*Poster*


# Newborn screening to the early detection of patients with spinal muscular atrophy and immunodeficiencies

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## ABSTRACT

**Motivation:** Severe combined immunodeficiencies (SCIDs) and spinal muscular atrophy (SMA) are rare but potentially fatal pathologies. Rapid detection allows prompt and efficient therapy, thereby improving the prognosis of patients. In January 2021 we initiated the first pilot study in Spain by using a RT-PCR based on a combined TREC/KREC/SMA determination assay from dried blood samples (DBS) of newborns.This study aims to evaluate the potential utility of this technique for the detection of both SCID and AME.

**Methods:** RT-PCR (LightMix®KIT, TREC KREC SMA Newborn, Roche-TIB molbiol) from prospectively collected DBS from neonates born in hospitals from Seville, Huelva and Cadiz was performed. Internal and external controls (SCID and AME) were included. TRECS and KRECS were quantified and cut-off points were 6 (copies/punch) for TRECS and 4 (copies/punch) for KRECS. SMA determination was based on positive/negative amplification. Beta-actin was used as sample quality control.

**Results:** From January 2021 to February 2022 a total of 11501 samples were analyzed. No cases of PID or SMA have been prospectively identified. We received two DBS samples (samples 1 and 2) of 3 and 6 months' old patients with suspected SCID and one DBS sample (sample 3) of a 2-week-old newborn with suspected SMA. The technique showed pathological results in the two patients with suspected SCID, that were later confirmed by flow cytometry and genetic analysis (Sample 1,SCID; Sample 2 XLA). Sample 3 did not amplify for SMN1 gene and diagnosis of SMA was confirmed by genetic techniques. All patients have received curative or supportive therapy with good clinical response.

**Conclusions:** Newborn screening of PID is a fast and sensitive technique for early diagnosis of SCID. This is the first prospective newborn screening study for SCID and SMA in Spain. No cases of SCID or SMA have been diagnosed, most likely due to the low incidence of these pathologies. The inclusion of SMA does not imply any extra cost whilst providing a great benefit. The study has allowed the rapid diagnosis of three patients, allowing prompt initiation of specific treatment and thereby avoiding serious sequelae. The inclusion of this technique in the rutinary newborn screening program will most likely be beneficial for the affected children and their families.

## REFERENCES

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