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CASE REPORT

Spinal Cord Compression in a Patient with Hereditary Sensory and Autonomia Neuropathic by Silent Spondylodiscitis

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ABSTRACT

Study Design. Case Report
Objective. To report a case of lumbar spinal cord compression caused by spondylodiscitis in a patient with Hereditary Sensory Autonomia Neuropathic (HSAN) type 3.
Summary of Background Data. To our knowledge, there have been no previous reports of spinal cord compression in patients with HSAN type 3 by spondylodiscitis.
Methods. A 53 years-old male with HSAN type 3 underwent surgical decompression L4-L5 and posterior fusion L3-S1 because of spinal cord compression due to silent spondylodiscitis. Medical history, clinical and laboratory findings, and imaging exams of the preoperative and postoperative course were collected and documented.
Results. The patient had resolution of his neurologic symptoms postoperatively. The histologic exam revealed chronic spondylodiscitis. Antibiotic treatment had been given for 4 weeks. The patient was free of symptoms and infections after 6 months of follow-up.
Conclusions. One must have high clinical suspicion during the clinical assessment of patients with HSAN because the hereditary loss of pain could cover a severe pathologic progress.

KEY WORDS. Hereditary Sensory Autonomia Neuropathic, Familial dysautonomia, syndrome Riley-Day, spinal cord compression.

Introduction

The rare "congenital insensitivity to pain" syndromes has been differentiated into rare hereditary sensory and autonomic neuropathies (HSAN) [1,2,3]. Five different entities (HSAN I-V) have been described [4]. Their incidence has been estimated to be about 1 in 25,000 [5].

Type I is transmitted as autosomal dominant trait and is characterized by a sensory deficit in the distal portion of the lower extremities, chron-

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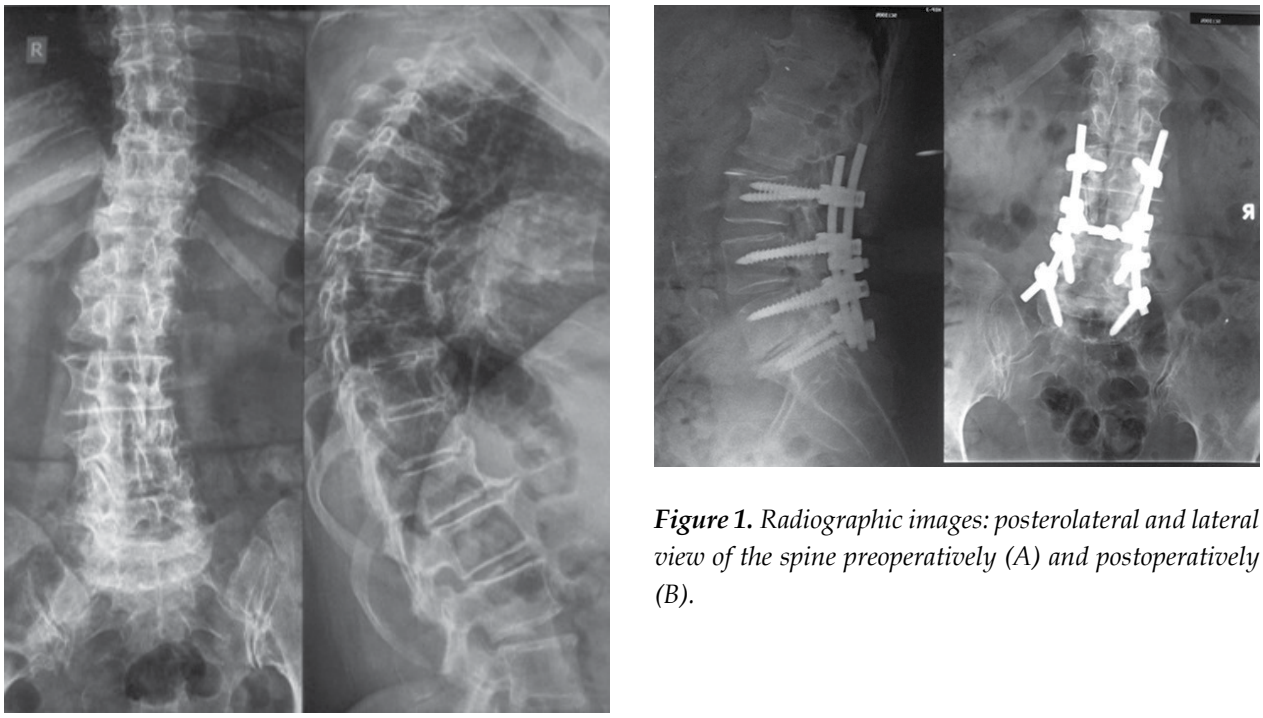

Figure 1. *Radiographic images: posterolateral and lateral view of the spine preoperatively (A) and postoperatively (B).*

ic perforating ulcerations of the feet and progressive destruction of the underlying bones. Symptoms appear in late early adolescence. Histologic examination reveals a marked reduction in the number of unmyelinated fibers.

