# Non-union of fractures in Riley Day Syndrome

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## ABSTRACT

Riley Day Syndrome, also known as familial dysautonomia, is a rare reported entity characterised by disturbance of pain and temperature perceptions, inability to produce tears, labile blood pressure and poor growth due to disorder of the autonomic and sensory nervous system. It is an autosomal recessive condition with the genetic locus mapped to chromosome $9^{q31 - q33}$. Traumatic fractures are common and due to lack of pain, may go unrecognised for prolonged periods of time, resulting in nonunion or pseudoarthrosis. Scoliosis is seen in up to 90% of the patients. Complications of are common in these patients and range from infection to wound breakdown to failure of fixation. We report a case (nine-year-old girl) of Riley Day Syndrome with general absence of pain and damage to the extremities to highlight this rare syndrome.

**Keywords: Pseudoarthrosis, familial dysautonomia, non union**

## INTRODUCTION

Riley Day Syndrome is a rare autosomal recessive disorder presenting at birth. [1, 2] It is of orthopaedic interest because of frequent nonunion and pseudoarthrosis of fractures. These patients have hypoactive or absent tendon reflexes. They are insensitive to pain and because of this they may be unaware of sprains and wounds and may develop Charcot's joints. [3]

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## CASE REPORT

A nine-year-old girl presented with a painless and deformed right elbow of five year duration and a painless and deformed right ankle over the past three months (Figure 1).

There was a history of consanguinous marriage of the parents, similar complaints in the sibling, lack of tears, anhydrosis, recurring bouts of fever and constipation. On examination there was generalised absence of pain with deformed right proximal forearm with pseudoarthrosis and a deformed right ankle joint. The patient had a Trendelenburg's gait and shortening of the right lower limb by three centimetres. The girl had a bifid tongue,

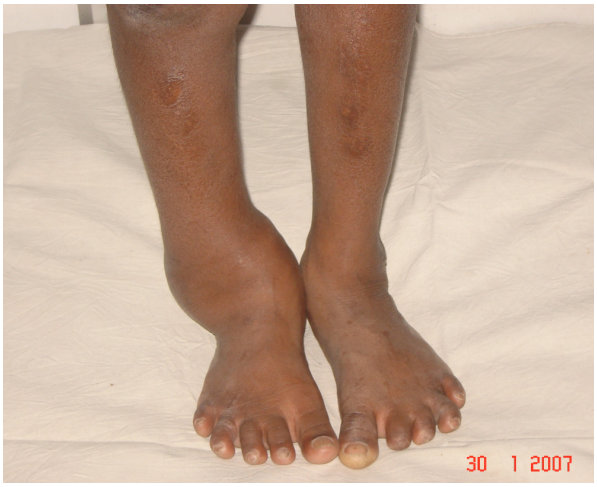


**Fig. 1: Painless deformed right ankle secondary to non-union of fracture.**

absence of most of the teeth, lack of response to painful stimulus, diminished temperature perception, normal touch perception, diminished deep reflexes, a fluctuating blood pressure, multiple scars over the body, scoliosis and ligamentous laxity. Radiographs of the patient revealed a nonunited displaced bimalleolar fracture with joint subluxation of the right ankle joint (Figure 2) and atrophic nonunion of proximal end fracture of both the bones of the right forearm.

Injection of 0.1 ml of 1:1,000 solution of histamine intra-dermally produced a wheal but no pain and no axon flare around it. Nerve conduction velocity was normal. The serum creatinine phosphokinase was elevated and the rest of the investigations were within normal limits. The patient underwent open reduction and internal fixation with malleolar screws for medial malleolar fracture of the right ankle and Kirschner wire stabilisation of the right lateral malleolus fracture under spi–

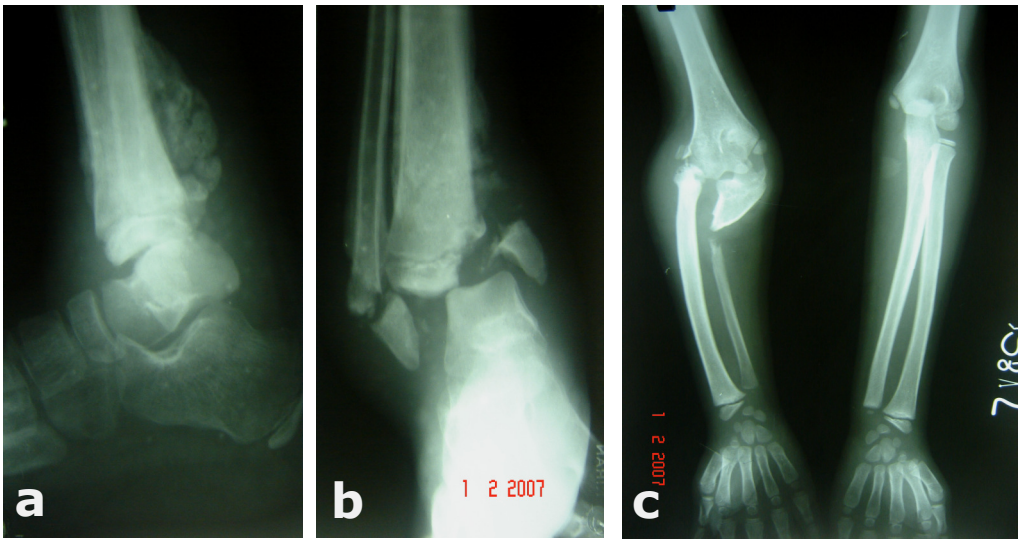


**Fig. 2: Radiographs showing a) and b) a non-union and displaced bimalleolar fracture with joint subluxation of the right ankle joint and c) showing an atrophic nonunion of proximal end fracture of both the bones of the right forearm.**

