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Newborn screening represents a major public health achievement, as early diagnosis leads to prompt treatment, which, in turn, decreases morbidity and mortality. However, current newborn screening programs rely on centralized laboratory screening that requires infrastructure for specimen transport, laboratory analysis, results reporting, and follow-up. As a result, regions of the world that lack this infrastructure, including the Global South, Indian subcontinent, and remote and resource-limited areas, are unable to provide newborn screening services. Recently developed smartphone point-of-care (POC) immunochromatographic assays for TSH, cortisol, and sickle cell hemoglobinopathies provide an alternative method to central lab testing for newborn screening. POC newborn screening eliminates the need to transport large number of samples to a central laboratory, decreases the requirement for capital-intensive laboratory equipment, and shortens turn-around-time for results reporting and patient follow-up. Using a smartphone for patient registration, test interpretation, results reporting, and data archiving enables real-time geomapping, patient-specific decision support, and automated case management. We have used smartphone POC TSH testing for newborn thyroid screening in a Bangalore, India slum and in western Uganda, POC cortisol testing to screen for Addison's disease in Kalahari Desert Bushmen, and POC sickle cell screening in a rural population on the Uganda Congo border. These studies demonstrate the feasibility and cost-effectiveness of POC newborn screening for endocrine and hematologic diseases, define the advantages, benefits, and limitations of POC newborn screening, and provide a template for developing POC methods to screen for other common congenital metabolic disorders such as PKU.

4. Posters

P01. Evaluation of Selective Newborn Screening for Inborn Errors of Metabolism in Macedonia during the Period 2014–2017

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The introduction of tandem mass spectrometry (LC/MS/MS) in newborn screening programs in many countries, has increased the capacity to test newborns for inborn errors of metabolism (IEM). LC/MS/MS can identify and quantify several acylcarnitines and amino acids in a single test, and is able to detect more than 40 metabolic disorders, with the combined incidence of about 1 in 5000 babies, not including phenylketonuria (PKU). Screening for IEM in Macedonia was performed by measuring of two groups of compounds, 12 amino acids and 13 acylcarnitines (Chromsystems Diagnostics, Germany), in dried blood spot (n = 16,075) collected 48 h after births, using LC/MS/MS method, during the 2014–2017. Total of 16,075 newborns (18.6% of neonatal population per year), selected from six birth centers in the country (coverage 89.2%), have been screened. 8407 (52.3%) were male and 7668 were (47.7%) female with male to female ratio of 1.09:1. Among screened neonates 56.2% were Macedonians of Slavic origin, followed by ethnic Albanians (31.8%), Roma (6.7%), Turks (3.3%), Bosnians (0.7), Boshnjac (0.6%) and others (0.7%). Were detected total of 8 newborns with IEM: 4 with medium-chain acyl-CoA dehydrogenase deficiency (MCAD), 2 with phenylketonuria (PKU), one with hypermethioninemia (MET), and one with tyrosinemia type I (confirmed by the second-tier test). Selective screening is an important diagnostic tool for the diagnosis of various types of IEM, and it can provide substantial benefits to patients and their families. Early diagnosis is important not only for treatment but also for genetic counseling. Activities to cover all newborns in Macedonia are underway.

Keywords: Metabolic disorders; Newborn screening; Tandem mass spectrometry

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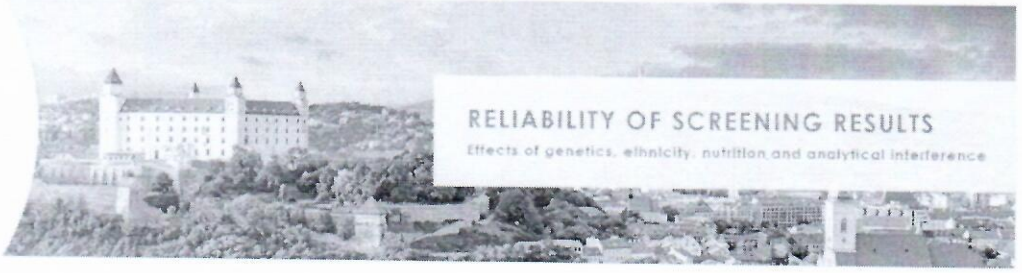

CERTIFICATE OF ATTENDANCE

We herewith certify that

Violeta Anastasovska
Macedonia, The Former Yugoslav Republic Of

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