



Figure 2. Demonstration of genetic, flow cytometric and radiographic changes in a patient with AR IFN- γ R1 deficiency. (A) Sanger sequencing confirmation of *IFNGR1* variants in patient and parents, arrows indicating sites of mutation (B) Schematic illustration of the wild type IFN γ R1 protein and the two variants; Variant 1: *p.E218del* and Variant 2: *p.S208Tfs*21* (C) Flow cytometric analysis in patient compared with healthy control, showing significantly reduced IFN γ R1 expression and normal IFN γ R2 expression (D) Flow cytometric analysis in patient compared with healthy control, showing absent STAT-1 phosphorylation in response to IFN- γ stimulation when gated on monocytes (E) Computed tomography images comparing hepatosplenomegaly at the peak of illness and at time of recovery.