Lymphoproliferation, immunodeficiency and early-onset inflammatory bowel disease associated with a novel mutation in Caspase 8

Veronika Kanderova, Hana Grombirikova, Irena Zentsova, Kamila Reblova, Adam Klocperk, Martina Fejtkova, Marketa Bloomfield, Barbora Ravcukova, Tomas Kalina, Tomas Freiberger, and Anna Sediva

Disclosures: none

Contributions: VK and IZ designed, carried out and analyzed ex vivo experiments and immunophenotyping; HG, BR and TF designed, performed and analyzed genetic tests, KR performed structural analyses, AK contributed to clinical characterization of the patient and analyzed ex vivo experiments, MF performed ex vivo experiments and immunophenotyping, MB analyzed bone marrow aspirate and contributed to clinical characterization of the CASP8def patients, VK wrote the paper, TK, TF, and AS designed the project, supervised research, and wrote the paper.