Genetics Uncoded:

NxGen MDx

Facts about Biotinidase Deficiency

What Your Test Results Mean

Carriers typically do not show any symptoms of biotinidase deficiency. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Biotinidase Deficiency Explained

Biotinidase deficiency is an inherited metabolic disorder that can present with seizures, hypotonia (weak muscle tone), and breathing issues in the newborn period. The *BTD* gene makes the enzyme biotinidase, which aids the body in reusing biotin, a B vitamin that is essential for breaking down fats, proteins, and carbohydrates. Individuals with biotinidase deficiency have decreased or eliminated activity of biotinidase. Without enough of this enzyme, biotin cannot be recycled normally within the body. If not properly treated, developmental delay, vision and hearing loss, ataxia (issues with movement and balance), skin rashes, and alopecia (hair loss) may develop. Treatment involves lifelong management with biotin supplements, which can lessen and prevent many of the complications from biotinidase deficiency.

How the Genetics Work

Biotinidase deficiency is an autosomal recessive disorder caused by variants in the *BTD* gene. In general, individuals have two copies of the *BTD* gene. Carriers of biotinidase deficiency have a variant in one copy of the *BTD* gene while individuals with biotinidase deficiency have variants in both copies of *BTD*, 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.

NxGenMDx, LLC
801 Broadway Ave NW, Suite 203
Grand Rapids, MI, 49504

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