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Case Report

Miglustat in Niemann-Pick Disease Type C: A Case Report

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Abstract

Background: Niemann-Pick disease type C (NPC) is a rare lysosomal storage disorder with neurodegeneration that often leads to cardio-respiratory collapse. Diagnosis requires a high index of suspicion, as patients can present with a wide diversity of signs, symptoms, and ages of onset.
Case presentation: A 31-year-old female presented with shortness of breath, tachycardia, and tachypnea of unknown etiology. Her past medical history includes NPC with seizures, dysphagia, and pneumonia secondary to aspiration. She is on miglustat therapy. Evaluation revealed aspiration pneumonia treated with antibiotics.
Conclusion: Aspiration pneumonia was confirmed via chest X-ray and CT and treated effectively. It remains the leading cause of death in NPC patients. Miglustat may have contributed to improving her swallowing capacity and delaying neurological decline.

Keywords: Niemann-Pick Disease Type C • Aspiration Pneumonia • Miglustat

Introduction

Niemann-Pick disease type C (NPC) is a rare autosomal recessive lysosomal storage disorder caused by mutations in the NPC1 or NPC2 genes, disrupting the trafficking of cholesterol and glycosphingolipids [1]. The estimated incidence ranges from 1 in 89,000 to 1 in 120,000 live births [2-4].

NPC can present anytime from the prenatal period to late adulthood [5-8]. Systemic involvement of the liver, spleen, or lungs is seen in over 85% of patients and typically in advance to neurological symptoms [2]. Disease onset between 6-15 years is referred to as juvenile onset, characterized by ataxia, cognitive decline, learning disabilities, vertical supranuclear gaze palsy, dystonia, dysarthria, dysphagia, and seizures [5].

Diagnostic evaluations include biomarker screening for oxysterols, genetic testing for NPC1/2 mutations, skin biopsy, fibroblast culture, and filipin staining [6,7]. Imaging may reveal brain atrophy, although it can also appear normal. NPC is frequently misdiagnosed, and if left untreated, most patients die from aspiration pneumonia between the second and fourth decades of life [5,8].

Case Presentation

A 31-year-old female presented to our community hospital with a 24-hour history of shortness of breath, tachycardia, and tachypnea. Her oxygen saturation was 92% on 100% oxygen via nasal cannula. Chest CT showed bilateral lower lobe infiltrates confirming pneumonia. She has a PEG tube for medication but eats orally, which may have led to aspiration.

Diagnosed with NPC at age 9, her history includes seizures, dysphagia, and cognitive impairment. She is wheelchair-bound but completed high school without developmental delays. She has been seizure-free for six months and has had only one prior aspiration pneumonia episode 10 years ago.

On examination, she was awake, alert, with dysarthria, dysphagia, and dementia-like features. Splenomegaly was noted. Respiratory exam revealed bilateral crackles; cardiac exam was unremarkable.were more medical students, 227 (64.5%) who participated in the study than non-medical students, 125 (35.5%). The highest percentage of participants are 1st year students (39.8%) while the lowest percentage are 3rd year students (37.2%), 2nd year students were 114 (32.4%).

Investigations:

1. **Chest CT:** Bilateral lower lobe infiltrates.
2. **Abdominal CT:** Enlarged spleen, pancolonic fecal retention indicating constipation.
3. **Bedside Flexible Endoscopic Evaluation of Swallowing:** Reduced tongue movement, tongue rocking, diminished tongue base retraction, premature spillage.

