# Rhizomelic Chondrodysplasia Punctata Type 1/Refsum Disease

### What Your Results Mean

Test results indicate that you are a carrier of rhizomelic chondrodysplasia punctata type 1 (RCDP1)/Refsum disease. Carriers typically show no symptoms of RCDP1. Risk for current or future pregnancies is dependent on your partner's carrier status. Carrier testing of your partner is recommended in addition to consultation with a genetic counselor for more detailed risk assessment.

Since this is an inherited gene change, this information may be helpful to share with family members as it may impact their family planning and their own personal clinical management.

### Recommended Next Steps

Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment. If both you and your partner or donor are carriers for RCDP1, each of your children has a 1 in 4 (25%) chance to have the condition.

## Rhizomelic Chondrodysplasia Punctata Type 1/Refsum Disease Explained

### What is Rhizomelic Chondrodysplasia Punctata Type 1/Refsum Disease?

Rhizomelic chondrodysplasia punctata type 1 (RCDP1)/Refsum disease is an inherited condition that impairs the body's ability to make plasmalogen, an important component of cell membranes. It is characterized by skeletal abnormalities, distinctive facial features, intellectual disability, and respiratory problems. There are three types, with type I being the most common. Type I rhizomelic chondrodysplasia punctata is caused by variants in the *PEX7* gene, which is involved in the formation of function of the peroxisomes in the cell. Some of the skeletal features include shortening of the bones in the upper arms and thighs (called rhizomelic shortening), a bone abnormality called chondrodysplasia punctata that affects the growth of the long bones, and joint deformities called contractures which make the joints very stiff and painful. The facial features include a prominent forehead, widely spaced eyes, a sunken appearance to the middle of the face, a small nose with upturned nostrils, and full cheeks. People with this condition can also develop cataracts very early in life. Other features include significant developmental delay, severe intellectual disability, growth delay, seizures, recurrent respiratory infections, and life-threatening breathing problems.

### Prognosis

RCDP1 has a very poor prognosis with death generally occurring during the first decade of life, mainly due to respiratory complications.

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## Treatment

Management is of this condition is usually focused on addressing specific symptoms; there is no cure. Physical therapy is usually recommended to assist in the improvement of contractures; orthopedic surgeries have also improved function in some individuals. Dietary restriction of phytanic acid to avoid the consequences of phytanic acid accumulation over time may be of benefit to some individuals with milder forms of this condition. Other management includes seizure control; vision, hearing, and orthopedic care; prevention of respiratory infections; and placement of a gastrostomy tube to assist with poor feeding and recurrent aspiration.

**Resources**
**Little People of America**
http://www.lpaonline.org/
**National Society of Genetic Counselors**
https://www.nsgc.org/

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