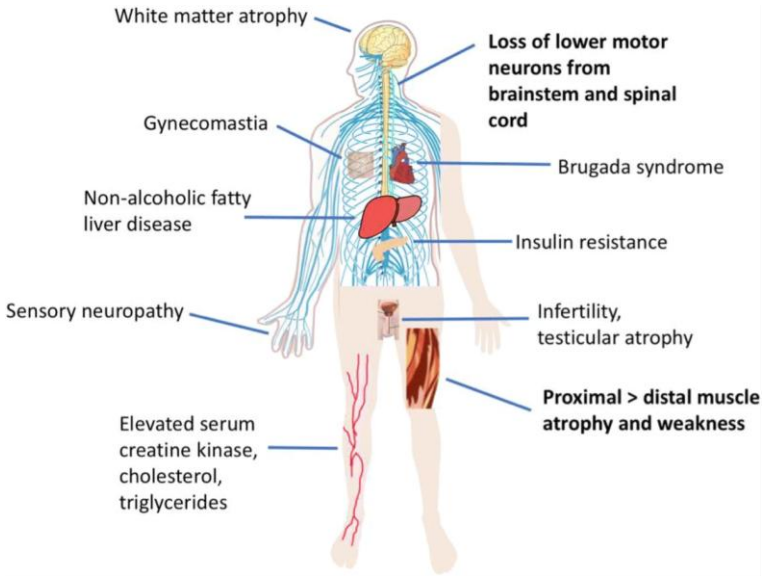
A Diagnostic Odyssey in Lower Motor Neuron Disorders: Recognizing Kennedy Disease

Tamil Kumaran.G, Aubin Mathew Varghese, Nandakumar.V,
Viveka Saravanan.R, Mugundhan.K
MMC & RGGGH, Chennai

INSTITUTE OF NEUROLOGY - MADRAS
1950

INTRODUCTION:

Kennedy Disease, or Spinal and Bulbar Muscular Atrophy (SBMA), is a rare X-linked recessive lower motor neuron disorder caused by a CAG repeat expansion in the androgen receptor gene. It typically presents with progressive proximal muscle weakness, bulbar symptoms, and characteristic fasciculations, often misdiagnosed in early stages.



CASE REPORT

- A 53-year-old male presented with progressive difficulty in walking for 10 years, followed by proximal upper limb weakness, tremors, and bulbar symptoms including slurred speech and difficulty chewing with twitching of proximal muscles.
- No significant family history
- Neurological examination revealed flaccid quadriparesis with proximal muscle wasting and fasciculations, postural hand tremors, head tremor (no-no type), tongue weakness with atrophy and fasciculations, with preserved sensory and cerebellar functions. He also had gynecomastia on general examination.
- Laboratory results were unremarkable with mildly elevated CPK.
- Electrophysiological studies demonstrated a neurogenic EMG pattern with normal nerve conduction studies. Repetitive nerve stimulation showed a decremental response.
- USG abdomen and testis and ECG were normal.
- Genetic confirmation of expanded CAG repeats in the androgen receptor gene established the diagnosis of SBMA.

DISCUSSION:

In this case, the combination of progressive proximal weakness, muscle fasciculations, postural tremors, bulbar involvement, and preserved sensory function raised suspicion for SBMA. Normal nerve conduction studies with a neurogenic EMG pattern and decremental response on RNS supported a motor neuron pathology with neuromuscular junction fatigue. Genetic testing confirming CAG repeat expansion in the androgen receptor gene established the diagnosis.

CONCLUSION:

- This case underscores the diagnostic challenge of Kennedy Disease due to its overlapping features with myopathy, myasthenia, and MND.
- Key clinical clues like tremor, tongue fasciculations, and androgen insensitivity-related features should raise suspicion. EMG and genetic testing remain the cornerstone for confirmation.
- Early recognition can guide appropriate management and genetic counseling.

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