

### **What are the Sulfate Transporter-related Osteochondrodysplasias?**

The sulfate transporter-related osteochondrodysplasias (STROs) are a group of inherited diseases characterized by short stature and short limbs, spine and joint abnormalities, and early-onset osteoarthritis.<sup>1</sup> The STROs involves abnormalities in a protein that transports sulfate into cells where it is needed for the development of cartilage. The symptoms of STRO are due to the abnormal formation of cartilage and its conversion to bone.<sup>2</sup> The four types of STRO are known as recessive multiple epiphyseal dysplasia, diastrophic dysplasia, atelosteogenesis type 2 (also known as de la Chapelle dysplasia or McAlister dysplasia), and achondrogenesis type 1B.<sup>1,3</sup>

### **What are the symptoms of Sulfate Transporter-related Osteochondrodysplasias and what treatment is available?**

Signs and symptoms of the sulfate transporter-related osteochondrodysplasias vary in severity, ranging from mild bone abnormalities with joint pain to lethal conditions.<sup>4</sup> The subtypes are classified based on clinical and radiological (x-ray) findings.

The mildest form is recessive multiple epiphyseal dysplasia (rMED). The majority of individuals are diagnosed in childhood, but some may not be diagnosed until adulthood. Symptoms are variable and may include<sup>4</sup>:

- Mild short stature
- Abnormalities of hands and fingers
- Abnormality of the kneecaps
- Clubfoot (foot turned inward and upward)
- Scoliosis (abnormal curvature of spine)
- Cleft palate (abnormal opening in the roof of the mouth)
- Joint pain, usually beginning in late childhood
- Early-onset arthritis

Symptoms of diastrophic dysplasia (DTD) are often recognized at birth or during infancy. Some infants have breathing difficulties due to a small rib cage and may require mechanical ventilation. Individuals who survive past infancy typically have a normal life expectancy. Symptoms of DTD may include<sup>5</sup>:

- Short stature
- Shortened limbs
- Clubfoot
- Lordosis, kyphosis, and scoliosis (abnormal curvatures of the spine)
- Cleft palate (abnormal opening in the roof of the mouth)
- Early-onset joint pain and joint contractures (tightening of the muscles or tendons preventing full extension)
- Early-onset arthritis

The skeletal findings of atelosteogenesis type 2 (AO2) resemble those of DTD but are more severe. Additional symptoms include a prominent abdomen and distinctive facial features. Affected infants have severe breathing difficulties and do not survive past early infancy.<sup>6</sup>



Achondrogenesis type 1B (ACG1B) is the most severe form of STRO with significant skeletal abnormalities. Some babies with ACG1B may have hydrops fetalis (abnormal build-up of fluid in the body) detected by prenatal ultrasound and may not survive pregnancy. Affected infants have very short limbs, fingers, and toes, a small chest, a prominent abdomen, and die shortly after birth.<sup>7</sup>

There is no cure for the sulfate transporter-related osteochondrodysplasias, and treatment includes supportive care for symptoms. For individuals with rMED and DTD, orthopedic intervention and pain management may be necessary.<sup>4,5</sup>

### **How are the Sulfate Transporter-related Osteochondrodysplasias inherited?**

The sulfate transporter-related osteochondrodysplasias are autosomal recessive diseases caused by mutations in the *SLC26A2* gene.<sup>2</sup> An individual who inherits one *SLC26A2* gene mutation is a “carrier” and is not expected to have related health problems. An individual who inherits two *SLC26A2* gene mutations, one from each parent, is expected to be affected with one of the sulfate transporter-related osteochondrodysplasias.

If both members of a couple are carriers, the risk of having an affected child is 25% in each pregnancy; therefore, it is especially important that the reproductive partner of a carrier be offered testing.

### **Who is at risk for the Sulfate Transporter-related Osteochondrodysplasias?**

The sulfate transporter-related osteochondrodysplasias can occur in individuals of all races and ethnicities, but it is most common in individuals of Finnish ancestry, with a carrier frequency estimated to be 1/50.<sup>8</sup> In the general population, the sulfate transporter-related osteochondrodysplasias are estimated to affect 1 in 100,000 individuals, with a calculated carrier frequency of 1/158.<sup>5</sup>

Having a relative who is a carrier or who is affected can increase an individual’s risk of being a carrier. Consultation with a genetics health professional may be helpful in determining carrier risk and appropriate testing.

### **What does a positive test result mean?**

If a gene mutation is identified, an individual should speak to a physician or genetics health professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members.

### **What does a negative test result mean?**

A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier is also influenced by family history, medical symptoms, and other relevant test results.

### **Where can I get more information?**

- Little People of America: [www.lpaonline.org](http://www.lpaonline.org)



## References

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