

### What is rhizomelic chondrodysplasia punctata type 1?

Rhizomelic chondrodysplasia punctata type 1 is an inherited disease characterized by skeletal abnormalities, growth retardation, intellectual disabilities, cataracts and decreased life expectancy.<sup>1</sup> The signs and symptoms associated with rhizomelic chondrodysplasia punctata type 1 are attributed to a defect in the body's ability to produce a specific type of fat called plasmalogen which is necessary for normal neurological function and skeletal formation.<sup>2</sup>

### What are the symptoms of rhizomelic chondrodysplasia punctata type 1 and what treatment is available?

Signs of rhizomelic chondrodysplasia punctata type 1 are usually apparent at birth. Rhizomelic chondrodysplasia punctata type 1 is characterized by:<sup>1,3</sup>

- Rhizomelia (shortening of the bones of the upper arms and legs)
- Chondroplasia punctata (x-ray findings of scattered calcifications at the end of the long bones)
- Joint contractures (limited range of joint movement)
- Cataracts (clouding of the eye lenses, impairing vision)
- Profound postnatal growth deficiency
- Severe intellectual disability
- Recurrent respiratory infections and breathing problems
- Ichthyosis (dry, scaly, or thickened skin)
- Seizures
- Distinctive facial features including prominent forehead, depressed nasal bridge and small nose

Although the majority of children with rhizomelic chondrodysplasia punctata type 1 have severe disease, some children have a milder form characterized by cataracts and chondrodysplasia punctata with milder degrees of rhizomelia, growth retardation and intellectual deficits.<sup>1</sup>

There is no cure for rhizomelic chondrodysplasia punctata type 1. Most children do not survive the first decade of life. Management of symptoms is supportive and may include physical therapy to improve joint contractures, orthopedic procedures to improve function, and cataract surgery.<sup>1</sup>

### How is rhizomelic chondrodysplasia punctata type 1 inherited?

Rhizomelic chondrodysplasia punctata type 1 is an autosomal recessive disease caused by mutations in the *PEX7* gene.<sup>2</sup> An individual who inherits one copy of a *PEX7* gene mutation is a carrier and is not expected to have related health problems. An individual who inherits two *PEX7* mutations, one from each parent, is expected to be affected with rhizomelic chondrodysplasia punctata type 1.

If both members of a couple are carriers of a mutation in the same gene, the risk of having an affected child is 25% in each pregnancy; therefore, it is especially important that the reproductive partner of a carrier be offered testing.

### Who is at risk for rhizomelic chondrodysplasia punctata type 1?

Rhizomelic chondrodysplasia punctata type 1 occurs in individuals of all races and ethnicities. The disease is estimated to affect 1 in 100,000 individuals with an approximate carrier frequency of 1 in 158.<sup>3</sup>

Having a relative who is a carrier or who is affected can increase an individual's risk of being a carrier. Consultation with a genetics health professional may be helpful in determining carrier risk and appropriate testing.

**What does a positive test result mean?**

If a gene mutation is identified, an individual should speak to a physician or genetics professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members.

**What does a negative test result mean?**

A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier is also influenced by family history, medical symptoms, and other relevant test results.

**Where can I get more information?**

Genetic Disorders UK: <http://www.geneticdisordersuk.org/gduknetwork/rhizomelicchondrodysplasiapunctata>

Children Living with Inherited Metabolic Diseases: [www.climb.org.uk](http://www.climb.org.uk)

Genetics Home Reference: <http://ghr.nlm.nih.gov/condition/rhizomelic-chondrodysplasia-punctata>

**References**

1. Braverman NE, et al. Rhizomelic chondrodysplasia punctata type 1. Gene Reviews NCBI Bookshelf National Library of Medicine, NIH Available at: <http://www.ncbi.nlm.nih.gov/books/NBK1270>. Accessed February 22, 2012.
2. Genetics Home Reference. *PEX7*. Available at: <http://ghr.nlm.nih.gov/gene/PEX7>. Accessed February 22, 2012.
3. Genetics Home Reference. *Rhizomelic chondrodysplasia punctata*. Available at: <http://ghr.nlm.nih.gov/condition/rhizomelic-chondrodysplasia-punctata>. Accessed March 1, 2012.