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What is Rhizomelic Chondrodysplasia Punctata Type 1?
Rhizomelic chondrodysplasia punctata type 1 (RCDP1) is an inherited disease characterized by skeletal abnormalities, growth retardation, intellectual disabilities, cataracts and decreased life expectancy.[1] The signs and symptoms associated with RCDP1 are attributed to a defect in the body's ability to produce a specific type of fat called plasmalogens which are necessary for normal neurological function and skeletal formation.[2]

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. RCDP1 is characterized by[1,3]

- Rhizomelia (shortening of the bones of the upper arms and legs ie long bones)
- 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 lenses of the eyes impairing vision)
- Profound postnatal growth deficiency
- Severe mental retardation
- Recurrent respiratory infections and breathing problems
- Icthyosis (dry, scaly, or thickened skin)
- Seizures
- Distinctive facial features including prominent forehead, depressed nasal bridge and small nose

Although the majority of children with RCDP1 have severe disease, some children have a milder form characterized by cataracts and chondrodysplasia punctata with milder degrees of rhizomelia, growth retardation and mental deficits.[1]

There is no cure for RCDP1. Unfortunately, most children do not survive the first decade of life. Management of RCDP1 symptoms is supportive and may include physical therapy to improve joint contractures, orthopedic procedures to improve function and cataract surgery.[1]

How is Rhizomelic Chondrodysplasia Punctata Type 1 inherited?
RCDP1 is an autosomal-recessive disease caused by mutations in the PEX7 gene.[2] 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 mutations in this gene, one from each parent, is expected to be affected with RCDP1.

If both members of a couple are carriers, the risk for 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. This disease is estimated to affect 1 in 100,000 individuals with an approximate carrier frequency of 1 in 158. [3]

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.

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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?
- National Organization for Rare Disorders (NORD) www.rarediseases.org
- Children Living with Inherited Metabolic Diseases www.climb.org.uk
- Genetic Alliance www.geneticalliance.org

References

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