Dyskeratosis Congenita, Autosomal Recessive

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of dyskeratosis congenita, autosomal recessive. Carriers of dyskeratosis congenita typically do not develop symptoms of the disease; however, some carriers may develop symptoms that appear later in life and carriers do have an increased risk of having a child with the disorder. You and your partner or donor would both have to be carriers of dyskeratosis congenita 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 or donor are carriers for dyskeratosis congenita, each of your children have a 25%, or 1 in 4, chance to have the condition.

Dyskeratosis Congenita, Autosomal Recessive Explained

What is Dyskeratosis Congenita, Autosomal Recessive?

Dyskeratosis congenita, autosomal recessive belongs to a group of inherited conditions known as dyskeratosis congenita that affect several different parts of the body. People with dyskeratosis congenita can have bone marrow failure with recurrent infections, scarred lungs, short stature, developmental delay, dental abnormalities, osteoporosis, liver disease, skin pigment differences, abnormal nails, or thickened, white patches inside the mouth. People with dyskeratosis congenita also have a higher chance of developing certain types of cancer. The severity of symptoms can vary widely, with some people having just a few of these symptoms and others having many.

Prognosis

The prognosis is variable and depends on the severity of symptoms. The condition is associated with shorter life expectancy for more severe cases, but many people live to at least age 60.

Treatment

There is no cure for dyskeratosis congenita. Symptoms are treated as they present with standard treatments. Bone marrow transplantation, if successful, can be curative for bone marrow failure and blood cancers.

Resources

Genetics Home Reference
https://medlineplus.gov/genetics/condition/dyskeratosis-congenita/
National Organization for Rare Disorders
https://rarediseases.org/rare-diseases/dyskeratosis-congenita
National Society of Genetic Counselors
https://www.nsgc.org/

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