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ANALYSIS AND THE IMPORTANCE OF THE DIFFERENTIAL DIAGNOSIS BETWEEN KENNEDY'S DISEASE AND AMYOTROPHIC LATERAL SCLEROSIS

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INTRODUCTION

Male, 57 years old felt weakness in the right upper limb in rural work, dropping the instrument used five years ago. Three years ago he presented difficulty in flexion of the right index finger and atrophy of the right upper limb, diagnosed with ALS. The symptoms started on the right side and progressed to the left side. He also reports polyuria, urgency, cries easily, imbalance and falling, night sweats and coldness in the upper limbs. History of 10kg loss and use of Riluzol 50mg. On physical exam he presented grade 3 strength deficit in the four limbs, fasciculations in the right upper limb, upper back and tongue that presented atrophied right muscles, interosseous atrophy of hands, hyporeflexia and staggering gait.

OBJECTIVES

- Correlate amyotrophic lateral sclerosis with kennedy's disease
- Analyze the clinical manifestations of the case
- Work on the differential diagnosis of Kennedy's disease

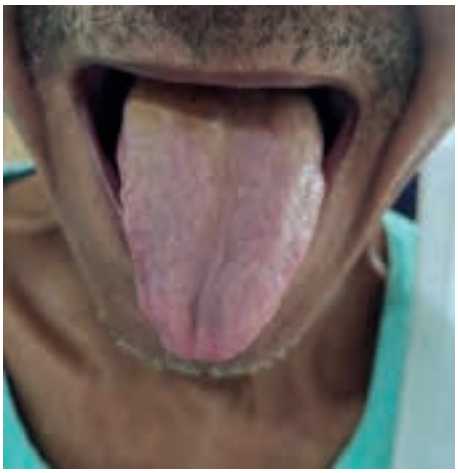
METHODS

Clinical discussion of the case, interview with the patient, documentation of clinical signs and analysis of information in the literature.

RESULTS

Spinal and bulbar muscular atrophy (SBMA) or Kennedy's disease, is a rare, X-linked, inherited lower motor neuron disease. It is characterized by progressive muscle weakness. An expanded trinucleotide repeat (CAG > 37) in the androgen receptor gene which encodes glutamine, is the responsible mutation (FISCHBECK, K. H., 2016).

Toxicity of this mutant protein affects both motor neurons and muscles. The main symptoms are weakness in the lower limbs, more proximal than distal, hyporeflexia, fasciculations, and muscle atrophy. Non-motor symptoms include dysphagia, dysarthria, testicular atrophy, Brugada syndrome, uriary disorders, and dementia due to reduced frontal brain volume (ATSUTA, WATANABE, et al, 2018). Amyotrophic Lateral Sclerosis is the main differential diagnosis and is ruled out by the absence of hyperreflexia and sensory changes. Early diagnosis is important because the prognosis is related to the risk of weakness-related falls and bronchoaspiration, and sudden death from Brugada Syndrome (NISHIMURA, MITNE-NETO, et al, 2004).



CONCLUSION

SBMA is not only a lower motor neuron disease, but a complex disorder affecting different systems, including the central nervous system. The differential diagnosis with (ALS) is of utmost importance, since the age of presentation of SBMA is 30-40 years and the patient is already in a very advanced picture at diagnosis.

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