# Hereditary sensory and autonomic neuropathy type 3 in non-Jewish child

## Neuropatia hereditária sensitivo-motora tipo 3 em uma criança não judia

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Hereditary sensory and autonomic neuropathy type 3 (HSAN3), also known as familial dysautonomia or Riley Day syndrome, is an rare hereditary autosomal recessive anomaly characterized by impairment of sensory and sympathetic innervation. The prevalence is 1:10,000 to 1:20,000 Ashkenazi Jews newborns. This condition is considered a rare condition among non-Jewish children[1-3].

## CASE REPORT

The patient was a 16 year-old girl, who presented no complications at birth and had no known disease in her family. Her parents were not consanguineous and had no-Jewish ancestry.

When she was two months old, she started presenting bronchial hyper reactivity, pneumonias, chronic diarrhea, and malnutrition that took her to several hospitalizations. By 11 months old, she started presenting an irritable behavior and episodes of self-harm, when she would bite her lips, tongue, hands and feet, which left permanent scars on these places (Fig 1). In order to stop the self-caused injuries, her teeth had to be removed. At that time she was noted to present generalized hypotonia and developmental delay; she was found to show no perception of pain or miotact reflexes.

From three to six years old, she went through many hospital admissions due to distal phalanges osteomyelitis (caused by herself-mutilation) and/or recurrent bone fractures (caused by frequent falls). In one admission, burn scars on the trunk and limbs where also noted which brought up the hypothesis that the patient would present no tactile and thermal sensitivity.

At eight years old, she started presenting some orthopedic changes (kyphoscoliosis and mandibular protrusion).

Currently, she presents a mild cognitive impairment, hypotonia, abolition of miotatic reflexes, global analgesia, absence of tactile and pain sensitivity. Besides, she presents with short stature, postural hypotension, decreased tear volume, absence of fungiform papillae on the tongue, chronic diarrhea, and acrodystrophy in hands (Fig 2) and feet.

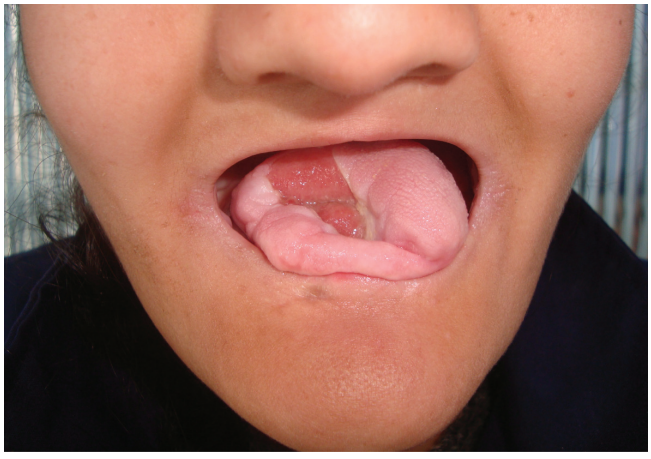

Fig 1. Smooth tongue, presenting a tongue scar and mandibular protrusion.

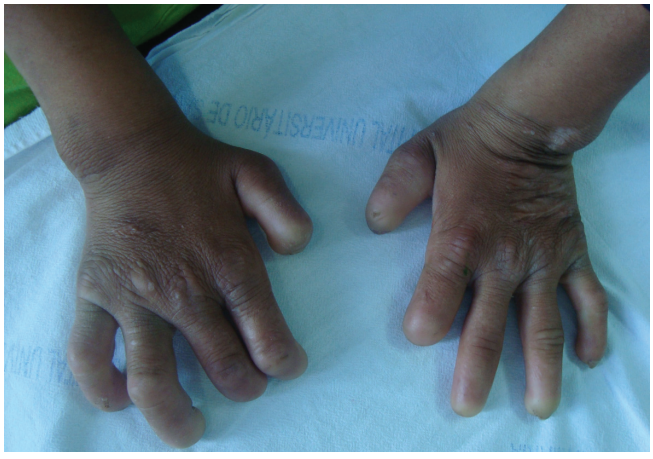

Fig 2. Acrodystrophy in hands.

She had undergone an electromyography, which showed reduced motor conduction velocity, reduced compound motor action potential amplitudes and absent sensory nerve action potentials.

## DISCUSSION

This disorder is a progressive sensorimotor neuropathy. The sympathetic autonomic dysfunction is considered the cause of the majority of the clinical manifestations[4,5]:

- Episodes of dysautonomic crises, characterized by nausea, vomiting and symptoms of sympathetic storm (irritability, tachycardia, hypertension, facial flushing, bronchorrhea and diminished oral coordination leading to swallowing and speech dysfunction);
- Additional autonomic symptoms include orthostatic hypotension, excessive salivation, gastrointestinal motility dysfunction, bladder dysfunction, decreased or absent tearing, pupil dilation, hypohidrosis, episodic hyperhidrosis and blotchy skin;
- Glomerulo sclerosis and chronic kidney disease unrelated to autonomic dysfunction;
- Neuropathic symptoms: loss of reflexes, hypotonia, decreased perception of pain and temperature;
- Additional clinical features: short stature, kyphoscoliosis, smooth tongue (no fungiform papillae), vomiting, recurrent aspiration, multiple skin scars, dysarthria, mental deficiencies, and emotional lability.

Linkage studies map to a candidate region on chromosome 9q31. Genetic evaluation is considered sensitive and specific for the diagnosis of HSAN3. Furthermore, it's the only specific diagnostic test currently available.

No particular treatment for familial dysautonomia has yet been described. Supportive care and symptomatic therapies are the main stay of management. The prognosis is poor.

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# Subcostal schwannoma in pregnancy

## Schwannoma subcostal na gravidez


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**in memoriam*.

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The schwannoma is a supporting cells tumor of peripheral nervous system, characterized by the formation of grayish, firm, circumscribed and usually solitary masses, which is located close to the roots of cranial and spinal nerves. Most of them have a benign character[1].

## CASE REPORT

A 27-year-old Caucasian female, with pregnancy at term, who was subjected to routine ultrasound, which observed the presence of hyperechoic lesion at the left hypochondrium