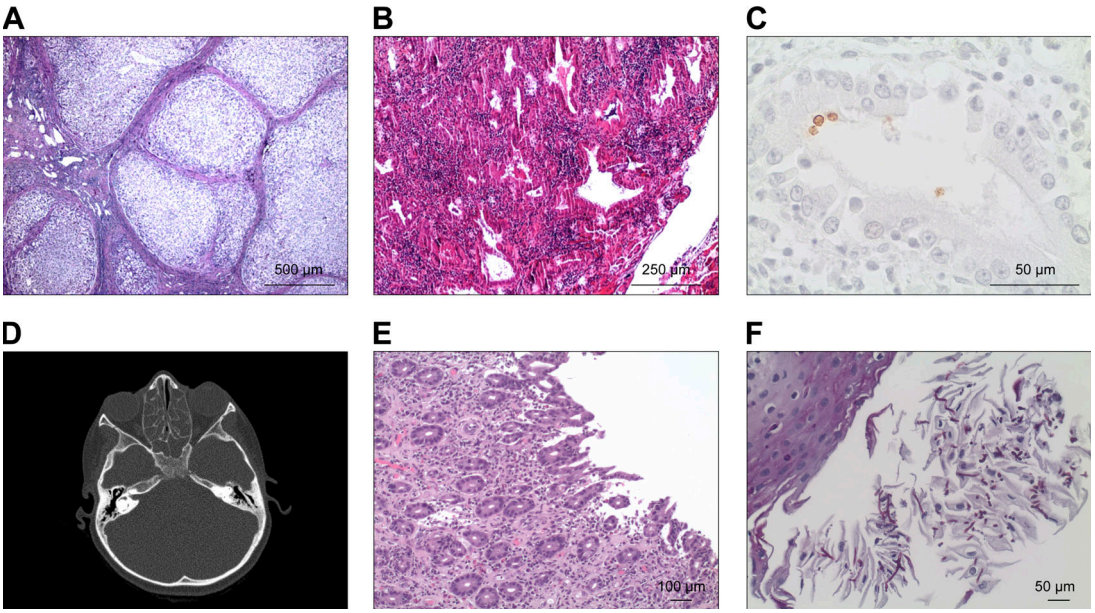

Figure S1. Clinical and histopathological phenotype of P1 and P2 from family A. (A–C) Histopathological analysis of the explanted liver of P1. (A) PAS staining revealed complete cirrhosis with prominent inflammation and lymphangiectasia. (B) The large bile ducts were dilated with pronounced epithelial proliferation and a dense lymphocytic infiltrate. (C) Some glands contained microorganisms on the luminal surface of the epithelial cells, which could be identified as cryptosporidia by immunohistochemistry. (D) Coronal magnetic resonance imaging of P2 illustrating an opacification with soft-tissue density and mucosal thickening in paranasal sinuses as a consequence of recurrent sinusitis. (E) Histopathological analysis demonstrates duodenal mucosa with inflammation and signs of epithelial regeneration due to ulceration and helicobacter gastritis in P2. (F) PAS-staining illustrating superficial candidiasis of the esophageal mucosa in P2.

Table S1, available as an excel file, show variant calling statistics for next-generation exome sequencing in kindred A.

Table S2, available as an excel file, shows the gene expression profile of Th1-, Th2-, and Th17-associated cytokines in IL-21R–deficient patient P2 of family A.

Table S3, available as an excel file, shows the primer for Sanger sequencing of *IL21R* and *RAB3IL1*.