Roberts syndrome

Description

Roberts syndrome is a genetic disorder characterized by limb and facial abnormalities. Affected individuals also grow slowly before and after birth. Mild to severe intellectual impairment occurs in about half of all people with Roberts syndrome.

Children with Roberts syndrome are born with abnormalities of all four limbs. They have shortened arm and leg bones (hypomelia), particularly the bones in their forearms and lower legs. In severe cases, the limbs may be so short that the hands and feet are located very close to the body (phocomelia). People with Roberts syndrome may also have abnormal or missing fingers and toes, and joint deformities (contractures) commonly occur at the elbows and knees. The limb abnormalities are very similar on the right and left sides of the body, but arms are usually more severely affected than legs.

Individuals with Roberts syndrome typically have numerous facial abnormalities, including an opening in the lip (a cleft lip) with or without an opening in the roof of the mouth (cleft palate), a small chin (micrognathia), ear abnormalities, wide-set eyes (hypertelorism), outer corners of the eyes that point downward (down-slanting palpebral fissures), small nostrils, and a beaked nose. They may have a small head size (microcephaly) or clouding of the clear front covering of the eyes (corneal opacities). In severe cases affected individuals have a sac-like protrusion of the brain (encephalocele) at the front of their head. In addition, people with Roberts syndrome may have heart, kidney, and genital abnormalities.

Infants with a severe form of Roberts syndrome are often stillborn or die shortly after birth. Mildly affected individuals may live into adulthood. A condition called SC phocomelia syndrome was originally thought to be distinct from Roberts syndrome; however, it is now considered to be a mild variant. "SC" represents the first letters of the surnames of the two families first diagnosed with this disorder.

Frequency

Roberts syndrome is a rare disorder. Its prevalence is unknown.

Causes

Mutations in the *ESCO2* gene cause Roberts syndrome. This gene provides instructions for making a protein that is important for proper chromosome separation during cell division. Before cells divide, they must copy all of their chromosomes. The copied DNA from each chromosome is arranged into two identical structures, called sister chromatids. The ESCO2 protein plays an important role in establishing the glue that holds the sister chromatids together until the chromosomes are ready to separate.

All identified mutations in the *ESCO2* gene prevent the cell from producing any functional ESCO2 protein, which causes some of the glue between sister chromatids to be missing around the chromosome's constriction point (centromere). In Roberts syndrome, cells respond to abnormal sister chromatid attachment by delaying cell division. Delayed cell division can be a signal that the cell should undergo self-destruction. The signs and symptoms of Roberts syndrome may result from the loss of cells from various tissues during early development. Because both mildly and severely affected individuals lack any functional ESCO2 protein, the underlying cause of the variation in disease severity remains unknown. Researchers suspect that other genetic and environmental factors may be involved.

Learn more about the gene associated with Roberts syndrome

- ESCO2

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

- Appelt-Gerken-Lenz syndrome
- Hypomelia hypotrichosis facial hemangioma syndrome
- Pseudothalidomide syndrome
- RBS
- Roberts-SC phocomelia syndrome
- SC phocomelia syndrome
- SC pseudothalidomide syndrome
- SC syndrome
- Tetraphocomelia-cleft palate syndrome

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Roberts-SC phocomelia syndrome (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0392475/)

Genetic and Rare Diseases Information Center

- Roberts syndrome (https://rarediseases.info.nih.gov/diseases/7387/index)

Patient Support and Advocacy Resources

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

Catalog of Genes and Diseases from OMIM

- ROBERTS-SC PHOCOMELIA SYNDROME; RBS (https://omim.org/entry/268300)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28roberts+syndrome%5BTIAB%5D%29+OR+%28hypomelia+hypotrichosis+facial+hemangioma+syndrome%5BTIAB%5D%29+OR+%28pseudothalidomide+syndrome%5BTIAB%5D%29+OR+%28roberts-sc+phocomelia+syndrome%5BTIAB%5D%29+OR+%28sc+phocomelia+syndrome%5BTIAB%5D%29+OR+%28sc+syndrome%5BTIAB%5D%29+OR+%28tetraphocomelia-cleft+palate+syndrome%5BTIAB%5D%29%29+NOT+%28Norman-Roberts+syndrome%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

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