Spondylocarpotarsal synostosis syndrome

Spondylocarpotarsal synostosis syndrome is a disorder that affects the development of bones throughout the body. Newborns with this disorder are of approximately normal length, but impaired growth of the torso results in short stature over time. The bones of the spine (vertebrae) are misshapen and abnormally joined together (fused). The vertebral abnormalities may result in an abnormally curved lower back (lordosis) and a spine that curves to the side (scoliosis).

People with spondylocarpotarsal synostosis syndrome have abnormalities and fusion of the bones of the wrist (carpal bones) and ankle (tarsal bones). They may also have inward- and upward-turning feet (clubfeet). Characteristic facial features include a round face, a large forehead (frontal bossing), and nostrils that open to the front rather than downward (anteverted nares).

Some people with spondylocarpotarsal synostosis syndrome have an opening in the roof of the mouth (a cleft palate), hearing loss, thin tooth enamel, flat feet, or an unusually large range of joint movement (hypermobility). Individuals with this disorder can survive into adulthood. Intelligence is generally unaffected, although mild developmental delay has been reported in some affected individuals.

Frequency

Spondylocarpotarsal synostosis syndrome is a rare disorder; its prevalence is unknown. At least 25 affected individuals have been identified.

Genetic Changes

Mutations in the FLNB gene cause spondylocarpotarsal synostosis syndrome. The FLNB gene provides instructions for making a protein called filamin B. This protein helps build the network of protein filaments (cytoskeleton) that gives structure to cells and allows them to change shape and move. Filamin B attaches (binds) to another protein called actin and helps the actin to form the branching network of filaments that makes up the cytoskeleton. It also links actin to many other proteins to perform various functions within the cell, including the cell signaling that helps determine how the cytoskeleton will change as tissues grow and take shape during development.

Filamin B is especially important in the development of the skeleton before birth. It is active (expressed) in the cell membranes of cartilage-forming cells (chondrocytes). Cartilage is a tough, flexible tissue that makes up much of the skeleton during early development. Most cartilage is later converted to bone (a process called ossification), except for the cartilage that continues to cover and protect the ends of bones and is present in the nose, airways (trachea and bronchi), and external ears.
B appears to be important for normal cell growth and division (proliferation) and maturation (differentiation) of chondrocytes and for the ossification of cartilage. 

*FLNB* gene mutations that cause spondylocarpotarsal synostosis syndrome result in the production of an abnormally short filamin B protein that is unstable and breaks down rapidly. Loss of the filamin B protein appears to result in out-of-place (ectopic) ossification, resulting in fusion of the bones in the spine, wrists, and ankles and other signs and symptoms of spondylocarpotarsal synostosis syndrome.

A few individuals who have been diagnosed with spondylocarpotarsal synostosis syndrome do not have mutations in the *FLNB* gene. Researchers are working to identify and confirm additional genetic changes that can cause this disorder.

**Inheritance Pattern**

Spondylocarpotarsal synostosis syndrome caused by *FLNB* gene mutations 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.

In a few individuals with signs and symptoms similar to those of spondylocarpotarsal synostosis syndrome but without *FLNB* gene mutations, the condition appears to have been inherited in an autosomal dominant pattern. Autosomal dominant means one copy of the altered gene in each cell is sufficient to cause the disorder.

**Other Names for This Condition**

- congenital scoliosis with unilateral unsegmented bar
- congenital synspondylism
- SCT
- SCT syndrome
- spondylocarpotarsal syndrome
- vertebral fusion with carpal coalition

**Diagnosis & Management**

**Genetic Testing**

- Genetic Testing Registry: Spondylocarpotarsal synostosis syndrome

**Other Diagnosis and Management Resources**

- GeneReview: FLNB-Related Disorders
  https://www.ncbi.nlm.nih.gov/books/NBK2534
General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html

- Drug Therapy
  https://medlineplus.gov/drugtherapy.html

- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html

- Palliative Care
  https://medlineplus.gov/palliativecare.html

- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources

MedlinePlus

- Health Topic: Bone Diseases
  https://medlineplus.gov/bonediseases.html

- Health Topic: Scoliosis
  https://medlineplus.gov/scoliosis.html

Genetic and Rare Diseases Information Center

- Spondyloptotic synostosis syndrome

Educational Resources

- Disease InfoSearch: Spondyloptotic synostosis syndrome
  http://www.diseaseinfosearch.org/Spondyloptotic+synostosis+syndrome/6801

- Disease InfoSearch: Spondyloptotic synostosis syndrome
  http://www.diseaseinfosearch.org/Spondyloptotic+synostosis+syndrome/6801

- MalaCards: spondyloptotic synostosis syndrome
  http://www.malacards.org/card/spondyloptotic_synostosis_syndrome

- Orphanet: Spondyloptotic synostosis
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=3275
Patient Support and Advocacy Resources

- Little People of America
  http://www.lpaonline.org/
- Little People UK
  http://littlepeopleuk.org/
- The MAGIC Foundation for Children's Growth
  https://www.magicfoundation.org/

GeneReviews

- FLNB-Related Disorders
  https://www.ncbi.nlm.nih.gov/books/NBK2534

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28spondylocarpotarsal+synostosis+syndrome%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh %5D

OMIM

- SPONDYLOCARPOTARSAL SYNOSTOSIS SYNDROME
  http://omim.org/entry/272460

MedGen

- Spondylocarpotarsal synostosis syndrome

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/27381093
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5117933/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18470895

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/14991055
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19505475

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/28145000

Reprinted from Genetics Home Reference:

Reviewed: April 2018
Published: July 24, 2018

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services