

## Isolated congenital asplenia

### Description

Isolated congenital asplenia is a condition in which affected individuals are missing their spleen (asplenia) but have no other developmental abnormalities. While most individuals with this condition have no spleen at all, some people have a very small, nonfunctional spleen (hyposplenism).

The spleen plays an important role in the immune system. This organ is part of the lymphatic system, which produces and transports fluids and immune cells throughout the body. The spleen produces certain immune system cells called phagocytes that help remove bacteria from the blood in order to prevent infections. The spleen also stores particular blood cells that fight foreign invaders until they are needed and filters old blood cells for removal. Because people with isolated congenital asplenia lack these immune functions, they are highly susceptible to bacterial infections.

People with isolated congenital asplenia are prone to developing severe, recurrent infections. Infections most commonly affect the whole body (sepsis), the membrane covering the brain and spinal cord (meningitis), or the ears (otitis media). Infections are most often caused by the *Streptococcus pneumoniae* bacteria.

Without preventative care and proper treatment, the frequent infections caused by isolated congenital asplenia can be life-threatening.

### Frequency

The worldwide prevalence of isolated congenital asplenia is unknown. One population study done in France estimated that the condition occurs in 1 per 2 million newborns.

### Causes

About 40 percent of cases of isolated congenital asplenia are caused by mutations in a gene called *RPSA*. This gene provides instructions for making a protein called ribosomal protein SA, which is a component of ribosomes. Ribosomes are cellular structures that process the cell's genetic instructions to create proteins.

Each ribosome has two parts (subunits) called the large and small subunits. Ribosomal protein SA is one of several proteins that make up the small subunit. Within the ribosome, the function of the ribosomal protein SA is unclear. Research suggests that it

helps the ribosome control the production of certain proteins, many of which are likely important for development before birth.

*RPSA* gene mutations are thought to reduce the amount of functional ribosomal protein SA. A shortage of the normal protein likely impairs the assembly of ribosomes, but the specific effects of the mutations are not known. It is unclear why *RPSA* gene mutations appear to solely affect development of the spleen.

When isolated congenital asplenia is not caused by mutations in the *RPSA* gene, the cause of the condition is unknown.

[Learn more about the gene associated with Isolated congenital asplenia](#)

- *RPSA*

## **Inheritance**

Isolated congenital asplenia caused by mutations in the *RPSA* gene 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, an affected person inherits the mutation from one affected parent. Other cases result from new mutations in the gene that occur during the formation of reproductive cells (eggs or sperm) in an affected individual's parent or in early embryonic development. These cases occur in people with no history of the disorder in their family.

For unknown reasons, some people with an *RPSA* gene mutation that has been associated with isolated congenital asplenia have a normal spleen. The condition is said to have incomplete penetrance because not everyone with an *RPSA* gene mutation develops the condition.

When the cause of isolated congenital asplenia is unknown, the inheritance of the condition is unclear.

## **Other Names for This Condition**

- Asplenia, familial
- Asplenia, isolated congenital
- Congenital hypoplasia of spleen
- Hypoplasia of spleen
- Hyposplenia, isolated congenital
- ICAS
- Splenic hypoplasia

## **Additional Information & Resources**

### Genetic Testing Information

- Genetic Testing Registry: Familial isolated congenital asplenia (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0685889/>)

### Patient Support and Advocacy Resources

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

### Clinical Trials

- ClinicalTrials.gov (<https://clinicaltrials.gov/search?cond=%22Isolated congenital asplenia%22>)

### Catalog of Genes and Diseases from OMIM

- ASPLENIA, ISOLATED CONGENITAL; ICAS (<https://omim.org/entry/271400>)

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28isolated+congenital+asplenia%5BTIAB%5D%29+OR+%28congenital+asplenia%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>)

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