

Poland syndrome

Description

Poland syndrome is a disorder in which affected individuals are born with missing or underdeveloped muscles on one side of the body, resulting in abnormalities that can affect the chest, shoulder, arm, and hand. The extent and severity of the abnormalities vary among affected individuals.

People with Poland syndrome are typically missing part of one of the major chest muscles, called the pectoralis major. In most affected individuals, the missing part is the large section of the muscle that normally runs from the upper arm to the breastbone (sternum). The abnormal pectoralis major muscle may cause the chest to appear concave. In some cases, additional muscles on the affected side of the torso, including muscles in the chest wall, side, and shoulder, may be missing or underdeveloped. There may also be rib cage abnormalities, such as shortened ribs, and the ribs may be noticeable due to less fat under the skin (subcutaneous fat). Breast and nipple abnormalities may also occur, and underarm (axillary) hair is sometimes sparse or abnormally placed. In most cases, the abnormalities in the chest area do not cause health problems or affect movement.

Many people with Poland syndrome have hand abnormalities on the affected side, commonly including an underdeveloped hand with abnormally short fingers (brachydactyly); small, underdeveloped (vestigial) fingers; and some fingers that are fused together (syndactyly). This combination of hand abnormalities is called symbrachydactyly. Some affected individuals have only one or two of these features, or have a mild hand abnormality that is hardly noticeable; more severe abnormalities can cause problems with use of the hand. The bones of the forearm (radius and ulna) are shortened in some people with Poland syndrome, but this shortening may also be difficult to detect unless measured.

Mild cases of Poland syndrome without hand involvement may not be evident until puberty, when the differences (asymmetry) between the two sides of the chest become more apparent. By contrast, severely affected individuals have abnormalities of the chest, hand, or both that are apparent at birth. In rare cases, severely affected individuals have abnormalities of internal organs such as a lung or a kidney, or the heart is abnormally located in the right side of the chest (dextrocardia).

Rarely, chest and hand abnormalities resembling those of Poland syndrome occur on both sides of the body, but researchers disagree as to whether this condition is a variant

of Poland syndrome or a different disorder.

Frequency

Poland syndrome has been estimated to occur in 1 in 20,000 newborns. For unknown reasons, this disorder occurs more than twice as often in males than in females. Poland syndrome may be underdiagnosed because mild cases without hand involvement may never come to medical attention.

Causes

The cause of Poland syndrome is unknown. Researchers have suggested that it may result from a disruption of blood flow during development before birth. This disruption is thought to occur at about the sixth week of embryonic development and affect blood vessels that will become the subclavian and vertebral arteries on each side of the body. The arteries normally supply blood to embryonic tissues that give rise to the chest wall and hand on their respective sides. Variations in the site and extent of the disruption may explain the range of signs and symptoms that occur in Poland syndrome. Abnormality of an embryonic structure called the apical ectodermal ridge, which helps direct early limb development, may also be involved in this disorder.

Rare cases of Poland syndrome are thought to be caused by a genetic change that can be passed down in families, but no related genes have been identified.

Inheritance

Most cases of Poland syndrome are sporadic, which means they are not inherited and occur in people with no history of the disorder in their families. Rarely, this condition is passed through generations in families. In these families the condition appears to be inherited in an autosomal dominant pattern, which means one copy of an altered gene in each cell is sufficient to cause the disorder, although no associated genes have been found.

Other Names for This Condition

- Poland anomaly
- Poland sequence
- Poland syndactyly
- Poland's anomaly
- Poland's syndrome
- Unilateral defect of pectoralis major and syndactyly of the hand

Additional Information & Resources

[Genetic and Rare Diseases Information Center](#)

- Poland syndrome (<https://rarediseases.info.nih.gov/diseases/7412/index>)

Patient Support and Advocacy Resources

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

Clinical Trials

- ClinicalTrials.gov (<https://clinicaltrials.gov/search?cond=%22Poland syndrome%22>)

Catalog of Genes and Diseases from OMIM

- POLAND SYNDROME (<https://omim.org/entry/173800>)

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28Poland+Syndrome%5BMAJR%5D%29+AND+%28Poland+syndrome%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>)

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