Genetics Uncoded:

NxGen MDx

Facts about Dyskeratosis Congenita

What Your Test Results Mean

Carriers show no symptoms of dyskeratosis congenita and are not at risk to develop symptoms of the disorder. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Dyskeratosis Congenita Explained

Dyskeratosis congenita is an inherited condition caused by defects in the maintenance of telomeres in the cell. The classical signs of this disorder are abnormal skin pigmentation, nail dystrophy, and white patches in the mouth. Most people with dyskeratosis congenita have normal intelligence and motor development, but they are very vulnerable to certain conditions such as bone marrow failure, pulmonary fibrosis, cancer, liver disease, osteoporosis, and blocked tear ducts. The severity of the disorder is variable, but seriously affected individuals die from bone marrow failure, cancer, or pulmonary fibrosis by early adulthood.

How the Genetics Work

Dyskeratosis congenita is an autosomal recessive disorder caused by pathogenic variants in the *NOLA3* gene. In general, individuals have two copies of the *NOLA3* gene. Carriers of dyskeratosis congenita have a single variant in one copy of the *NOLA3* gene while individuals with dyskeratosis congenita 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.

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Revised 12/15/16