# L1CAM Gene

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## 1. Introduction

The *L1CAM* gene provides instructions for producing the L1 cell adhesion molecule protein (shortened to L1 protein), which is found on the surface of nerve cells (neurons) throughout the nervous system. The L1 protein spans the cell membrane, with one end of the protein inside the cell and the other end projecting from the outer surface of the cell. This positioning allows the L1 protein to attach (bind) to other proteins, including other L1 proteins, on neighboring neurons to help these cells stick to one another (cell-cell adhesion).

The L1 protein plays a role in the movement (migration) and organization of neurons and the outgrowth of axons, which are specialized extensions of neurons that transmit nerve impulses. The protein also plays a role in the formation of the protective sheath (myelin) that surrounds certain neurons and the formation of junctions between nerve cells (synapses), where cell-to-cell communication occurs. These neuronal functions contribute to brain development, thinking ability, memory, and movement.

## 2. Health Conditions Related to Genetic Changes

### 2.1. L1 Syndrome

More than 350 mutations in the *L1CAM* gene have been found to cause L1 syndrome. L1 syndrome describes a group of conditions that vary in severity, primarily affect the nervous system, and occur almost exclusively in males. People with L1 syndrome often have brain abnormalities, intellectual disability, and movement problems. The *L1CAM* gene mutations that cause this condition lead to an L1 protein that cannot facilitate cell-cell adhesion or participate in various neuronal functions. Disruption of these functions likely impedes the growth and development of the brain, leading to the signs and symptoms of L1 syndrome.

Some *L1CAM* gene mutations result in the production of a protein that is abnormally short and nonfunctional or result in a complete absence of protein. These mutations typically lead to severe cases of L1 syndrome. Other mutations change single protein building blocks (amino acids) in the L1 protein, impairing the protein's ability to interact with other proteins at the cell surface or preventing the protein from reaching the cell surface where it is needed. These mutations typically lead to the milder forms of L1 syndrome. While a gene mutation's effect on the L1 protein can sometimes provide a clue to the severity of the condition, individuals with the same or similar mutations often have very different signs and symptoms.

### 2.2. Other Disorders

Rarely, mutations in the *L1CAM* gene have also been found in individuals with both a buildup of fluid in the brain (hydrocephalus) and Hirschsprung disease. Hirschsprung disease is an intestinal disorder characterized by the absence of nerves in parts of the intestine. It is unclear whether this set of features should be classified as part of the L1 syndrome group of disorders (described above) or as a separate condition. It is likely that the *L1CAM* gene mutations that cause these features contribute to disruption of the normal migration and function of nerve cells in the brain and intestine, leading to hydrocephalus and Hirschsprung disease.

## 3. Other Names for This Gene

- antigen identified by monoclonal antibody R1

- CAML1
- CD171
- L1CAM_HUMAN
- MIC5
- N-CAML1
- neural cell adhesion molecule L1

## References


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