

Frasier Syndrome

Subjects: Genetics & Heredity

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Frasier syndrome is a condition that affects the kidneys and genitalia.

Keywords: genetic conditions

1. Introduction

Frasier syndrome is characterized by kidney disease that begins in early childhood. Affected individuals have a condition called focal segmental glomerulosclerosis, in which scar tissue forms in some glomeruli, which are the tiny blood vessels in the kidneys that filter waste from blood. In people with Frasier syndrome, this condition often leads to kidney failure by adolescence.

Although males with Frasier syndrome have the typical male chromosome pattern (46,XY), they have gonadal dysgenesis, in which external genitalia do not look clearly male or clearly female (ambiguous genitalia) or the genitalia appear completely female. The internal reproductive organs (gonads) are typically undeveloped and referred to as streak gonads. These abnormal gonads are nonfunctional and often become cancerous, so they are usually removed surgically early in life.

Affected females usually have normal genitalia and gonads and have only the kidney features of the condition. Because they do not have all the features of the condition, females are usually given the diagnosis of isolated nephrotic syndrome.

2. Frequency

Frasier syndrome is thought to be a rare condition; approximately 50 cases have been described in the scientific literature.

3. Causes

Mutations in the *WT1* gene cause Frasier syndrome. The *WT1* gene provides instructions for making a protein that regulates the activity of other genes by attaching (binding) to specific regions of DNA. On the basis of this action, the *WT1* protein is called a transcription factor. The *WT1* protein plays a role in the development of the kidneys and gonads (ovaries in females and testes in males) before birth.

The *WT1* gene mutations that cause Frasier syndrome lead to the production of a protein with an impaired ability to control gene activity and regulate the development of the kidneys and reproductive organs, resulting in the signs and symptoms of Frasier syndrome.

Frasier syndrome has features similar to another condition called Denys-Drash syndrome, which is also caused by mutations in the *WT1* gene. Because these two conditions share a genetic cause and have overlapping features, some researchers have suggested that they are part of a spectrum and not two distinct conditions.

3.1. The Gene Associated with Frasier Syndrome

- *WT1*

4. Inheritance

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

5. Other Names for This Condition

- FS

References

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