Ehlers-Danlos Syndrome, Dermatosparaxis Type

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of Ehlers-Danlos syndrome, dermatosparaxis type. Carriers are not expected to show symptoms. You and your partner or donor would both have to be carriers of Ehlers-Danlos syndrome, dermatosparaxis type 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 Ehlers-Danlos syndrome, dermatosparaxis type, each of your children has a 1 in 4 (25%) chance to have the condition.

Ehlers-Danlos Syndrome, Dermatosparaxis Type Explained

What is Ehlers-Danlos Syndrome, Dermatosparaxis Type?

Ehlers-Danlos syndrome, dermatosparaxis type is an inherited condition that impairs the body's ability to properly make collagen, an important component of connective tissue. Affected individuals have soft, velvety, and fragile skin that stretches and tears easily, in addition to severe joint hyperextensibility. Individuals with Ehlers-Danlos syndrome, dermatosparaxis type bruise and scar very easily and their fragile connective tissue can cause health complications such as spontaneous rupture of the bladder or diaphragm.

Prognosis

The prognosis of Ehlers-Danlos syndrome, dermatosparaxis type is not well-defined. Ehlers-Danlos syndrome, dermatosparaxis type is not known to affect intelligence or mental function.

Treatment

Treatment is symptomatic as there is no cure for Ehlers-Danlos syndrome, dermatosparaxis type. Anti-inflammatory drugs can be beneficial in treating joint pain. Padding and bandages help protect against skin tearing, and physical therapy can be considered to help treat joint hypermobility.

Resources

The Ehlers-Danlos Society
https://www.ehlers-danlos.com/
National Organization for Rare Disorders (NORD)
https://rarediseases.org/rare-diseases/ehlers-danlos-syndrome/
National Society of Genetic Counselors
https://www.nsgc.org/

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