



Facts about

Sulfate Transporter-Related Osteochondrodysplasia



What Your Test Results Mean

Carriers typically show no symptoms of sulfate transporter-related osteochondrodysplasia; however, carriers are at an increased risk of having a child with sulfate transporter-related osteochondrodysplasia.

Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

● Sulfate Transporter-Related Osteochondrodysplasia Explained

Sulfate transporter-related osteochondrodysplasias are a group of inherited disorders of cartilage and bone formation. While caused by variants in the same gene, the four disorders differ greatly in severity. Atelosteogenesis type 2 is the most serious form followed by achondrogenesis type 1B, fatal before or shortly following birth. Diastrophic dysplasia is a more mild form characterized by short stature (very short extremities), protruding abdomens, and narrow chests. Affected individuals live to adulthood and typically have normal intelligence and mental function. Recessive multiple epiphyseal dysplasia is the mildest form of sulfate transporter-related osteochondrodysplasia characterized by joint pain, hand/foot deformities, and scoliosis. Treatment for the more severe forms is palliative while treatment for the milder forms is symptomatic. Lifespan varies greatly depending on the severity of the osteochondrodysplasia.

● How the Genetics Work

Sulfate transporter-related osteochondrodysplasia is an autosomal recessive disorder caused by variants in the *SLC26A2* gene. In general, individuals have two copies of the *SLC26A2* gene. Carriers of sulfate transporter-related osteochondrodysplasia have a single variant in one copy of the *SLC26A2* gene while individuals with sulfate transporter-related osteochondrodysplasia 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 **1-866-661-7966** to set up an appointment to discuss your results in more detail with a Clarity Genetics genetic counselor.