OFD1 Gene

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1. Introduction

The *OFD1* gene provides instructions for making a protein whose function is not fully understood. It appears to play a critical role in the early development of many parts of the body, including the brain, face, limbs, and kidneys.

The OFD1 protein is located at the base of cilia, which are finger-like projections that stick out from the surface of cells. Cilia are involved in cell movement and in many different chemical signaling pathways. They play important roles in the development and function of many parts of the body. Researchers suspect that the OFD1 protein is essential for the normal formation of cilia.

Studies suggest that the OFD1 protein may have additional functions. In the earliest stages of development, it appears to be involved in determining the left-right axis (the imaginary line that separates the left and right sides of the body). The OFD1 protein is also found in the nucleus, although its function in this cell structure is unknown.

2. Health Conditions Related to Genetic Changes

2.1. Oral-facial-digital syndrome

About 100 mutations in the *OFD1* gene have been found in people with oral-facial-digital syndrome type I, which is the most common form of the disorder. These mutations include changes in single DNA building blocks (base pairs) and larger deletions of genetic material from the *OFD1* gene. Most of these genetic changes lead to the production of an abnormally short, nonfunctional version of the OFD1 protein. It is unclear how a shortage of functional OFD1 protein leads to the specific features of oral-facial-digital syndrome type I. However, studies suggest that a lack of this protein prevents the normal formation of cilia, which affects the development of many tissues and organs.

2.2. Other disorders

Mutations in the *OFD1* gene can also cause several other disorders with features that overlap with those of oral-facial-digital syndrome. At least two mutations have been identified in people with Joubert syndrome. This disorder is characterized by particular brain abnormalities, weak muscle tone (hypotonia), delayed development, unusual eye movements, and breathing problems. Another *OFD1* gene mutation has been found in a family with a form of X-linked intellectual disability. Affected individuals have had severe intellectual disability, an unusually large head size (macrocephaly), and breathing problems.

The *OFD1* mutations responsible for Joubert syndrome and X-linked intellectual disability lead to the production of an abnormally short version of the OFD1 protein. However, studies suggest that these mutations result in a somewhat longer protein than the mutations that cause oral-facial-digital syndrome. This protein may retain some of its usual function in cilia. The abnormal protein probably disrupts the development or function of cilia in some way, although it is unclear how these changes result in the signs and symptoms of Joubert syndrome or X-linked intellectual disability.

3. ther Names for This Gene

- 71-7A
- CXorf5
- JBTS10

- MGC117039
- MGC117040
- OFD1_HUMAN
- oral-facial-digital syndrome 1
- SGBS2

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