Type II is transmitted as autosomal recessive trait and is characterized by pain of upper and lower extremities affected with chronic ulcerations and multiple injuries to fingers and feet. Autoamputation of the distal phalanges is common. Symptoms appear in early infancy. Histologic examination reveals total loss of myelinated fibers and a reduced number of unmyelinated fibers.

Type III (familial dysautonomia, Riley-Day syndrome) is transmitted as an autosomal recessive trait seen predominantly in Jew descents of Eastern Europe. Newborns have weak sucking reflex, hypotonia and hypothermia. Other features include absence of the sense of pain, reduced or absent tears, depressed deep tendon reflexes, absent corneal reflex, absence of fungiform papillae of the tongue and postural hypotension. Scoliosis is frequent. Histologic examination reveals a reduction in the number of myelinated and unmyelinated fibers. Recently the responsible gene was localized in the long arm of chromosome 9(9q31)(7,22) [6].

Type IV is transmitted as an autosomal recessive trait. In infants, it is expressed with hyperthermia, anhidrosis and insensitivity to pain. Histologic examination reveals a small reduction in the number of unmyelinated fibers.

Type V has similar clinical features to type IV but there is a selective absence of myelinated fibers.

Cardiovascular, pulmonary and gastrointestinal manifestations may be severe and lead to early death. The insensitivity to pain may mask symptoms of underlying severe pathologic conditions such as in the reported case.

Case Report

A 53 years old man was admitted because of claudicatio intermittens. He was able to walk about 150 meters. He suffered of familial dysautonomia. He had an intravenous antibiotic treatment for spondylodiscitis L4/L5 twelve years ago. At that instance, histological culture revealed Staphylococcus aureus before antibiotic treatment. On clinical examination he could walk on heals and

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toes. The muscular power of the lower limb was 4/5. In both legs, Lasegue sign was negative. Sensory sense was very decreased. There were multiple amputations of the distal phalanges of both upper and lower limbs. There was an important dental loss because of recurrent caries. Blood count exams, VES and CRP were normal on admission. PET revealed increased signal in L4/L5 disc. Magnetic resonance imaging revealed severe spinal stenosis at L4/L5 level. The patient underwent surgical decompression of L4/L5 and L3-S1 posterior fusion with instrumentation (**Fig.1**). Postoperatively the patient had hyperpyrexia for 3 days. The hyperpyrexia resolved spontaneously. His neurologic symptoms also resolved. Intraoperative cultures did not reveal any microorganism. Percutaneous disc biopsy revealed chronic inflammation. Appropriate intravenous antibiotic treatment was given for 4 weeks. The patient was free of symptoms 6 months after the operation.

Discussion

The HSAN type III (familial dysautonomia, Rilley-Day syndrome) is a disorder caused by incomplete maturation of the unmyelinated neurons of the sensory pathway, the sympathetic and part of the parasympathetic system [7]. Pathological findings indicate that within the peripheral sensory and autonomic system, individuals affected by HSAN type III have incomplete neuronal development, as well as progressive neuronal degeneration. The autonomic dysfunction results in protein functional abnormalities, also affecting other systems, and yielding myriads of clinical manifestations which include orthostatic hypotension without compensatory tachycardia, episodic hypertension, oropharyngeal incoordination, gastrointestinal dysmotility, excessive sweating, and absence or overflow of emotional tearing [8]. Recurrent infections occur in these patients. Severe infections may have subclinical symptoms because of decreased sense of pain.

The case described suffered from recurrent infections in various parts of the body. Repeated trauma, and infections at the level of upper and lower limbs resulted in multiple amputations of the distal phalange. Recurrent caries resulted in severe dental loss. Spondylodiscitis caused severe spinal stenosis. Preoperatively, the patient was not able to walk more than 150 meters because of loss of his muscular power. After spinal decompression the patient had a gradual gait improvement.

Conclusions

The decreased sense of pain in patients affected by HSAN may mask severe underlying pathologic conditions and hazard their health status. High clinical suspicion is needed during assessment of these patients.

Key Points

- A patient affected by Hereditary Sensory and Autonomia Neuropathic (HSAN) had a gradual loss of his walking capacity.
- In these patients, there is a partial or complete loss of the sense of pain. That condition may mask severe pathological conditions.
- In this patient, MRI revealed severe spinal stenosis due to spondylodiscitis.
- After spinal decompression the patient had a gradual gait improvement.

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