

## **Massachusetts Neurologic Association**

March 25, 2017

### **CALL FOR ABSTRACTS**

Deadline for submission: March 10, 2017

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The Poster Presentation Session at the MNA Spring Meeting aspires to allow residents and fellows in training to present their research or interesting cases in a poster session. Please follow the following abstract guidelines:

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Title: Evidence of Small-fiber Neuropathy in two patients with unexplained Genital Sensory loss and Sensory Urinary Cystopathy

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Abstract:

(There is a limit of **350** words for your abstract submission).

**Objective:**

To describe an atypical presentation of biopsy-proven small fiber neuropathy (SFN). The chief manifestations are genital and perineal sensory loss with a sensory urinary cystopathy.

**Background:**

In the past few years many different presentations of SFN have been reported, including attempts to link SFN to syndromes of unclear pathogenesis such as fibromyalgia and complex regional pain syndrome. To our knowledge, sensory loss predominantly affecting genital and perianal areas has not been previously reported.

**Design:**

Case series of two patients with atypical presentation of SFN.

**Results:**

Two young men, 20 and 29 years old, presented with gradually progressive sensory loss of the genital area symmetrically involving the penile, perineal and peri-anal skin bilaterally. Both men also had a sensory cystopathy on urologic studies. Electromyography (EMG) and nerve conduction studies (NCS) in the limbs were normal. MRI of the lumbar spine, pelvis and lumbar plexus were unrevealing for a structural etiology. Microscopic examination of PGP9.5-immunolabeled thick sections from a punch skin biopsy from distal leg showed reduced density of the epidermal nerve endings (less than the 3<sup>rd</sup> percentile of normal). One individual was positive for a variant of uncertain significance in the LRSAM1 gene responsible for Charcot-Marie-Tooth type 2P, but given the very different phenotype, the relevance of the mutation to this syndrome is unclear. Laboratory testing for endocrine, nutritional, infectious and inflammatory etiologies of SNF was otherwise unrevealing.

**Conclusion:**

The etiology of genital sensory disturbances often remains undetermined and is frequently attributed to psychogenic factors. Given the novelty of this syndrome, the frequency of underlying SFN in otherwise unexplained genital numbness is unknown. Identification of SFN may yield insights into the anatomical basis for some such cases including a basis for the sensory cystopathy.

