O-021 - SCREENING FOR CONGENITAL IMMUNODEFICIENCIES - SCID, AGAMAGLOBULINEMIA AND OTHER T AND B CELL LYMPHOPENIAS IN BRAZIL

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INTRODUCTION: Congenital immunodeficiencies are a group of diseases very often considered pediatric emergencies because of their ability to compromise severely newborn's health. Those are usually associated with a high mortality rate when undiagnosed and receive early treatment. In general, they are caused by genetic changes with an autosomal recessive or X-linked pattern. ***OBJECTIVE:*** Implementation of a screening exam for early identification and/or monitoring of patients with congenital immunodeficiencies, SCID, agammaglobulinemia and other T-cell and B-cell lymphopenias - also known as primary immunodeficiencies. ***METHOD:*** DNA isolation in 3.2 mm dried blood spot samples (DBS) followed by amplification using RT-qPCR Multiplex (Real Time Quantitative PCR) technique. Quantification of TREC (T Cell Receptor Excision Circles) and KREC (Kappa-Deleting Excision Circles) amplified fragments by RT-qPCR in DNA samples extracted from DBS. ***RESULTS:*** Sample from 1200 geographically dispersed patients were selected, associated with two undetectable positive controls for the TREC and KREC targets - previously validated - and amplified by the RT-qPCR methodology. Through statistical analysis the minimum healthy cut-off value of 35 copies/µL of TREC and KREC in blood was established. None of the 1,200 patients was classified as undetectable or inferior to the internal cut off. The assay was validated and established routinely in the laboratory. During the period, a patient with two independent collections with an interval of two weeks had KREC amplification profile classified as undetectable, indicating the need for cautious medical monitoring and follow up investigation by other techniques. ***CONCLUSIONS:*** DNA extraction steps, amplification and analysis were successfully optimized for the absolute quantification of TREC and KREC copies in DBS samples using RT-qPCR. The result of the TREC and KREC assay has shown to be highly reproducible, robust and low cost, associated with satisfactory profiles of sensitivity and specificity. The test was also incorporated in the routines of different neonatal screening tests, since it is possible to be performed in the same sample harvested for these purposes.