

Primary Spontaneous Pneumothorax

Subjects: Genetics

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Definition

Primary spontaneous pneumothorax is an abnormal accumulation of air in the space between the lungs and the chest cavity (called the pleural space) that can result in the partial or complete collapse of a lung. This type of pneumothorax is described as primary because it occurs in the absence of lung disease such as emphysema. Spontaneous means the pneumothorax was not caused by an injury such as a rib fracture. Primary spontaneous pneumothorax is likely due to the formation of small sacs of air (blebs) in lung tissue that rupture, causing air to leak into the pleural space. Air in the pleural space creates pressure on the lung and can lead to its collapse. A person with this condition may feel chest pain on the side of the collapsed lung and shortness of breath.

1. Introduction

Blebs may be present on an individual's lung (or lungs) for a long time before they rupture. Many things can cause a bleb to rupture, such as changes in air pressure or a very sudden deep breath. Often, people who experience a primary spontaneous pneumothorax have no prior sign of illness; the blebs themselves typically do not cause any symptoms and are visible only on medical imaging. Affected individuals may have one bleb to more than thirty blebs. Once a bleb ruptures and causes a pneumothorax, there is an estimated 13 to 60 percent chance that the condition will recur.

2. Frequency

Primary spontaneous pneumothorax is more common in men than in women. This condition occurs in 7.4 to 18 per 100,000 men each year and 1.2 to 6 per 100,000 women each year.

3. Causes

Mutations in the *FLCN* gene can cause primary spontaneous pneumothorax, although these mutations appear to be a very rare cause of this condition. The *FLCN* gene provides instructions for making a protein called folliculin. In the lungs, folliculin is found in the connective tissue cells that allow the lungs to contract and expand when breathing. Folliculin is also produced in cells that line the small air sacs (alveoli). Researchers have not determined the protein's function, but they believe it may help control the growth and division of cells. Folliculin may play a role in repairing and re-forming lung tissue following damage. Researchers have not determined how *FLCN* gene mutations lead to the formation of blebs and increase the risk of primary spontaneous pneumothorax. One theory is that the altered folliculin protein may trigger inflammation within the lung tissue that could alter and damage the tissue, causing blebs.

Primary spontaneous pneumothorax most often occurs in people without an identified gene mutation. The cause of the condition in these individuals is often unknown. Tall young men are at increased risk of developing primary spontaneous pneumothorax; researchers suggest that rapid growth of the chest during growth spurts may increase the likelihood of forming blebs. Long-term smoking also greatly increases the risk of developing primary spontaneous pneumothorax in both men and women.

3.1. The gene associated with Primary spontaneous pneumothorax

- *FLCN*

4. Inheritance

When this condition is caused by mutations in the *FLCN* gene, it is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. In most

cases, a person inherits the *FLCN* gene mutation from an affected parent. People who have an *FLCN* gene mutation associated with primary spontaneous pneumothorax all appear to develop blebs, but it is estimated that only 40 percent of those individuals go on to have a primary spontaneous pneumothorax.

5. Other Names for This Condition

- pneumothorax
- PSP
- spontaneous pneumothorax

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Keywords

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