JMSCR Vol||13||Issue||08||Page 07-09||August 2025

http://jmscr.igmpublication.org/home/
ISSN (e)-2347-176x ISSN (p) 2455-0450
crossref DOI: https://dx.doi.org/10.18535/jmscr/v13i08.02

IGM Publication
Journal Of Medical Science And Clinical Research
An Official Publication Of IGM Publication

Probable Anti-IgLON5 Antibody Disease Presenting with Bulbar Weakness, REM Sleep Behavior Disorder, and Cognitive Impairment

Author
Dr Paul Joy
Consultant – Radiology, Dr Gigy Varkey Consultant – Neurology and Interventional Neurology & Dr Ajith Toms Consultant – Radiology.
Corresponding Author
Dr Paul Joy
Institution- Rajagiri Hospital,
Address Dept of Radio diagnosis, Rajagiri hospital, Aluva, Ernakulum district, kerala

Abstract
Anti-IgLON5 antibody disease is a rare neurological disorder characterized by a combination of sleep dysfunction, neurodegeneration, and movement abnormalities. We report a case of a 62-year-old male who presented with excessive daytime sleepiness (EDS), bulbar weakness, and cognitive decline over three years. The patient exhibited REM sleep behavior disorder (RBD) with violent and complex movements during sleep, suggestive of a sleep-regulatory dysfunction. Clinical, polysomnographic, and serological findings strongly suggested anti-IgLON5 antibody disease. This case highlights the diagnostic challenges and the importance of early recognition and intervention in this emerging disorder.

Introduction

Anti-IgLON5 antibody disease is an autoimmune neurodegenerative disorder with a strong association to sleep-related dysfunction, bulbar symptoms, and movement disorders. First described in 2014, this rare entity has overlapping features with autoimmune encephalitis, multiple system atrophy (MSA), and progressive supranuclear palsy (PSP). The presence of IgLON5 autoantibodies correlates with tau pathology, making it a complex and often misdiagnosed condition. This case adds to the growing literature on its diverse clinical presentation and the role of early diagnosis.

Case Presentation

Patient History and Initial Presentation

A 62-year-old male presented with a progressive history of excessive daytime sleepiness (EDS) since 2021, along with severe fatigue, non-refreshing sleep, and early morning headaches. His sleep history, assessed via a sleep diary, revealed a consistent sleep-wake pattern (9 PM – 6 AM) but with persistent somnolence and cognitive impairment over the past three years. He also had bulbar symptoms, including dysphagia to both solids and liquids, nasal regurgitation, and frequent choking episodes. He had no history of depression but

reported intermittent episodes of joint pain and swelling, which responded to a short course of steroids in September 2024.

His past medical history was significant for obstructive airway disease. There was no history of bradykinesia, rigidity, or other parkinsonian features. His neurological symptoms included episodes of transient loss of consciousness, sudden sleep attacks, and abnormal sleep movements. His cognitive impairment primarily manifested as difficulty learning new information, suggesting bilateral medial temporal dysfunction.

Clinical Examination

On initial examination, the patient was conscious and oriented. Neurological examination revealed:

- **Cranial nerves:** No deficits noted.
- **Motor system:** Normal tone, power 5/5 in all four limbs, deep tendon reflexes 2+, and flexor plantar responses.
- **Cerebellar signs:** Subtle gait ataxia on tandem walking.
- **Sleep-related findings:** Finalistic and complex movements during sleep, including thrashing, picking, and vocalizations.

Investigations

- **Polysomnography (PSG, 27.01.25):** Poor sleep efficiency with absence of REM sleep.
- **Multiple Sleep Latency Test (MSLT, 28.01.25):** Mean sleep latency of 9 minutes (4 out of 5 naps); no sleep-onset REM (SOREM).
- **MRI Brain (previously done):** Essentially normal.
- **Cerebrospinal fluid (CSF) analysis:** Elevated CSF protein with normal cytology; no malignant cells.
- **Serum autoimmune encephalitis panel:** Negative for NMDA, VGKC, AMPA-1 & 2, GABA-B.
- **Echocardiogram (28.01.25):** Mild concentric left ventricular hypertrophy (LVH) with no regional wall motion abnormalities.
- **Endoscopic evaluation (FESS):** Significant dysphagia with pooling of saliva, high risk of aspiration.
- **Serum anti-IgLON5 antibody (30.01.25):** STRONGLY POSITIVE by both Tissue-Based Assay (TBA) and Cell-Based Assay (CBA).