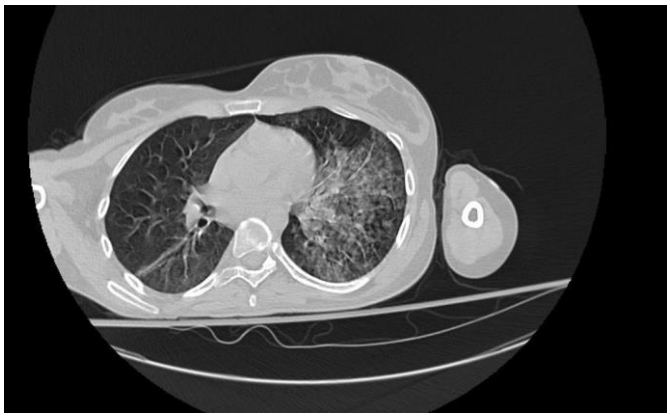

Figure 1: Showing bilateral lung infiltrates but more prominent in right lung suggesting aspiration pneumonia.

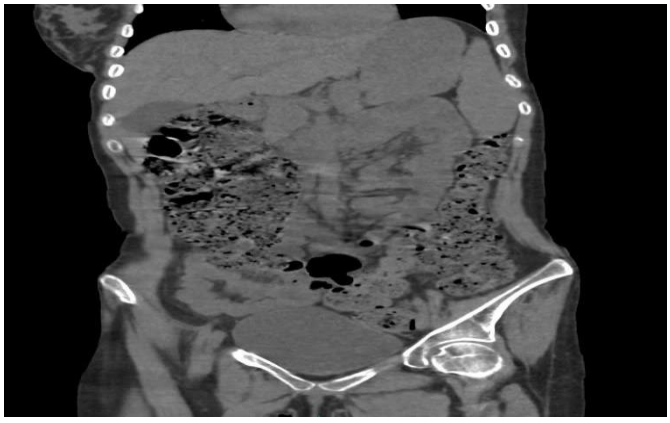

Figure 2: CT abdomen and pelvis shows enlarged spleen and swirl sign involving the terminal ileum with wall thickening just proximal to the valve. Large pancolonic fecal residue suggesting chronic constipation.

Discussion

NPC is caused by mutations in the NPC1 or NPC2 genes, resulting in disrupted intracellular lipid trafficking and accumulation of cholesterol and glycosphingolipids in lysosomes [1]. This leads to progressive neurodegeneration, hepatic dysfunction, and systemic involvement [2]. The neurological decline is typically the most significant contributor to reduced quality of life. Most patients die between ages 10 and 25, commonly from aspiration pneumonia due to dysphagia [2].

Our patient's survival into her early 30s is uncommon and suggests protective factors like early diagnosis and proactive care. Diagnosed at age 9, she was started on miglustat, although the exact initiation age remains to be clarified. Miglustat inhibits glucosylceramide synthase, reducing glycosphingolipid synthesis and subsequent lysosomal accumulation [7]. Studies demonstrate that miglustat can slow neurological deterioration, particularly in swallowing and ambulation—critical functions in preventing respiratory complications [7].

Preserving swallowing capacity is vital since dysphagia significantly increases the risk of aspiration pneumonia, the leading cause of death in NPC [2]. Some evidence suggests miglustat may stabilize or improve swallowing function, aligning with our patient's clinical trajectory [9].

Constipation in our patient could be multifactorial: neurodegeneration of the gut, side effects from medications like glycopyrrolate (prescribed for secretion control), and potential gastrointestinal effects of miglustat itself [10]. Management of these complications is essential, as constipation can contribute to regurgitation and further risk of aspiration.

Conclusion

Niemann-Pick disease type C remains a challenging neurodegenerative disorder with significant morbidity and mortality, primarily driven by complications such as aspiration pneumonia. This case highlights the importance of early diagnosis, comprehensive management and the potential benefits of therapy such as miglustat; there are cases in which miglustat improves dysphagia in Niemann disease type C [11-13]. The patient's survival into her 30s suggests that even though targeted therapy is not approved for type c, proactive interventions, including attention to swallowing function, seizure control and gastrointestinal management, may positively influence outcomes. Continued research and clinical awareness are essential to improve quality of life and longevity in individuals with NPC, as well as improve techniques for treatment.

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