SPECC1L Gene

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sperm antigen with calponin homology and coiled-coil domains 1 like

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1. Normal Function

The *SPECC1L* gene provides instructions for making a protein called cytospin-A. This protein stabilizes components of the cell's structural framework (cytoskeleton) called microtubules, which are rigid, hollow fibers that help maintain the cell's shape. Stabilization of microtubules is necessary for these fibers to regulate various cell processes including the movement of cells to their proper location (cell migration). In order for cells to move, microtubules elongate in a specific direction, changing the shape of the cytoskeleton and allowing the cell to move in that direction. Migration of cells to their proper location during development ensures normal tissue formation.

During development of the embryo, cytospin-A plays a role in the migration of cells called neural crest cells, which originate in the developing spinal cord and migrate to specific regions in the embryo to form different structures. Cytospin-A is specifically involved in the migration of neural crest cells that come together to form the forehead, nasal bridge, and lower jaw.

2. Health Conditions Related to Genetic Changes

2.1. Opitz G/BBB syndrome

At least two mutations in the *SPECC1L* gene have been found to cause Opitz G/BBB syndrome. This condition causes several abnormalities along the midline of the body, including widely spaced eyes (ocular hypertelorism), throat malformations that can cause difficulty breathing or swallowing, brain malformations, distinct facial features, and genital abnormalities in males. The *SPECC1L* gene mutations that cause Opitz G/BBB syndrome change single protein building blocks (amino acids) in the cytospin-A protein, reducing the protein's ability to interact with components of the cytoskeleton. As a result, microtubules are disorganized and cells have trouble migrating to their proper location. Because the *SPECC1L* gene plays a role in facial development, mutations in this gene likely account for an opening in the lip (cleft lip) with or without an opening in the roof of the mouth (cleft palate) seen in some individuals with Opitz G/BBB syndrome. However, it is unclear how *SPECC1L* gene mutations cause the other features of this disorder.

2.2. Other disorders

Mutations in the *SPECC1L* gene have also been found to cause openings in facial structures called oblique facial clefts, also known as Tessier clefts. These clefts are similar to cleft lip and palate, but are more severe. The facial clefts can occur on one or both sides of the face; they typically involve the mouth and can include tissues up to the eyes. Health problems experienced by affected individuals depend on the severity of the cleft, but generally include problems eating and breathing.

The *SPECC1L* gene mutations that cause oblique facial clefts diminish the function of the cytospin-A protein. As a result, the protein cannot interact with the cytoskeleton, and microtubules are disorganized. These problems with the cytoskeleton impair cell migration, particularly of cells that come together to form the structures of the face. A decrease in cytospin-A function impairs cell migration during facial development, leaving openings, or clefts, where tissue should normally be.

3. Other Names for This Gene

- cytokinesis and spindle organization A

- cytospin A
- CYTSA
- KIAA0376
- sperm antigen with calponin homology and coiled-coil domains 1-like

References

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