Frasier syndrome

Description

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

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 or the genitalia appear female-typical. 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.

Frequency

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

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.

Learn more about the gene associated with Frasier syndrome

- WT1

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.

Other Names for This Condition

- FS

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Frasier syndrome (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0950122/)

Genetic and Rare Diseases Information Center

- Frasier syndrome (https://rarediseases.info.nih.gov/diseases/2375/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Frasier syndrome%22)

Catalog of Genes and Diseases from OMIM

- FRASIER SYNDROME (https://omim.org/entry/136680)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28Frasier+syndrome%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D)

References

- Andrade JG, Guaragna MS, Soardi FC, Guerra-Junior G, Mello MP, Maciel-GuerraAT. Clinical and genetic findings of five patients with WT1-related disorders. Arq Bras Endocrinol Metabol. 2008 Nov;52(8):1236-43. doi:10.1590/s0004-27302008000800006. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/19169475)
- Fujita S, Sugimoto K, Miyazawa T, Yanagida H, Tabata N, Okada M, Takemura T. Afemale infant with Frasier syndrome showing splice site mutation in Wilms' tumorgene (WT1) intron 9. Clin Nephrol. 2010 Jun;73(6):487-91. doi: 10.5414/cnp73487. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/20497763)
- Niaudet P, Gubler MC. WT1 and glomerular diseases. Pediatr Nephrol. 2006Nov;21(11):1653-60. doi: 10.1007/s00467-006-0208-1. Epub 2006 Aug 23. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16927106)

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