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Sympathetic skin response in Kennedy disease patients

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Background

Spinal and bulbar muscular atrophy (SBMA), also known as Kennedy's disease is a rare X-linked neuromuscular disease, caused by a CAG repeat expansion >35CAGs in the exon 1 of the androgen receptor gene.

SBMA classically manifests with lower-motor neuron symptoms. Patients commonly present with muscle cramps, tremor, leg weakness, dysarthria and dysphagia.

Autonomic nervous system involvement in SBMA is not well studied nor fully known.

Sympathetic skin response (SSR) is used in diagnosing the impairment of non-myelinated sympathetic fibres in peripheral neuropathies.

Aim of the study

Investigate the autonomic nervous system's involvement in SBMA by using sympathetic skin response test.

Methods

All Kennedy disease patients in Latvia (n=5), unrelated Caucasian male patients, aged 34 to 68.

We deeply phenotyped all patients and carried out sympathetic skin response test as a non-invasive approach to investigate the sympathetic system, as well as nerve conduction studies.

Results

Neurological examination revealed **typical manifestations of lower-motor neuron damage**.

Motor and sensory nerve conduction velocities and compound muscle action potentials and sensory action potential amplitudes were abnormal for 2 out of 5 patients indicating **peripheral large nerve fibre neuropathy**.

All patients had complaints about **sweating disturbances:**

- 3 patients complained about increased sweating;
- 2 patients about decreased sweating.

Sympathetic skin response test results indicated that 3 of 5 patients had **peripheral sympathetic nervous system dysfunctions:**

- All of those 3 patients demonstrated an increased latency;
- And 2 of them also had a decreased amplitude during examination.

Conclusions

Our data supports the evidence that Kennedy disease is multisystemic disease with peripheral nervous system involvement, including sympathetic nervous system.

However, our study group size was limited, and bigger SBMA patient cohorts should be evaluated for autonomic nervous system involvement.