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Assessment of awareness of the early clinical features of Spinal Muscular Atrophy (SMA) amongst pediatricians

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Background: Spinal muscular atrophy (SMA) is an autosomal recessive neuromuscular disease characterized by progressive atrophy of the voluntary muscles of the limbs and trunk. The rarity of SMA along with an overlap of early clinical features with other neuromuscular disorders contributes to persistent diagnostic delay that varies from 3.6 months (type I) to 43.6 months (type III); these timeframes overlap with a period of irreversible denervation. Clinical trial data suggests early treatment is critical to modifying the rapid loss of motor neurons while improving life expectancy and clinical outcomes.

Objective: To further understand the diagnostic awareness of SMA amongst

general pediatricians as they interact with undiagnosed infants presenting early symptoms.

Methods: The survey was distributed to general pediatricians via email by Medscape Education. The survey was designed to review the clinical decision making of general pediatricians upon observation of hypotonia and/or gross motor delay, as well as overall awareness of the early signs of SMA and neuromuscular disorders.

Results: 300 general pediatricians completed the survey. Upon observation of hypotonia, 55.59% of pediatricians indicated they would immediately refer to early intervention, while 51.64% would immediately refer to a pediatric neurologist for further evaluation. 52.67% correctly indicated that genetic testing is required to make a definitive diagnosis of SMA. Additionally, 70% of respondents indicate comfort identifying the early signs of neuromuscular disease (Extremely comfortable 3.33%; Very comfortable 18.67%; Moderately comfortable

48.33%). Likewise, 67.3% of respondents noted a familiarity with SMA (Extremely familiar 4.33%; Very Familiar 13.67%; Moderately familiar 49.33%), yet of this group, only 59% identified the genetic testing requirement.

Conclusion: This research yielded important insights into current clinical knowledge gaps regarding the diagnosis of SMA and a varied clinical response upon observation of hypotonia amongst pediatricians. Practice guidelines and continuing education may alleviate variation while reducing diagnostic delay.

Biography

Mary Curry has dedicated her career to the improvement of health general well-being of individuals. She currently leads the Education and Awareness Topic Group of the Cure SMA Industry Collaborative (SMA-IC). The SMA-IC was established to leverage the expertise of pharmaceutical companies working in SMA drug development as we collectively address concerns that impact the SMA community. Via the Education Topic Group, she has contributed to the development of Cure SMA's recently launched SMARt Moves, a new disease awareness and educational campaign to empower parents, pediatricians and other healthcare professionals to promptly recognize the early signs and quickly diagnose SMA.

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