Pycnodysostosis

What Your Results Mean

Test results indicate that you are a carrier of pycnodysostosis. Carriers are not expected to show symptoms. You and your partner would both have to be carriers of pycnodysostosis for there to be an increased chance to have a child with symptoms; this is known as autosomal recessive inheritance. Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a 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.

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 are carriers for pycnodysostosis, each of your children has a 1 in 4 (25%) chance to have the condition.

Pycnodysostosis Explained

What is Pycnodysostosis?

Pycnodysostosis is an inherited lysosomal disorder caused by deficient levels of the lysosomal enzyme cathepsin K. Decreased cathepsin K leads to multiple bones issues in the individual such as short stature and dense and brittle bones causing the individual to easily suffer from fractures. Delayed or abnormal development of the teeth is also seen with this disorder.

Prognosis

Prognosis for pycnodysostosis is generally considered to be good. Lifespan is typically not affected by pycnodysostosis with proper treatment.

Treatment

Treatment for pycnodysostosis involves managing the symptoms along with good dental hygiene. Growth hormones can help to improve the height of the individual, as well as surgery to correct deformities in the face and jaw.

Resources

National Organization for Rare Disorders (NORD)
https://rarediseases.org/rare-diseases/pyknodysostosis/

National Society of Genetic Counselors
https://www.nsgc.org/

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