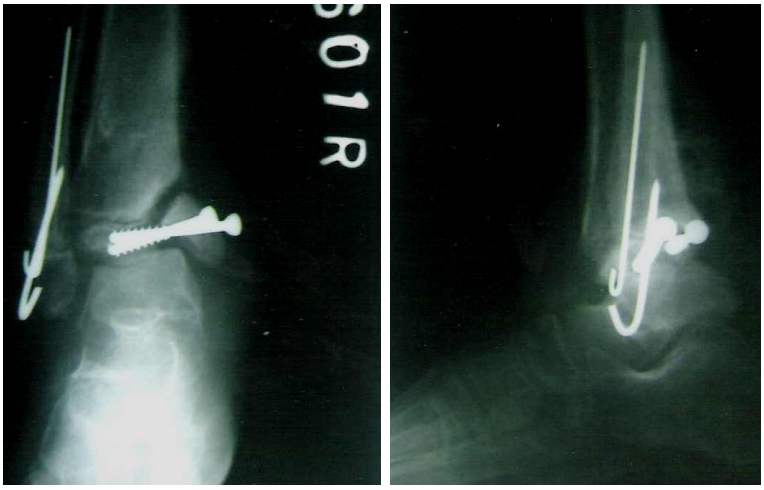

**Fig. 3: Painless deformed right ankle secondary to non-union of fracture.**

nal anaesthesia (Figure 3). Post-operatively, the right ankle joint was immobilised in a below knee slab for six weeks. Intra-operative and post-operative recovery was uneventful. However there were no radiological signs of fracture healing after twelve months. The pseudoarthrosis of the proximal right forearm was left alone without any operation.

## DISCUSSION

Riley Day Syndrome, also known as the Hereditary Sensory and Autonomic Neuropathy Type III or Familial Dysautonomia is an autosomal recessive disorder common in Eastern European Jews with an incidence of one in ten thousand to twenty thousand, but is very rare in other ethnic groups. [2, 3] The genetic locus has been mapped to chromosome $9^{q31 - q33}$ (IKBKAP gene). [2, 4] This gene encodes for the IKP protein (IkB kinase complex associated protein). To date, there have been three mutations in IKBKAP identified in individuals with this syndrome. The decreased in the amount of functional IKAP protein in cells causes Familial Dysautonomia. The reported patient had all the features consistent with this syndrome but unfortunately genetic testing was not done as the test is not available.

In this disorder, the development and survival of the autonomic nervous system is affected resulting in abnormalities in the sensory, sympathetic and some parasympathetic neurons. This is characterised pathologically by a reduced number of small unmyelinated nerve fibers that carry pain, temperature and taste sensations and that mediate autonomic functions. [2] Injection of 0.1 ml of 1:1,000 solution of histamine should produce a triple response, but because of the lack of C fibres no flare develops in these patients.

These patients present with diminished or absent pain and temperature perception but normal touch perception. [1] They present with anhydrosis, decreased lacrimation, tongue ulcerations, recurrent GI upset, diminished or absent deep tendon reflexes, poorly controlled temperature, a fluctuating blood pressure (labile blood pressure resulting in episodic hypertension and postural hypotension) and an absent axon reflex. [2-3] Orthopae-

dic manifestations include fractures, Charcot joints, osteomyelitis, scoliosis and ligamentous laxity. [1-3] Traumatic fractures are common and result in nonunion and pseudoarthrosis. [1] Orthopaedic treatment complications are very common and range from infection to wound breakdown to failure of fixation. [1]

The diagnosis of this rare syndrome is mainly clinical but decreased catecholamines and normal nerve conduction velocity is helpful. [1-2] Most of the patients die in childhood, mainly of pulmonary infections but fifty percent of the patients may reach the age of thirty years. [1-3]

In conclusion, this case highlights the orthopaedic manifestations of a rare autosomal recessive syndrome. The patient had a poor operative outcome as the bimalleolar fracture did not show radiological signs of healing even after twelve months of stablisation and the resultant pseudoarthrosis of proximal right forearm.

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# WORLD PARKINSON'S DAY
# 11th APRIL 2012

**The story behind the Parkinson Tulip**

In 1981, a Dutch horticulturalist (J.W.S. Van der Wereld) who had Parkinson's disease named his prized cultivar (red and white tulip) the 'Dr. James Parkinson' tulip, to honour Dr James Parkinson who first described this medical condition and also to commemorate the International Year of the Disabled. The tulip received the Award of Merit from the Royal Horticultural Society in London England, and also received the Trial Garden Award from the Royal General Bulb Growers of Holland. It is described as a flower: 'exterior, glowing cardinal red, small feathered white edge, outer base whitish; inside, currant-red to turkey-red, broad feathered white edge, anthers pale yellow'.

The Red Tulip was launched as the Worldwide Symbol of Parkinson's disease at the 9th World Parkinson's disease Day Conference in Luxembourg on 11th April 2005.