Meckel Syndrome

Subjects: Genetics & Heredity
Contributor: Rita Xu

Meckel syndrome is a disorder with severe signs and symptoms that affect many parts of the body.

Keywords: genetic conditions

1. Introduction

The most common features are enlarged kidneys with numerous fluid-filled cysts; an occipital encephalocele, which is a sac-like protrusion of the brain through an opening at the back of the skull; and the presence of extra fingers and toes (polydactyly). Most affected individuals also have a buildup of scar tissue (fibrosis) in the liver.

Other signs and symptoms of Meckel syndrome vary widely among affected individuals. Numerous abnormalities of the brain and spinal cord (central nervous system) have been reported in people with Meckel syndrome, including a group of birth defects known as neural tube defects. These defects occur when a structure called the neural tube, a layer of cells that ultimately develops into the brain and spinal cord, fails to close completely during the first few weeks of embryonic development. Meckel syndrome can also cause problems with development of the eyes and other facial features, heart, bones, urinary system, and genitalia.

Because of their serious health problems, most individuals with Meckel syndrome die before or shortly after birth. Most often, affected infants die of respiratory problems or kidney failure.

2. Frequency

Meckel syndrome affects 1 in 13,250 to 1 in 140,000 people worldwide. It is more common in certain populations; for example, the condition affects about 1 in 9,000 people of Finnish ancestry and about 1 in 3,000 people of Belgian ancestry.

3. Causes

Meckel syndrome can be caused by mutations in one of at least eight genes. The proteins produced from these genes are known or suspected to play roles in cell structures called cilia. Cilia are microscopic, finger-like projections that stick out from the surface of cells and are involved in signaling pathways that transmit information between cells. Cilia are important for the structure and function of many types of cells, including brain cells and certain cells in the kidneys and liver.

Mutations in the genes associated with Meckel syndrome lead to problems with the structure and function of cilia. Defects in these cell structures probably disrupt important chemical signaling pathways during early development. Although researchers believe that defective cilia are responsible for most of the features of this disorder, it remains unclear how they lead to specific developmental abnormalities of the brain, kidneys, and other parts of the body.

Mutations in the eight genes known to be associated with Meckel syndrome account for about 75 percent of all cases of the condition. In the remaining cases, the genetic cause is unknown. Mutations in several other genes have been identified in people with features similar to those of Meckel syndrome, although it is unclear whether these individuals actually have Meckel syndrome or a related disorder (often described as a "Meckel-like phenotype").

3.1. The Gene Associated with Meckel Syndrome

CEP290

3.1.1. Additional Information from NCBI Gene:

- B9D1
- B9D2
- CC2D2A
- MKS1
- RPGRIP1L
- TMEM216
- TMEM67

4. Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

5. Other Names for This Condition

- dysencephalia splanchnocystica
- Meckel-Gruber syndrome
- MKS

References

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