RESULTS OF A MEXICAN NEWBORN SCREENING PROGRAM: GLOBAL INCIDENCE COMPARISON

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\textbf{INTRODUCTION:} Newborn screening (NBS) is fundamental for early diagnosis of inborn errors of metabolism (IEM) and other disorders. Although, these are known as rare, as group they become a relevant cause of morbidity and mortality. The disparity in the NBS performed, different laboratory methodologies, and suboptimal coverage, among others, explain the international statistics’ variability.

\textbf{OBJECTIVE:} To analyze and compare the incidence obtained by a Mexican NBS program with the described in the world literature. \textbf{MATERIALS AND METHODS:} We reviewed 199,065 NBS reports performed by Genomi-k’s NBS program from January 1, 2008 to January 31, 2018. All newborns (NB) were screened for 68 pathologies; 62,584 were also evaluated for SCID and 56,979 for six lysosomal storage diseases. We collected and filtered the published data per screened diseases. Further, we estimated their incidence and compared it with our results. \textbf{RESULTS:} An overall incidence of 48.3 affected NB and 101.6 heterozygotes per 10,000 screened NB (from the latter, 63% for Hb S) was found. Particularly, the IEM incidence that could be compared to previous studies was 5.8: 10,000 screened NB. The five most frequent diagnosed pathologies comprehend 89% of the positive results. These were: glucose-6-phosphate dehydrogenase deficiency (G6PD), congenital hypothyroidism (CH), Fabry disease, Cystic Fibrosis (CF), and Pompe disease. \textbf{DISCUSSION AND CONCLUSIONS:} Overall, the incidence we estimated exceeded other countries’ incidence by more than 100%. This could be explained by the consideration of more diseases besides IEM (42.5: 10,000 NB). Even though, IEM’s incidence is one of the highest, like the described in Spain and United Kingdom (6.4 and 6.5: 10,000 NB, respectively). Our estimated incidences are generally greater than those presented in the literature. This proves that the data variability is due to the number of biomarkers, the NBS methodology, and the population distribution. We highlight the frequency of G6PD, CH, and Fabry disease in the studied population. Since the patient's prognosis is proportional to early diagnosis and treatment, we recommend the inclusion of these biomarkers within any NBS program in Mexico.