PROSPECTIVE AND RETROSPECTIVE TREC TESTING: A CROSSROADS BETWEEN SCREENING AND DIAGNOSIS OF PIDs

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Primary immune deficiencies (PID) is a large group of inborn errors encompassing more than 400 conditions. Some, such as severe combined immune deficiency (SCID), are fatal without treatment, while others have heterogeneous consequences depending on the extent of the immune system dysfunction. Most forms of SCID can be detected by measuring the levels of T-cell recombination excision circles (TREC) in dried blood spots. We present preliminary data from our prospective study aiming to augment the introduction of TREC technology in Bulgaria as a screening tool. A total of 1276 newborns were tested. Additionally TREC technoology was applied retrospectively to selected patients, divided into 3 categories: with proven PID, with possible PID and with secondary immune deficiencies (SIDs). In all of them, cellular immunity was assessed by immunophenotyping panels. The results of neonatal screening reveled that, 0.62% were rated as likely positive (possible PID). This results demonstrated a relatively high percentage of newborns with a possible PID and the need for urgent introduction of the technology in the National screening program. All patients with proven T cell PID had very low, even to zero TREC values. In individuals with a probable diagnosis of PID, TREC were reduced in 43.2%. TREC values of that group as well as of individuals with secondary immune deficiencies correlated with flow cytometrically defined number of T-cells . With the present study, we demonstrated that TREC may have a place in aiding PID diagnosis, and would be a suitable and cheap method for immune health evaluation of large population cohorts.