E-Poster

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Neonatal Screening Results for Spinal Muscular Atrophy in Romania: Insights from a Pilot Program

Topic: Diagnosis

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BACKGROUND

Spinal Muscular Atrophy (SMA) is a severe genetic neuromuscular disorder caused by deletions or mutations in the SMN1 gene. Early diagnosis through neonatal screening is essential for timely therapeutic intervention, significantly improving clinical outcomes. In August 2022, a pilot neonatal screening program for SMA was launched in Romania, aiming to assess feasibility and impact.

To present the preliminary results of the ongoing SMA neonatal screening pilot program in Romania, evaluating its effectiveness in early detection and referral for treatment.

METHODS

The program started in August 2022 with four maternity hospitals and has progressively expanded to 25 maternities nationwide. Dried blood spot samples from newborns were analyzed for SMN1 gene deletions using RT-PCR. Positive results were confirmed through genetic testing, and affected infants, along with their families, were referred for further medical evaluation and early therapeutic intervention.

RESULTS

Approximately 50,000 newborns have been screened since the program's inception. Eight newborns tested positive for SMN1 deletions, resulting in an estimated incidence rate of 1 in 6,250 live births. Additionally, two relatives of affected infants were identified as SMA-positive through cascade genetic testing. All confirmed cases were promptly referred for specialized care, with early access to disease-modifying therapies. The program has faced challenges in logistics, parental awareness, and equitable access to treatment, but its expansion from 4 to 25 maternities demonstrates increasing feasibility and acceptance.

CONCLUSION

The Romanian pilot neonatal screening program for SMA has successfully identified affected infants early, proving its feasibility and clinical impact. The ongoing expansion suggests a strong foundation for a future national program, which could significantly improve early SMA diagnosis and patient outcomes in Romania.

Statement of Disclosures

None

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