

DIFFERENTIAL DIAGNOSIS BETWEEN BULBOSPINAL MUSCULAR ATROPHY - KENNEDY'S DISEASE AND AMYOTROPHIC LATERAL SCLEROSIS

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Amyotrophic lateral sclerosis (ALS) includes a number of disorders causing degeneration of lower and upper motor neurons and findings in the bulbar region and at least two spinal regions or UMN and LMN in three spinal regions. ALS is typically presented with bulbar or asymmetric limb weakness, loss of ability to speak, to swallow and to breathe. Kennedy disease is a form of MND that is associated with bulbar involvement and X linked recessive inheritance. The symptoms include muscular cramps, a limb-girdle distribution of muscle weakness, bulbar symptoms and distinguishing clinical features include facial and perioral fasciculations in particular. This is a case study of a 43-year-old man who suffered from recurrent muscle cramping and progressive symmetric lower extremity weakness, with a prominent fatigable component to this weakness, shoulder weakness, difficulty swallowing and facial twitching. EMNG showed widespread reinnervation changes and fasciculations in arms, legs, tongue, and thoracic paraspinals, with minimal fibrillation potentials. Motor NCS were normal or borderline in amplitude, and sensory responses were absent in upper and lower limbs. Genetic testing was positive (45 CAG). Kennedy disease may be underdiagnosed, owing in part to misdiagnosis and to the mild symptoms exhibited by some patients. The electro-physiological examinations are the key point to the diagnosis of Kennedy disease. In our case, we found the symmetric weakness, sensory abnormalities on electrophysiological testing, prominent facial fasciculations, and gynecomastia which were not characteristic of the ALS, and indicated that was one of MND syndromes such as Kennedy disease and that was confirmed by genetic testing.

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