NxGen MDx

SLC26A2-Related Disorders

What Your Results Mean

Test results indicate that you are a carrier of an *SLC26A2*-related disorder. Carriers are not expected to show symptoms. You and your partner would both have to be carriers of *SLC26A2*-related disorders 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 *SLC26A2*-related disorders, each of your children has a 1 in 4 (25%) chance to have the condition.

SLC26A2-Related Disorders Explained

What is an *SLC26A2*-Related Disorder?

SLC26A2-related disorders are a group of inherited disorders of cartilage and bone formation. These diseases include: achondrogenesis type 1B, diastrophic dysplasia, and recessive multiple epiphyseal dysplasia.

Achondrogenesis type 1B is a severe skeletal dysplasia characterized by extremely short limbs, a narrow chest, and a prominent, rounded abdomen. Fingers and toes are short, and feet may be rotated inwards. Affected individuals may have a soft outpouching near the belly button (umbilical hernia) or the groin (inguinal hernia).

Diastrophic dysplasia is characterized by short stature, short arms and legs, clubfoot, cleft palate, swelling of the ears, hitchhiker thumbs, progressive abnormal curvature of the spine, joint pain, and joint abnormalities called contractures, which restrict movement. These individuals typically have a normal head size and the condition does not usually affect intelligence.

Multiple epiphyseal dysplasia is characterized by joint pain, early-onset arthritis, malformations of the hands, cleft palate, club foot, and abnormal curvature of the spine. Individuals with this condition typically reach normal height.

Prognosis

Prognosis varies depending on the specific variant in the *SLC26A2* gene. Infants with achondrogenesis type 1B typically die prenatally or soon after birth. The mechanism of prenatal death is unknown, but newborns typically die of respiratory failure. Individuals with diastrophic dysplasia typically live into adulthood and do not have life-threatening breathing problems. These individuals will face physical challenges with walking and other movement and may rely on various mechanical aids for mobility. Individuals with multiple epiphyseal dysplasia have a better prognosis, as they have a normal lifespan and can perform most daily activities. However, they may have chronic joint pain and some bone abnormalities.

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Treatment

Treatment varies depending on the specific change in the *SLC26A2* gene. Treatment for individuals with achondrogenesis type 1B is strictly palliative care. Treatment of diastrophic dysplasia focuses on maintaining joint mobility through physiotherapy and casting. Surgery may be necessary to address scoliosis and clubfoot for affected individuals as well. Physiotherapy may also be used to treat individuals with multiple epiphyseal dysplasia to maintain optimal muscle strength.

Resources

MAGIC Foundation for Children's Growth
https://www.magicfoundation.org/
Little People of America (LPA)
http://www.lpaonline.org/
National Society of Genetic Counselors
https://www.nsgc.org/

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