

Severe combined immunodeficiency disease (SCID) encompasses a set of disorders with varying phenotypic expression, typically characterized by early onset of infections. SCID is caused by genetic variation in more than nine genes, due to disrupted development of functional T cells and B cells. RAG-deficient SCID, a subclass of SCID, is caused by complete or partial reduction in functionality of the RAG1 and RAG2 proteins. RAG proteins serve as crucial contributors to the genetic diversity of lymphocyte populations in the immune system, promoting combinatorial genetic rearrangement. Mutations in the RAG domains result in a lack of mature lymphocyte populations, contributing to the onset of SCID. This analysis aims to improve the immunological characterization of RAG-deficient SCID by associating phenotype with type of genetic variation through observation of clinical presentation and genotype. We aim to contribute to an increased understanding of the physical characteristics prevalent in a diverse patient population, to assist clinicians in diagnosis of SCID due to RAG-deficiency. A parsing protocol, developed in python, was utilized to extract HTML data from annotated clinical literature on RAG-deficient SCID patients. Patient data was collected from demographic, clinical, and laboratory data, followed by categorizing types of genetic variants: missense, nonsense, and frameshift. We expect to observe an increasing level of severity in phenotypes associated with more deleterious mutations. This review aims to derive phenotypic and genotypic patterns in patient data, serving as a foundation for future assessment of common treatments administered to each patient population, allowing for a comprehensive analysis of personalized therapeutic strategies based on genotype.