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**Development of a T-cell receptor excision circle (TREC) assay to detect severe combined immunodeficiency (SCID) in the newborn population.**

Author (s)

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Severe combined immunodeficiency (SCID) is a group of inherited genetic disorders characterized by defects in T- and B-lymphocyte development. Due to the compromise of the immune system patients with SCID usually die within the first year of life from serious infections. For this reason and the fact that a bone marrow transplant within the first month of life successfully reconstitutes the immune function in many patients, it is being considered for addition to newborn screening panels in several states. The most common form of this disorder is X-linked SCID, mutations in the common cytokine receptor gamma chain account for approximately 50% of cases. Mutations in genes encoding adenosine deaminase, an enzyme involved in lymphocyte maturation, Janus kinase 3 (JAK3), and in the recombination activating genes RAG1 and RAG2 have also been implicated. Due to the diversity of genes affected in SCID we aim to develop a T-cell receptor excision circle (TREC) assay to detect SCID. During the maturation of T-cells, the rearrangement of the T-cell antigen receptor genes gives rise to the T-cell repertoire and to DNA excision circles known as TRECs. The most abundant TREC species is  $\gamma$ Rec-J $\delta$ , produced by 70% of T-cells and is the target of this study. Our assay utilizes polymerase chain reaction and detection with fluorescent probes to quantify the amount of TRECs present regardless of the underlying cause of the immunodeficiency. SCID patients would have very few detectable TRECs, while normal individuals have a high number of TRECs.