IFNγR1 Deficiency Presenting with Visceral Leishmaniasis and Mycobacterium Avium infections mimicking HLH

Muhammad Khalid¹, Sonia Lemos², Katherine Myint-Hpu¹, Debbie Draper¹, Jennifer Stoddard³, Julie Niemela³, Stefania Pittaluga⁴, Sergio Rosenzweig³, Ottavia Delmonte³, and Luigi D. Notarangelo¹

¹National Institute of Allergy and Infectious Diseases
²Centro Hospitalar e Universitario de Coimbra EPE
³National Institutes of Health
⁴National Cancer Institute

July 12, 2021

Hosted file

Manuscript, Compound Heterozygous IFNGR-1 Deficiency_PA1_revised.docx available at https://authorea.com/users/425145/articles/530019-ifn%CE%B3r1-deficiency-presenting-with-visceral-leishmaniasis-and-mycobacterium-avium-infections-mimicking-hlh
Figure 2. Demonstration of genetic, flow cytometric and radiographic changes in a patient with AR IFN-γR1 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-γR1 protein and its 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-γR1 expression and normal IFN-γR2 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.