Laboratory Findings

- **Thyroid function tests:** TSH 0.761 uIU/mL (within normal limits).
- **Parathyroid hormone:** 58.8 pg/mL (normal).
- **Inflammatory markers:** CRP 5.4 mg/L (mildly elevated).
- **Complete blood count:** Total WBC count 11.5 x10^3/µL (mild leukocytosis), neutrophilia (73.6%), relative lymphopenia.
- **Metabolic panel:** Normal renal, hepatic, and electrolyte levels.

Diagnosis

Based on the clinical, polysomnographic, and serological findings, a **probable diagnosis of anti-IgLON5 antibody disease** was made, with the following differential considerations:

1. Seronegative autoimmune encephalitis
2. Atypical Parkinsonism – Multiple System Atrophy (MSA-C)
3. Neurodegenerative sleep disorder spectrum

Early Diagnosis and Relevance

- **Biomarkers for early detection**: Identifying IgLON5 antibodies in serum/CSF can enable diagnosis before advanced neurodegeneration occurs.
- **Polysomnography in high-risk cases**: Patients with unexplained REM sleep disorder or bulbar dysfunction should undergo detailed PSG studies.
- **MRI and PET imaging**: Tauopathy-related changes may provide early diagnostic clues (Sabater et al., 2014).

- **Epidemiology and prevalence**: Still underrecognized, with a global prevalence yet to be established (Höftberger et al., 2015).

Hospital Course and Management

The patient was admitted for a detailed evaluation. Given his high risk of aspiration, a Ryle's tube was inserted, and enteral feeding was initiated. Symptomatic treatment included:

- **Tablet Modalert 100 mg (morning):** To manage excessive daytime sleepiness.
- **Tablet Ecosprin AV 75 mg (night):** Cardiovascular protection.
- **Tablet Quicnac AB (twice daily):** Anti-inflammatory support.
- **Calamine lotion & Acivir ointment:** For symptomatic relief of skin lesions.

Given the confirmed anti-IgLON5 positivity, plans were made for **immunomodulatory therapy**, including IV immunoglobulins (IVIG) or plasmapheresis. The patient's relatives requested discharge with a plan to return for aggressive immunotherapy and further evaluation.

Discussion

Anti-IgLON5 disease presents with an overlapping spectrum of sleep disorders, neurodegeneration, and bulbar dysfunction. The presence of strongly positive IgLON5 antibodies, in combination with sleep dysregulation and cognitive impairment, makes this a striking case. The absence of parkinsonian features or rigidity differentiates it from other neurodegenerative disorders like MSA or PSP. This case highlights the importance of recognizing IgLON5 disease in patients presenting with combined sleep and neurological symptoms, as early immunomodulatory intervention can potentially alter disease progression.

Source of Support: None

References

1. Honorat JA, Komorowski L, Josephs KA et al. IgLON5 antibody Neurological accompaniments and outcomes in 20 patients. Neurology-Neuroimmunology Neuroinflammation. 2017; 4(5): e385.
2. Sabater L, Gaig C, Gelpi E, et al. A novel non-rapid eye movement and rapid eye movement parasomnia with sleep breathing disorder associated with antibodies to IgLON5. Lancet Neurol. 2014; 13(6): 575-586.
3. Höftberger R, Titulaer MJ, Sabater L, et al. Encephalitis and tauopathy in IgLON5 antibody disease. Brain. 2015; 138(Pt 6): 1583-1595.
4. Cortese I, Giannoccaro MP, Antelmi E, et al. Treatment response in anti-IgLON5 disease: A multicenter international study. *J Neurol* 2022;269(12):6592–606. doi:10.1007/s00415-022-11300-7.