P-227 - COMPARISON OF FDA-CLEARED NEWBORN SCREENING PLATFORMS FOR LYSOSOMAL STORAGE DISORDERS - POMPE AND MPS I

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INTRODUCTION: As Latin American countries seek to expand newborn screening to include lysosomal storage disorders (LSDs), prospective results from active LSD screening programs can be used to compare the performance of available screening methodologies. Two FDA-cleared platforms are currently available for LSD enzyme testing from dried blood spot specimens - tandem mass spectrometry (MS/MS) and digital microfluidic fluorometry (DMF). Both methods are currently being actively used for newborn screening (NBS) of Pompe disease and Mucopolysaccharidosis Type I (MPS I) in several states and both platforms offer FDA-cleared specific reagent kits that use synthetic substrates and have the ability to screen for multiple LSDs in a single run. The ability of each method to discriminate normal from affected samples must take into account the multiple sources of variability (biological/genetic variability, DBS sample quality, gestational age at sampling, for example) that exist between newborn samples.

OBJECTIVES: We will use published data sets from active LSD screening programs to evaluate the clinical performance of MS/MS and DMF for Pompe and MPS I newborn screening. RESULTS: Substantial data sets are available from the prospective LSD screening programs in the United States from the states of Missouri (N=441,000 infants screened using DMF) and Illinois (N=220,000 infants screened using MS/MS). The data sets do not show significant differences between the two testing platforms in terms of the positive predictive value (PPV). It is clear, however, from these and other data, that further testing algorithms are required to improve the PPV of both the MPS I and Pompe disease screening tests using any of the currently available platforms. CONCLUSIONS: We conclude that the active and pilot LSD screening programs in the United States are excellent, unbiased resources for those interested in initiating NBS for LSDs into their laboratories. The emerging clinical results from these prospective screening programs reflect the real world performance of each platform and should be considered alongside the associated costs (equipment, maintenance, personnel, etc.) and workflow of each platform to determine the best fit for each NBS laboratory.