Control/Tracking Number: 2026-A-2308-ESHG
Activity: ESHG Abstract
Current Date/Time: 3/12/2026 5:13:03 AM

International assessment of newborn screening for spinal muscular atrophy using advanced sequencing and artificial intelligence

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Abstract:
Background: Spinal muscular atrophy (SMA) is an autosomal recessive neuromuscular disease with low prevalence (1/10,000 births) and heterogeneous clinical and genetic presentation, caused mainly by homozygous deletions in exon 7 of the SMN1 gene. Clinical severity is modulated by the number of copies of the SMN2 gene, and symptomatology varies from severe forms with neonatal onset to mild manifestations in adulthood. Early diagnosis is essential given the availability of disease-modifying therapies, with newborn screening (NBS) being a key tool for the presymptomatic identification of affected individuals.
Material and Methods: As part of a pilot study conducted in collaboration with Paraguay and Denmark, 92 anonymous dried blood samples were received. After excluding low-quality samples, 71 samples (41 diagnosed SMA cases, 19 carriers, and 11 healthy donors) were analysed along with 3 controls. The Phivea® platform (gMendel Test - SMA), based on long-read sequencing and artificial intelligence (AI), was used. The analysis allowed classification of samples into homozygous deletion of SMN1 exon 7, heterozygous status, or normal genotype, and the number of SMN2 copies was also assessed.
Results: An analytical sensitivity of 100%, specificity of 96.8%, and overall accuracy of 98.6% were obtained. Diagnostic sensitivity was 97.6%, and diagnostic specificity was 100%. No false positives were detected, and the negative likelihood ratio was 0.024.
Conclusion: These data demonstrate the high robustness and reliability of the gMendel-SMA test, validating its application in population-based programs. International collaboration and the use of advanced technologies reinforce the feasibility of SMA NBS in resource-limited countries.