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Orthopaedic Surgery

Pediatric Orthopaedics

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Patient Guide to Spondyloepiphyseal Dysplasia Congenita

Paul Sponseller, M.D.
Michael Ain, M.D.
Nancy Miller, M.D.
Michael Bahk, MSIV

What is Spondyloepiphyseal Dysplasia Congenita?
Spondyloepiphyseal Dysplasia Congenita (SEDC) is a rare genetic disorder where bone and joints do not form properly. 'Spondylo' refers to spine, 'epiphyseal' refers to the growing ends of bones, 'dysplasia' means abnormal growth, 'congenita' means congenital or present from birth. Therefore, this is a condition from birth where the spine and ends of bones do not grow properly. People with SEDC are shorter and often have spine, hip, and feet problems. Children with the condition have unique problems and require special care.

SED often comes in two forms. The more severe form is SED Congenita (SEDC) while the milder form is SED Tarda (SEDT).

What causes Spondyloepiphyseal Dysplasia Congenita?
SEDC is a genetic condition. This means that a gene that directs growth in the body does not work properly. In this particular condition, the gene affects a protein called type II collagen. This is the main protein used in cartilage. Cartilage is important because it covers the bones used in joints. Bone also

develops from cartilage meaning that many of the bones in people with SEDC grow incorrectly. The severity of the disease varies a lot from person to person.

Most of the time the abnormal gene forms spontaneously or is a random mutation. Sometimes, the gene can be inherited from a parent as an autosomal dominant mutation. This means that collecting only one copy of the abnormal gene from either parent results in a child with SEDC.

How common in Spondyloepiphyseal Dysplasia Congenita?
SEDC is a very rare condition. It is estimated to occur in about 3-4 people out of a million.

How do you know your child has Spondyloepiphyseal Dysplasia Congenita?
The severity of SEDC varies a lot from person to person. In general, people with SEDC often have a characteristic appearance. The face often looks tight and is described as taut. People with SEDC often also have small mouths and may be born with cleft palates. The trunk, arms and legs are shortened. The chest may also protrude somewhat and form a characteristic shape called pectus carinatum.

How will my child do in the long run with Spondyloepiphyseal Dysplasia Congenita?
Overall, people with SEDC in the long run do well. Most people with SEDC live just as long as people without SEDC and have productive and active lives. They can obtain advanced education. There are several people with SEDC who are doctors, lawyers, and scientists. People with SEDC are able to have families and contribute their talents to society.

What are some of the problems that people with Spondyloepiphyseal Dysplasia Congenita develop?
People with SEDC often develop spine problems. The most serious spine problem is neck instability. Sometimes one of the neck bones called the axis does not grow properly. This allows for the neck to be unstable and endanger the spinal cord. The spinal cord can be compressed or injured. The spinal cord is important because it helps control breathing, bowel and bladder function as well as control movement of the hands and feet. Other spine problems called scoliosis and kyphosis can also develop. Both are abnormal curvatures where the spine bends incorrectly. Scoliosis is an abnormal curvature from side-to-side while kyphosis is an extreme bending forward in the front-to-back plane. Both scoliosis and kyphosis develop before the teen years.

People with SEDC also develop hip problems called coxa vara. This means that the hips develop abnormally so that the legs

point inward toward the midline of the body too much. 'Coxa' refers to the hips and 'vara' refers to the inward direction the legs point. The more severe the disease the more the hips develop in varus. Sometimes, this causes the child to walk abnormally with the head held back.

The most common foot problem is equinovarus where the foot is held pointing down 'equino' and the bottom of the foot points in or toward the midline or 'varus.'

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 will ask questions, physically exam the child and order X-rays or other tests if necessary to follow the progress of your child.

People with SEDC can have eye problems such as retinal detachment. Some may also develop hearing problems. It is important for physicians to monitor these concerns and keep your child as healthy as possible.

What is the treatment for Spondyloepiphyseal Dysplasia Congenita?
There are various forms of treatment than can help correct specific skeletal abnormalities. The most serious problem that should be watched and treated because it is life-threatening is neck instability. If the neck is unstable and the spinal cord is endangered, surgery is performed to stabilize the neck. This involves fusion of the upper neck bones. The spine is fixed in place and the neck bones are allowed to grow together to prevent unwanted motion.

Scoliosis and kyphosis are monitored over time and may initially be treated with braces. If the curve is too severe or does not correct with bracing, surgical fusion is also recommended. Surgery will straighten the spine and allow the spine bones to grow together again to keep the spine straight for the long-term.

Hip surgeries called osteotomies can be performed to correct the abnormal angles and positions of the hip bones. Cuts in the bones are made and the bones repositioned in normal alignment so that patients can walk more normally.

The foot problems can be treated with physical therapy, casting, and surgery if required.

For more information please contact:
Michael C Ain, M.D.

Assistant Professor
Pediatric Orthopaedics
Specializing in Skeletal Dysplasias
601 North Carolina Street
Baltimore, MD 21287-0882
Phone: (410) 955-3135
Fax: (410) 502-6816

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