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Review Article

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Nutritional Management Evolution in Children with Spinal Muscular Atrophy Type 1 Undergoing New Therapies: A Retrospective Study from a Reference Center

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ABSTRACT

Spinal muscular atrophy (SMA) type 1 is a severe neuromuscular disorder caused by mutations in the SMN1 gene. Historically associated with poor survival, recent advances in targeted therapies have significantly improved prognosis. This study aimed to describe the evolution of nutritional support in patients with SMA type 1 at a tertiary care reference center between 2008 and 2023. A retrospective descriptive analysis was conducted including 19 patients, classified into three treatment groups: supportive care, pharmacologic treatment (intrathecal Nusinersen or Risdiplam), and gene therapy. Nutritional interventions, respiratory support, complications, and clinical evolution were recorded. Among patients receiving only supportive care, 83% required nasogastric feeding and experienced 100% mortality by a median of 5 months. In the pharmacologic treatment group, 63% required long-term enteral feeding and one death occurred due to non-neurological causes. In the gene therapy group, only one child required long-term nutritional support, and all patients showed favorable clinical progression. Our findings highlight the transformation in nutritional needs prompted by disease-modifying therapies, underscoring the importance of early and specialized nutritional intervention. Multidisciplinary management including pediatric nutrition specialists is essential for optimal care of SMA type 1 patients.

Keywords: Spinal Muscular Atrophy Type 1, Nutritional Support, Gene Therapy, Pediatric Nutrition, Enteral Nutrition, Multidisciplinary Care

Introduction

Spinal muscular atrophy (SMA) type 1 is an autosomal recessive neuromuscular disease characterized by degeneration of anterior horn motor neurons due to mutations in the survival motor neuron 1 (SMN1) gene. It is the most severe form of SMA and typically presents within the first six months of life, leading to progressive hypotonia, respiratory failure, and early mortality if untreated. Until recently, treatment was limited to supportive care, and nutritional management focused on palliative feeding strategies. The advent of disease-modifying treatments, including intrathecal Nusinersen, oral Risdiplam, and gene therapy, has markedly improved survival and functional outcomes. As a result, nutritional intervention must adapt to evolving patient needs. This study aims to evaluate how the nutritional approach to SMA type 1 has evolved in a reference hospital following the implementation of these novel therapies.

Review of Literature

Previous literature establishes that malnutrition and feeding difficulties are prominent in SMA type 1 due to bulbar dysfunction, oral motor incoordination, and overall muscle weakness. Traditional approaches emphasized short-term enteral feeding, often through nasogastric tubes. The introduction of Nusinersen (Spinraza) and later, gene therapy (onasemnogene abeparvovec) has improved survival and motor function, altering the disease trajectory and increasing the relevance of long-term nutritional strategies. Emerging data supports the integration of pediatric nutritionists into multidisciplinary teams, as these patients may

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require gastrostomy placement, management of oral aversion, and tailored caloric plans to support growth and development. However, real-world data on how these changes are reflected in clinical practice remains limited.

Materials and Methods

This was a retrospective descriptive study conducted at the Vall d'Hebron Barcelona Hospital Campus from 2008 to 2023. Inclusion criteria included confirmed diagnosis of SMA type 1 and treatment or follow-up at our center. Nineteen patients (9 males) were included. Data collected included sex, age at diagnosis, type of nutritional intervention (nasogastric tube or gastrostomy), respiratory support, complications, and clinical evolution. Patients were classified into three groups based on treatment modality:

* Group A: Supportive care only
* Group B: Intrathecal Nusinersen or oral Risdiplam
* Group C: Gene therapy

Discussion

Findings indicate a clear shift in nutritional intervention practices correlating with improved prognosis due to new therapies. In group A, short-term nutritional devices predominated, reflecting limited survival and palliative care goals. Group B showed an increase in long-term nutritional device use and survival, supporting the role of proactive nutrition in treatment plans. Group C patients required minimal nutritional intervention, suggesting improved bulbar function and feeding capacity. However, all still required respiratory support, indicating the ongoing need for comprehensive care. Notably, the single death in group B was unrelated to SMA progression, highlighting the changing landscape of morbidity and mortality. These findings underscore the necessity for dynamic nutritional protocols tailored to therapeutic advances.

Findings

* 100% of supportive care patients (Group A) died by 5 months, with most requiring nasogastric feeding.
* In the pharmacologic therapy group (Group B), 63% required gastrostomy and one death occurred due to abdominal sepsis.
* In the gene therapy group (Group C), only one child required long-term feeding due to oral aversion, and all survived with favorable outcomes up to 1.5 years.
* Improved survival was associated with a shift from short-term to long-term nutritional strategies.

Limitations Conclusions and Recommendations

Limitations include small sample size and single-center design, which may limit generalizability. Additionally, the retrospective nature of the study restricts causality assessment. Nonetheless, the results strongly support integrating pediatric nutrition specialists into SMA care teams. Nutritional needs are evolving with treatment advancements and must be proactively addressed to optimize outcomes. Further prospective studies are recommended to validate these findings and refine nutritional protocols.

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