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A Case of Kennedy's Disease Presented as Chronic Dysphagia Mimicking Sjogren's Syndrome

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INTRODUCTION

• X-linked recessive spinal and bulbar muscular atrophy (SBMA), also known as Kennedy's disease, is a rare lower motor neuron disorder. Due to its insidious onset and slowly progressive nature, it is frequently misdiagnosed as more common conditions, such as gastroesophageal reflux disease (GERD), Sjogren's syndrome, or degenerative spinal pathologies.

• We report a case where a six-year history of chronic dysphagia was ultimately diagnosed as SBMA, highlighting the critical importance of identifying subtle bulbar lower motor neuron signs.

Case Presentation

• A 59-year-old male presented with a six-year history of progressive dysphagia, primarily localized to solid foods. His symptoms had been refractive to previous treatments for suspected GERD and Sjogren's syndrome, the latter of which was considered due to his "liquid wash" requirement.

• Neurological examination revealed preserved limb strength (MRC grade V/V) and no overt gait imbalance, yet deep tendon reflexes were globally hypoactive (1+). Physical examination was notable for prominent facial and tongue fasciculations, tongue atrophy, and mild dysarthria. Further history revealed that his maternal grandfather had experienced similar progressive bulbar symptoms, strongly suggesting an X-linked recessive inheritance pattern.

• Laboratory findings showed significantly elevated serum creatine kinase (2,992 IU/L; Dec 2025) and high testosterone levels (7.49 ng/mL; Apr 2023), indicating androgen insensitivity.

• A video-fluoroscopic swallowing study (VFSS) performed in July 2025 demonstrated impaired tongue base retraction and significant vallecular residue after solid ingestion, consistent with bulbar muscle weakness (Figures 1–2). Confirmatory genetic testing identified an abnormal expansion of 45 CAG repeats in the androgen receptor gene.

