The PCR assay is comprised of two reagents: one containing a reaction buffer and enzyme and one with the primers and the probes for the multiplex assay. In the second study, we used the simple buffer DNA extraction material and swapped the reaction buffer with Perfecta ToughMix® which is commonly used for SCID testing.

The results of these tests suggest that the in-situ assay is compatible with the PCR chemistry developed to identify the homozygous deletion of exon 7 in the SMN1 gene and simultaneously evaluate the copy number of TREC and KREC using a simple elution solution or Extracta DBS solution. The results also confirm that the PCR System developed is optimized fully to detect the four targets in the qPCR assay as compared to Perfecta ToughMix®.

P111. Newborn Screening Facilitates Early Diagnosis of Spinal Muscular AtrophyNewborn Screening Facilitates Early Diagnosis of Spinal Muscular Atrophy

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Background: Spinal muscular atrophy (SMA) is an autosomal recessive disease characterized by defect in the survival of motor neuron 1 (*SMN1*) gene. Type 1 SMA patients typically develop symptoms within the first six months after birth and most patients die before age 2 years. Therefore, early detection by SMA newborn screening (NBS) provides neonates the opportunity for early SMA diagnosis and standard care. We report our results of 18-month period of SMA NBS.

Methods: Two NBS centers, Taipei Institute of Pathology (TIP) and The Chinese Foundation of Health (CFOH), screened for SMA using dried blood spots between September 2017 to February 2019. TIP detected the homozygous SMN1 deletion by genotyping 4 polymorphisms between the SMN1 and SMN2, including the c.840C/T alleles, using Sequenom MassArray. CFOH performed a real-time polymerase chain reaction (RT-PCR) assay to detect the presence of the c.888+100A in the SMN1 intron 7. Molecular diagnosis of neonates with SMN1-deleted status was confirmed by multiplex ligation-dependent probe amplification (MLPA) using genomic DNA samples.

Results: of the 164,352 screened newborns, 10 and 12 newborns were screened positive by RT-PCR and Mass spectrometry, respectively. Thirteen cases with the absence of SMN1 were confirmed by MLPA and then all remained asymptomatic. During 6-month follow-up, five of them with 2 copies of SMN2 became symptomatic and diagnosed as type I SMA, 2 of then died of necrotizing enterocolitis and acute respiratory failure, respectively. While six cases with 3 or 4 copies of SMN2 remained asymptomatic, another 2 lost follow up. The other 9 cases revealed 1 copy of SMN1 by MLPA and eight of the nine false-positive cases were resulted from gene conversions between SMN1 and SMN2.

Conclusion: SMA NBS by RT-PCR and Mass spectrometry is feasible and can provide early diagnosis of SMA. Our results revealed an incidence of newborns with homozygous SMN1 deletion in Taiwan is about 1 in 12,642.

P112. Newborn Screening of Congenital Toxoplasmosis Infection in northern Taiwan

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Background Congenital Toxoplasmosis is a cause leading to development of central nervous system anomalies and neurological sequelae in neonates if their mother acquired infection during pregnancy. Early detection and accurate management can reduce the burden from these long-term neurological sequelae. This study aimed to investigate the seropositive rate of Toxoplasma infection in pregnant women and for early diagnosis and treatment of congenital Toxoplasmosis in newborns.