Stromme syndrome is a very rare genetic condition. It affects multiple bodily systems and causes anomalies in the intestines, eyes, and skull.

It can also affect other areas of the body, such as the renal and cardiac systems.

Genetic mutations in the CENPF gene cause Stromme syndrome to occur. The CENPF gene is involved in DNA regulation and synthesis, so mutations to it can affect skeletal development.

Infants with Stromme syndrome are born with an incomplete intestine, ocular anomalies, and, in most cases, a smaller-than-average skull.

Causes

Genetic mutations to the CENPF gene cause Stromme syndrome.

This gene codes for centromere protein F. The position of this protein suggests that it plays a role in chromosome segregation.

Chromosome segregation occurs when two sister chromatids migrate to opposite poles of the cell nucleus after separating from each other.

Symptoms

Three typical symptoms characterize Stromme syndrome. These are intestinal atresia, ocular anomalies, and cranial anomalies.

The following sections will look at these in more detail.

1. Intestinal atresia

One of the main symptoms of Stromme syndrome is jejunal, or intestinal, atresia. Infants with Stromme syndrome are born with intestinal atresia, which refers to the incomplete formation of part of the small intestine.

Intestinal atresia, also known as apple peel syndrome, causes a section of the small intestine to twist around the main artery that sends blood to the colon. This can cause blockages to the intestine.

2. Ocular anomalies

Infants born with Stromme syndrome tend to have underdeveloped eyes and a variety of eye anomalies. These may include:

- Sclerocornea: This is an eye abnormality wherein the cornea blends with the white outer layer of the eyeball, causing there to be no clear-cut boundary between them.
- Microphthalmia: This is a condition wherein one or both of the eyes are unusually small.
- Microcornea: This is a condition wherein one or both of the corneas are unusually small.
- Ptosis: This refers to the drooping or falling of the upper eyelid.
- Epicanthus: This refers to a fold of skin on the upper eyelid that covers the inner corner of the eye.

3. Cranial anomalies

Another common symptom of Stromme syndrome is the presence of cranial anomalies. Infants with Stromme syndrome are likely to have microcephaly. This is a condition that causes an infant to have a much smaller head than usual.

However, not all infants with Stromme syndrome have microcephaly. Some infants with this condition have a typical head circumference.