CALR Gene

Subjects: Genetics & Heredity
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calreticulin

Keywords: genes

1. Normal Function

The *CALR* gene provides instructions for making a multi-functional protein called calreticulin. This protein is found in several parts of the cell, including inside a structure called the endoplasmic reticulum (ER), in the fluid-filled space inside the cell (the cytoplasm), and at the outer surface of the cell. The ER is involved in protein processing and transport, and within this structure, calreticulin plays a role in ensuring the proper folding of newly formed proteins. The ER is also a storage location for charged calcium atoms (calcium ions), and calreticulin is involved in maintaining the correct levels of calcium ions in this structure. Through calcium regulation and other mechanisms, calreticulin is thought to play a role in the control of gene activity, cell growth and division (proliferation) and movement (migration), the attachment of cells to one another (adhesion), and regulation of programmed cell death (apoptosis). The function of this protein is important for immune system function and wound healing.

2. Health Conditions Related to Genetic Changes

2.1. 19p13.13 Deletion Syndrome

19p13.13 deletion syndrome

2.2. Essential Thrombocythemia

Some gene mutations are acquired during a person's lifetime and are present only in certain cells. These changes, which are called somatic mutations, are not inherited. Somatic mutations in the *CALR* gene in early blood-forming cells are associated with essential thrombocythemia, a disorder characterized by an increased number of platelets, the blood cells involved in normal blood clotting. The mutations associated with this condition remove or add small amounts of genetic material to a region of the *CALR* gene called exon 9. These genetic changes lead to production of an altered calreticulin protein with a different sequence of building blocks (amino acids) at one end. It is not clear how the alteration in calreticulin affects the protein's function or leads to the signs and symptoms of essential thrombocythemia.

2.3. Primary Myelofibrosis

Somatic mutations in exon 9 of the *CALR* gene are also associated with primary myelofibrosis, a condition in which bone marrow is replaced by scar tissue (fibrosis). The effect of the genetic changes on calreticulin function is unknown, and researchers are working to determine how the altered protein is involved in primary myelofibrosis.

3. Other Names for This Gene

- calregulin
- calreticulin precursor
- cC1qR
- CRP55
- CRT
- endoplasmic reticulum resident protein 60
- epididymis secretory sperm binding protein Li 99n
- ERp60
- FLJ26680

- grp60
- HACBP
- HEL-S-99n
- RO
- Sicca syndrome antigen A (autoantigen Ro; calreticulin)
- SSA

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