



Genetics Uncoded:



Facts about

Rhizomelic Chondrodysplasia Punctata Type 1



What Your Test Results Mean

Carriers typically show no symptoms of rhizomelic chondrodysplasia punctata type 1 (RCDP1); however, carriers are at an increased risk of having a child with RCDP1. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

● Rhizomelic Chondrodysplasia Punctata Type 1 Explained

RCDP1 is an inherited condition that impairs the body's ability to make plasmalogen, an important component of cell membranes. This disorder causes abnormal development of many body parts, leading to skeletal abnormalities, distinctive facial features, severe intellectual disability, cataracts, and respiratory problems. Most children with this condition grow and develop more slowly than other children their age and do not achieve developmental milestones such as sitting without support, feeding themselves, or speaking in phrases. Affected individuals may also be prone to seizures and recurrent respiratory infections. Most people with RCDP1 have a shortened lifespan and do not live into adulthood.

● How the Genetics Work

RCDP1 is an autosomal recessive disorder caused by variants in the *PEX7* gene. In general, individuals have two copies of the *PEX7* gene. Carriers of RCDP1 have a single variant in one copy of the *PEX7* gene while individuals with RCDP1 have variants in both copies of their genes, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

Questions?

Contact us at **1-855-776-9436** to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.