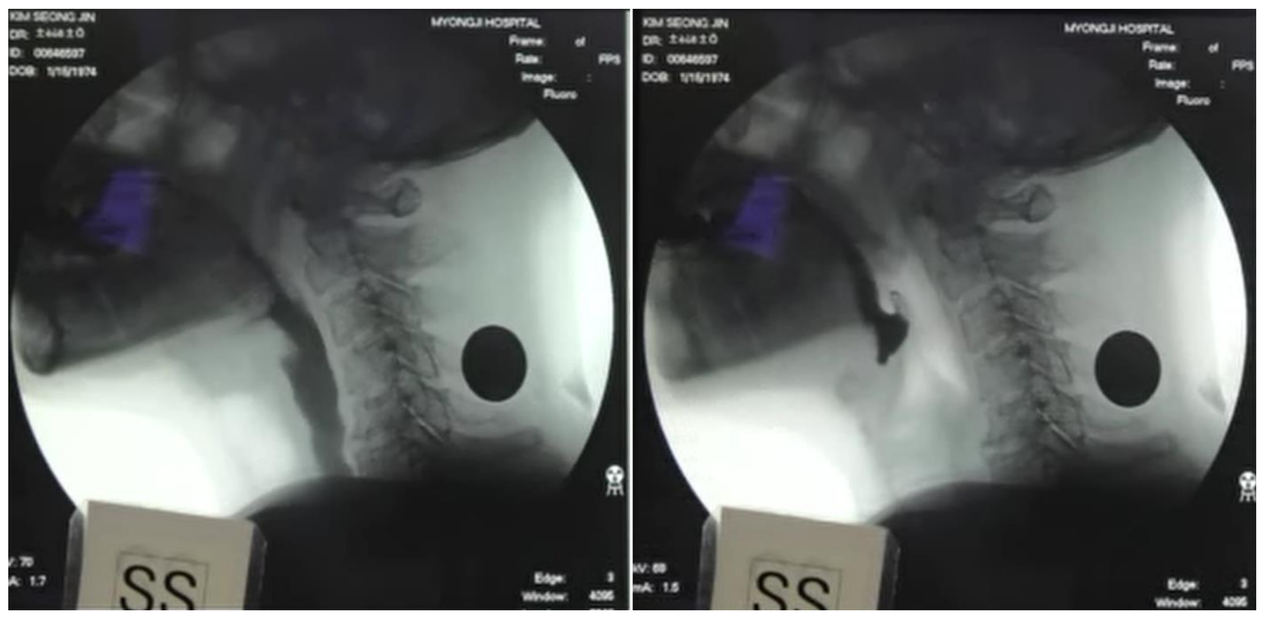

Figure 1. 25.07.31 VFSS image (Semisolid 4cc during-swallow)
Video-fluoroscopic swallowing study (VFSS) taken on July 31, 2025, demonstrating post-swallow remnent of semisolid (4cc) diet during swallowing, dominantly in vallecular space.

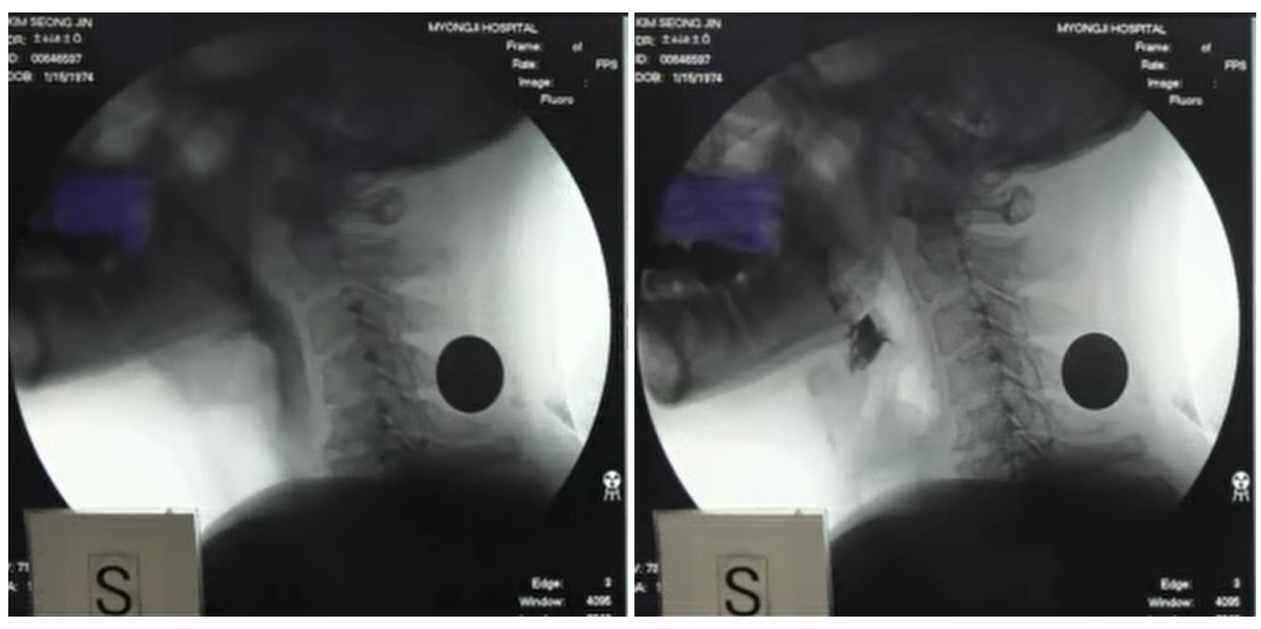

Figure 2. 25.07.31 VFSS image (Solid during-swallow)
Video-fluoroscopic swallowing study (VFSS) taken on July 31, 2025, showing significant bolus residue in vallecular fossa after swallowing solid diet.

CONCLUSION

•This case illustrates that chronic dysphagia in male patients should not be prematurely attributed to common structural or systemic causes. The triad of bulbar fasciculations, elevated CK, and high testosterone levels serves as a diagnostic "red flag" for Kennedy's disease.

• Clinicians must exercise caution to prevent incidental degenerative spinal findings from leading to inappropriate surgical interventions in patients with unrecognized motor neuron disease.

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