P-57

Management of Respiratory Failure in ARSACS Using Non-Invasive Ventilation: A Case Report

Hee Jae Park[1,2], Juhyun Kwon[1,2], Han Eol Cho[1,2]

[1] Department of Rehabilitation Medicine, Gangnam Severance Hospital
[2] Rehabilitation Institute of Neuromuscular Disease, Yonsei University College of Medicine

INTRODUCTION

Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS) is a rare neurodegenerative disorder primarily affecting the nervous system. Respiratory failure is not a typical feature, but neuromuscular weakness may lead to respiratory complications. This case highlights the first reported instance of respiratory failure in ARSACS, successfully managed with non-invasive ventilation (NIV).

Case Report

A 52-year-old male was admitted to the Pulmonary Rehabilitation Center at Gangnam Severance Hospital due to complaints of chronic fatigue, headaches, and shortness of breath, particularly during speech and with exertion. He also reported difficulty clearing his airway due to excessive sputum production and a reduced ability to cough. The patient had a history of progressive neurological decline starting in early childhood. By high school, he could no longer walk independently, became wheelchair-bound in his 30s, and was bedridden in his 40s. Neurological assessment revealed cerebellar dysfunction, limb weakness, impaired proprioception, and severe coordination difficulties. Brain MRI showed cerebellar atrophy, and genetic testing identified a pathogenic mutation in the SACS gene, confirming the diagnosis of ARSACS. Pulmonary function tests revealed severe restrictive lung disease and hypercapnia. Overnight transcutaneous CO_2 monitoring showed chronic CO_2 retention. NIV was initiated with BiPAP, leading to symptom improvement. At the five-year follow-up, the patient's CO_2 levels had normalized with consistent NIV use, despite overall decline in physical and respiratory function.

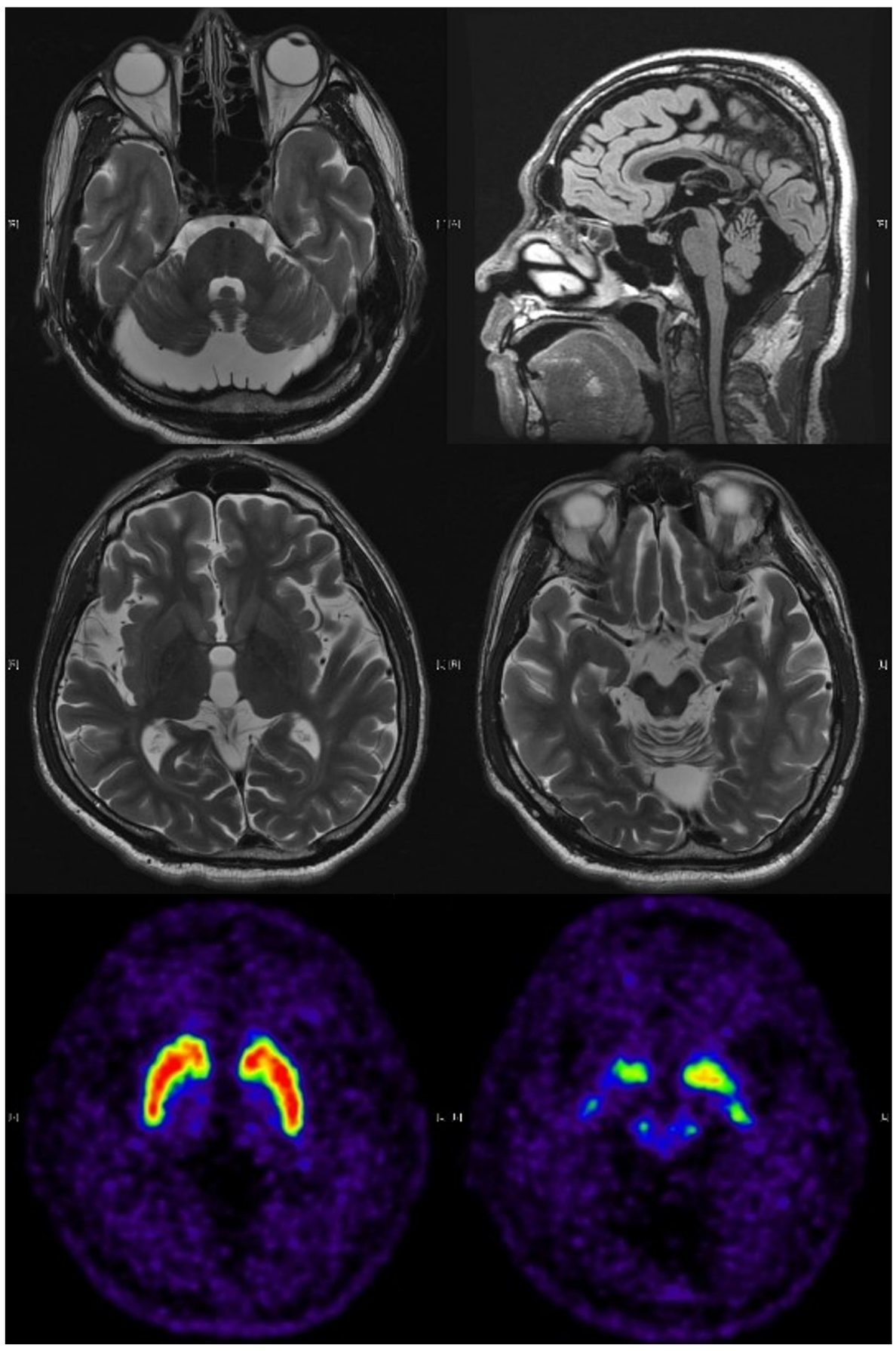

Figure 1. Brain MRI and FP-CIT images of the patient with ARSACS
Top row (A, B, C, D): T2-weighted brain MRI shows marked cerebellar atrophy, particularly prominent in the vermis and superior cerebellum. No significant brainstem atrophy is observed. Axial sections show enlarged cerebrospinal fluid (CSF) spaces in the posterior cranial fossa without fourth ventricle enlargement, suggesting mega cisterna magna.
Bottom row (E, F): FP-CIT images showing normal dopamine transporter activity

CONCLUSIONS

This is the first reported case of respiratory insufficiency in a patient with ARSACS. While ARSACS is primarily a neurological disease, this case demonstrates that respiratory muscle weakness and chronic ventilatory failure can occur. NIV proved to be a safe and effective intervention, alleviating symptoms and stabilizing hypercapnia over five years. Regular respiratory assessments should be considered in ARSACS patients to ensure timely intervention. Further studies are warranted to explore the prevalence and management of respiratory complications in ARSACS.