

Spondylothoracic dysostosis

Description

Spondylothoracic dysostosis is a condition characterized by malformation of the bones of the spine and ribs. The bones of the spine (vertebrae) do not develop properly, which causes them to be misshapen and abnormally joined together (fused). The ribs are also fused at the part nearest the spine (posteriorly), which gives the rib cage its characteristic fan-like or "crab" appearance in x-rays. Affected individuals have short, rigid necks and short torsos because of the bone malformations. As a result, people with spondylothoracic dysostosis have short bodies but normal-length arms and legs, called short-trunk dwarfism.

The spine and rib abnormalities, which are present from birth, cause other signs and symptoms of spondylothoracic dysostosis. Infants with this condition have small chests that cannot expand adequately, often leading to life-threatening breathing problems. As the lungs expand in the narrow chest, the muscle that separates the abdomen from the chest cavity (the diaphragm) is forced down and the abdomen is pushed out. The increased pressure in the abdomen can cause a soft out-pouching around the lower abdomen (inguinal hernia) or belly-button (umbilical hernia).

Breathing problems can be fatal early in life; however, some affected individuals live into adulthood.

Spondylothoracic dysostosis is sometimes called spondylocostal dysostosis, a similar condition with abnormalities of the spine and ribs. The two conditions have been grouped in the past, and both are sometimes referred to as Jarcho-Levin syndrome; however, they are now considered distinct conditions.

Frequency

Spondylothoracic dysostosis affects about one in 200,000 people worldwide. However, it is much more common in people of Puerto Rican ancestry, affecting approximately one in 12,000 people.

Causes

The *MESP2* gene provides instructions for a protein that plays a critical role in the development of vertebrae. Specifically, it is involved in separating vertebrae and ribs from one another during early development, a process called somite segmentation.

Mutations in the *MESP2* gene prevent the production of any protein or lead to the production of an abnormally short, nonfunctional protein. When the MESP2 protein is nonfunctional or absent, somite segmentation does not occur properly, which results in malformation and fusion of the bones of the spine and ribs seen in spondylothoracic dysostosis.

[Learn more about the gene associated with Spondylothoracic dysostosis](#)

- MESP2

Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- Jarcho-Levin syndrome
- STD

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Spondylocostal dysostosis (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0265343/>)

Genetic and Rare Diseases Information Center

- Autosomal recessive spondylocostal dysostosis (<https://rarediseases.info.nih.gov/diseases/6798/index>)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (<https://rarediseases.org/>)

Catalog of Genes and Diseases from OMIM

- SPONDYLOCOSTAL DYSOSTOSIS 1, AUTOSOMAL RECESSIVE; SCDO1 (<https://omim.org/entry/277300>)
- SPONDYLOCOSTAL DYSOSTOSIS 2, AUTOSOMAL RECESSIVE; SCDO2 (<https://omim.org/entry/608681>)

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28spondylothoracic+dysostosis%5BALL%5D%29+OR+%28Jarcho-Levin+Syndrome%5BTIAB%5D%29+AND+engli sh%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>)

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Last updated June 1, 2016