PAX2 Gene

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paired box 2

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1. Introduction

The *PAX2* gene belongs to a family of genes that plays a critical role in the formation of tissues and organs during embryonic development. The members of the PAX gene family are also important for maintaining the normal function of certain cells after birth. To carry out these roles, the PAX genes provide instructions for making proteins that attach to specific areas of DNA and help control the activity (expression) of particular genes. On the basis of this action, PAX proteins are called transcription factors.

During embryonic development, the *PAX2* gene provides instructions for producing a protein that is involved in the formation of the eyes, ears, brain and spinal cord (central nervous system), kidneys, urinary tract, and genital tract. After birth, the PAX2 protein is thought to protect against cell death during periods of cellular stress.

2. Health Conditions Related to Genetic Changes

2.1. Renal coloboma syndrome

More than 40 mutations in the *PAX2* gene have been found to cause renal coloboma syndrome. Most mutations are specific to each affected family; however, one mutation has been found in multiple affected individuals. This mutation inserts one DNA building block (nucleotide) into the *PAX2* gene (written as 619insG). Most mutations occur in the region of the protein that attaches to DNA, impairing its function as a transcription factor. A lack of functional PAX2 protein disrupts the formation of certain tissues (particularly the kidneys and eyes) during embryonic development, causing the signs and symptoms of renal coloboma syndrome.

2.2. Congenital anomalies of kidney and urinary tract

More than 20 mutations in the *PAX2* gene have been found in people with abnormalities of the kidneys and other structures of the urinary system but without the eye problems of renal coloboma syndrome (described above). The urinary system abnormalities vary in severity and are grouped together as congenital anomalies of kidney and urinary tract (CAKUT). The most severe CAKUT abnormalities can cause kidney damage and life-threatening kidney failure.

The effects of CAKUT-associated *PAX2* gene mutations are not fully understood, but it is likely that they impair the function of the PAX2 protein, disrupting formation of the kidneys and urinary system during embryonic development. It is unclear why only structures of the urinary system are affected in these individuals.

2.3. Other disorders

PAX2 gene mutations are also found in individuals with abnormalities of the optic nerve, which carries visual information from the eyes to the brain. These individuals do not have the kidney anomalies associated with renal coloboma syndrome (described above). As in renal coloboma syndrome, the *PAX2* gene mutations associated with eye abnormalities likely disrupt regulation of genes that help direct normal eye development. Researchers are working to understand why mutations in this gene can affect different organ systems in different people.

3. Other Names for This Gene

- paired box gene 2

- paired box homeotic gene 2
- paired box protein 2

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