

Patient Guide to Diastrophic Dysplasia

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What is Diastrophic Dysplasia?

Diastrophic Dysplasia (DD) is a condition from birth where cartilage and bone do not form properly. This leads to abnormal skeletal growth and joint problems. The severity and features that develop can differ a lot from person to person. Children with the condition have characteristic growth problems and require special care.

What causes Diastrophic Dysplasia?

DD is a genetic condition. This means that a gene that directs growth in the body does not work properly. It is present from birth. The condition is genetically passed down as an autosomal recessive trait. This means that each parent has one copy of the DD gene and that the child collected genes from each parent by random chance.

The DD gene codes for a sulfate transporter protein that is named the "diastrophic dysplasia sulfate transporter." This protein helps cartilage keep its natural properties. Cartilage is important because it is found at the end of bones in joints and is also a material that bone forms from.

How common is Diastrophic Dysplasia?

DD is extremely rare. DD seems to be the most common in Finland where over 160 people have the condition. The original mutation may have occurred there.

How do you know your child has Diastrophic Dysplasia?

DD may initially be brought up during an ultrasound examination in the second trimester of pregnancy. The ultrasound may reveal skeletal features that suggest the condition. This may include very short bones. These bones are often the upper arm or thigh bone. It may also reveal clubfeet and a characteristic thumb condition called abducted thumbs meaning that the thumb is away from the palm in a "hitchhiking" position.

At birth the child may also have features of DD and as he or she grows older more features may become evident. Some of these characteristic features are explained below.

What are the signs and symptoms of Diastrophic Dysplasia?

Children with DD have unique facial characteristics. Half of DD children are born with a cleft palate. This is an important problem because children can often have problems breathing and swallowing correctly. This can cause a lung infection like pneumonia. The cartilage in the throat can also be very soft and cause breathing difficulties for the infant. Children with DD may also have flattened nose bridges, full cheeks, and a characteristic ear appearance that typically occurs at 3 to 6 weeks of age. The cartilage in the ears hardens into a unique shape termed "cauliflower ears."

As the child grows, the skeleton also develops abnormally. They often have short arms, legs and trunk. There are also problems with the spine, arms and hands, hips, knees, feet, and overall height.

How will my child do in the long run with Diastrophic Dysplasia?

Overall, people with DD in the long run do well. They have become attorneys, accountants and succeed in other types of professions. Most people with DD live just as long as people without DD. The life expectancy is not significantly reduced. People with DD can live productive and active lives.

What kind of problems do children with Diastrophic Dysplasia have?

Children usually learn to sit, crawl and walk at later dates than average. A small number of children with DD develop lung and breathing problems because of the cleft palate and soft cartilage in the throat. Children with DD also often have spine problems. The spine may not completely grow together or have abnormal curvatures. Sometimes the back part of the bones of the spine

called vertebra do not grow together to fully protect the spinal cord. Other times, abnormal curves called kyphosis and scoliosis may develop endangering the spinal cord. Kyphosis is a front-to-back curve while scoliosis is a side-to-side curve. Watching these problems is important because the spinal cord helps control breathing, bladder and bowel function as well as provide hand and feet control.

People with DD also develop hip problems. The hips do not grow properly and they may have hips that move out of place, become stiff and lose mobility, develop early arthritis. The knees can also develop abnormally becoming knock-kneed and develop early arthritis.

The arms and legs are often short. The shortest bones tend to be the upper arm bone and the thigh bone. The hands are also usually short and broad. The thumb is also often kept in a characteristic "hitchhiker" position. Clubfeet also often develops. This is when the feet often points down and in.

Height is also often affected to varying amounts. Children with more severe DD tend to be shorter. On average, the male adult DD patient grows to a height of 136cm or 4 feet 6 inches. Adult female patients often grow to a height of 129 cm or 4 feet 3 inches.

Should my child see a doctor regularly?

Yes. It is important to have a doctor follow your child to make sure spine problems do not become life-threatening and to correct any other skeletal problems that may hinder the child.

The physician can monitor your child's spine. The orthopedist will perform regular physical examinations testing the spine, perform X-rays to look at the spine bones, and order other tests if necessary. If a problem is detected, the pediatric orthopedic surgeon may recommend a brace or surgery to stabilize the spine.

The hips, knees and feet are also monitored with regular visits. Physical therapy, bracing, and surgeries may be performed to keep your child's mobility and health.

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