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Implementation and progress of SMA neonatal genetic screening in republic of moldova

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Background: Spinal Muscular Atrophy (SMA) is a life-threatening autosomal recessive neuromuscular disorder, primarily caused by mutations in the SMN1 gene. Early detection of SMA through newborn screening (NBS) is essential to prevent irreversible neuronal damage, guide treatment and follow-up. Aim: to develop and implement a pilot newborn genetic screening program for SMA.

Materials and Methods: The Human Molecular Genetics Lab at the Mother and Child Institute (IMC) undertook a pilot genetic screening approach for newborns, utilizing an algorithm designed for SMA mutation analysis. Newborn blood samples were collected on filter paper cards after securing informed consent. These samples were subjected to thorough analysis using advanced molecular-genetic methods, including qPCR and MLPA techniques.

Results: The research design and protocol received approval from the Research Ethics Committee. The study successfully implemented necessary molecular genetic methods, including real-time PCR with specially designed probes and arrays. For comparison and validation, a commercial set was employed. The MLPA method was integral in confirming and assessing the copy number variations of SMN1/SMN2 genes. A total of 300 informed consents were obtained, and 265 samples screened. Three samples were flagged as potentially positive and one was confirmed as positive upon re-screening and then by MLPA

Conclusions: The screening algorithm is primarily designed to detect spinal muscular atrophy (SMA) in newborns by identifying homozygous deletions in exon 7 of the SMN1 gene. About 5% of SMA cases with compound heterozygous mutations are undetected. This screening enables early identification of SMA, allowing for timely intervention. Implementing this screening at the IMC evaluates its practicality as an early diagnostic tool for SMA, potentially benefiting Moldova's healthcare system.

Biography

Iulia Coliban is a distinguished Molecular Biologist and Research Scientist at the Human Molecular Genetics Laboratory of the Mother and Child Institute, pioneering in genetic diagnostics. She is spearheading an innovative newborn screening program for Spinal Muscular Atrophy (SMA), aiming to mitigate the irreversible neuronal damage caused by this condition. With a PhD fellow in Molecular Biology and Medical Genetics from Nicolae Testemițeanu State University of Medicine and Pharmacy, Mrs. Coliban has a profound background in neonatal diagnostics, emphasized by her novel approach to presymptomatic detection of the SMN1 gene exon 7 deletion. Her method merges real-time PCR method with High-Resolution Melt analysis, significantly enhancing the early detection and patient outcomes for SMA. The design's novelty lies in its integration of both commercial and customized reagents, balancing sensitivity with cost-effectiveness, while ensuring broad utility for all ages. This has a substantial socio-economic impact by fostering early and accurate diagnoses.