PITX2 Gene

Subjects: Genetics & Heredity
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paired like homeodomain 2

genes

1. Introduction

The *PITX2* gene provides instructions for making a protein that attaches (binds) to specific regions of DNA and regulates the activity of other genes. On the basis of this action, the PITX2 protein is called a transcription factor. The *PITX2* gene is part of a family of homeobox genes, which act during early embryonic development to control the formation of many parts of the body.

The PITX2 protein plays a critical role in early development, particularly in the formation of structures in the front part of the eye (the anterior segment). These structures include the colored part of the eye (the iris), the lens of the eye, and the clear front covering of the eye (the cornea). Studies suggest that the PITX2 protein also has functions in the adult eye, such as helping cells respond to oxidative stress. Oxidative stress occurs when unstable molecules called free radicals accumulate to levels that can damage or kill cells.

The PITX2 protein is also involved in the normal development of other parts of the body, including the teeth, heart, and abdominal organs.

2. Health Conditions Related to Genetic Changes

2.1. Axenfeld-Rieger syndrome

More than 45 mutations in the *PITX2* gene have been found to cause Axenfeld-Rieger syndrome type 1, a condition that affects the development of the anterior segment of the eye and other parts of the body. Most *PITX2* gene mutations reduce the amount of functional PITX2 protein that is produced in cells. However, some genetic changes (such as a duplication of the *PITX2* gene) increase the amount or function of the PITX2 protein. Having either too little or too much of this protein disrupts the regulation of other genes needed for normal development.

Eye development appears to be the most sensitive to changes in PITX2 protein activity, and abnormalities of the anterior segment of the eye are the predominant features of Axenfeld-Rieger syndrome. However, changes in the

amount of PITX2 protein can also lead to distinctive facial features, tooth abnormalities, and problems with development of other parts of the body in people with this condition.

2.2. Peters anomaly

At least one mutation in the *PITX2* gene has been found to cause Peters anomaly. This condition is characterized by abnormal development of the anterior segment of the eye and clouding of the cornea. This mutation (written IVS3AS,A>T,-2) alters the way the PITX2 protein is pieced together. It is thought that this altered protein disrupts the regulation of movement of cells to their proper location in the developing anterior segment, leading to abnormal formation of the structures in this area of the eye and other features of Peters anomaly.

2.3. Other disorders

Mutations in the *PITX2* gene have also been identified in other eye disorders. Like Axenfeld-Rieger syndrome and Peters anomaly, these conditions primarily involve the anterior segment of the eye. Mutations in the *PITX2* gene can cause ring dermoid of the cornea, a condition associated with tumor-like growths on the cornea. Additionally, conditions that cause underdevelopment of the iris can occur, including iris hypoplasia or iridogoniodysgenesis type 2. Iridogoniodysgenesis type 2 is also associated with an elevated risk of increased pressure in the eye (glaucoma).

3. Other Names for This Gene

- all1-responsive gene 1
- ARP1
- Brx1
- IDG2
- IGDS
- IGDS2
- IHG2
- IRID2
- Otlx2
- paired-like homeodomain 2
- paired-like homeodomain transcription factor 2
- pituitary homeobox 2
- PITX2_HUMAN
- PTX2
- RGS
- RIEG
- rieg bicoid-related homeobox transcription factor 1
- RIEG1

- RS
- solurshin

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