Senior-Løken Syndrome

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Senior-Løken syndrome is a rare disorder characterized by the combination of two specific features: a kidney condition called nephronophthisis and an eye condition known as Leber congenital amaurosis.

Keywords: genetic conditions

1. Introduction

Nephronophthisis causes fluid-filled cysts to develop in the kidneys beginning in childhood. These cysts impair kidney function, initially causing increased urine production (polyuria), excessive thirst (polydipsia), general weakness, and extreme tiredness (fatigue). Nephronophthisis leads to end-stage renal disease (ESRD) later in childhood or in adolescence. ESRD is a life-threatening failure of kidney function that occurs when the kidneys are no longer able to filter fluids and waste products from the body effectively.

Leber congenital amaurosis primarily affects the retina, which is the specialized tissue at the back of the eye that detects light and color. This condition causes vision problems, including an increased sensitivity to light (photophobia), involuntary movements of the eyes (nystagmus), and extreme farsightedness (hyperopia). Some people with Senior-Løken syndrome develop the signs of Leber congenital amaurosis within the first few years of life, while others do not develop vision problems until later in childhood.

2. Frequency

Senior-Løken syndrome is a rare disorder, with an estimated prevalence of about 1 in 1 million people worldwide. Only a few families with the condition have been described in the medical literature.

3. Causes

Senior-Løken syndrome can be caused by mutations in one of at least five 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; they are involved in signaling pathways that transmit information between cells. Cilia are important for the structure and function of many types of cells, including certain cells in the kidneys. They are also necessary for the perception of sensory input (such as vision, hearing, and smell).

Mutations in the genes associated with Senior-Løken syndrome likely lead to problems with the structure and function of cilia. Defects in these cell structures probably disrupt important chemical signaling pathways within cells. Although researchers believe that defective cilia are responsible for the features of this disorder, it remains unclear how they lead specifically to nephronophthisis and Leber congenital amaurosis.

Some people with Senior-Løken syndrome do not have identified mutations in one of the five genes known to be associated with the condition. In these cases, the genetic cause of the disorder is unknown.

3.1. The Genes Associated with Senior-Løken Syndrome

- CEP290
- NPHP1
- WDR19

3.2. Additional Information from NCBI Gene:

- IQCB1
- NPHP4
- SDCCAG8

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

- Loken-Senior syndrome
- renal dysplasia and retinal aplasia
- renal-retinal syndrome
- Senior-Loken syndrome

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