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## RILEY-DAY SYNDROME

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**ABSTRACT:** Riley-Day syndrome is a hereditary sensory and autonomic neuropathy type III (HSAN III) resulting in insensitivity to pain, inability to produce tears, poor growth and labile blood pressure. It is caused by mutations in IKBKAP gene on chromosome 9. On clinical diagnosis no fungiform papillae on the tongue, there is deep tendon reflexes and lack of an axon flare, genetic test denotes presence of chromosome 9. Treatment of Riley-Day syndrome is preventative, symptomatic, and supportive. Medicines are used to control vomiting, eye dryness, and blood pressure. Central nervous system degeneration have a poor long term prognosis.

**Key word:** Syndrome, Neuropathy, Mutations, Chromosome, Fungiform Papillae

### INTRODUCTION:

Riley Day syndrome is a type of autonomic sensory neuropathy of hereditary genetic origin that is part of peripheral neuropathies, resulting in dysfunction of the sensory and autonomic nervous structures as a consequence of a genetic alteration. Riley Day syndrome presents a high rate of morbidity and mortality. However, although there are no curative treatments, can improve the medical prognosis, survival, and quality of life of affected individuals.

### DEFINITION

Riley-Day syndrome is an autonomic sensory neuropathy of origin Heredity that generalizes nervous affection that results in an autonomic and sensorial dysfunction inherited disorder that affects nerves throughout the body. Riley-Day syndromes also known as family dysautonomia, hereditary sensory and autonomic neuropathy - type III (HSAN III).

### CAUSES

Riley-Day syndrome is passed down through families (inherited). A person must inherit a copy of the defective gene from each parent to develop the condition. This condition occurs most often in people of Eastern European Jewish ancestry (Ashkenazi Jews). It is caused by a change (mutation) to a gene. It is rare in the general population.

## CLASSIFICATION

There are different types of peripheral neuropathies:

- Motor neuropathy
- Sensory neuropathy
- Autonomic neuropathy
- Mixed or Combined Neuropathy (Foundation for Peripheral Neuropathy, 2016)

According to the function of the type of nerve fibre that is affected:

- Motor nerves
- Sensory nerves
- Autonomic nerves

## SYMPTOMS

- Breath-holding spells (can lose consciousness)
- Constipation
- Decrease in sense of taste
- Diarrhoea
- Dry eyes
- Feeding problems
- Inability to feel pain and changes in temperature (can lead to injuries)
- Lack of tears when crying
- Long periods of vomiting
- Poor coordination and unsteady walk
- Poor growth
- Repeated fevers
- Repeated [pneumonia](#)
- [Seizures](#)
- Skin blotching
- Sweating while eating
- Unusually smooth, pale tongue surface

## INVESTIGATIONS

- Absent or decreased deep tendon reflexes
- Lack of a response after receiving a histamine injection (normally redness and swelling would occur)
- Lack of tears with crying
- Low muscle tone, especially in babies
- Repeated episodes of [high blood pressure](#)
- Severe curving of the spine ([scoliosis](#))
- Tiny pupils after receiving certain eye drops
- Blood tests are for the gene mutation that causes Riley-Day syndrome.

## TREATMENT

- Therapy to help prevent seizures
- Feeding in an upright position and giving textured formula to prevent [gastroesophageal reflux](#) (stomach acid and food coming back up, also called GERD)
- Measures to prevent low blood pressure when standing, such as increasing intake of fluid, salt, and caffeine, and wearing elastic stockings
- Medicines to control vomiting
- Medicines to prevent dry eyes
- Physical therapy of the chest
- Measures to protect against injury
- Providing enough nutrition and fluids
- Surgery or spinal fusion
- Treating aspiration pneumonia

## PROGNOSIS

Advances in diagnosis and treatment are increasing the survival rate. About half of babies born with Riley-Day will live to age 30.

## COMPLICATIONS

- Blotching of the face and torso
- Excessive sweating of the head and torso
- High blood pressure and rapid heart rate
- Insomnia
- Irritability
- Patchy skin tone on the hands and feet
- Nausea and vomiting
- Severe problems swallowing, drooling
- Worsening of muscle tone

## PREVENTION

Genetic DNA testing is very accurate for Riley-Day syndrome. It may be used for diagnosing people with the condition or those who carry the gene. It can also be used for prenatal diagnosis.

People of Eastern European Jewish background and families with a history of Riley-Day syndrome may wish to seek genetic counselling if they are thinking of having children.

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