Frasier syndrome

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 (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.

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.
Inheritance Pattern

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

Diagnosis & Management

Genetic Testing Information

• What is genetic testing?
  /primer/testing/genetictesting

• Genetic Testing Registry: Frasier syndrome

Research Studies from ClinicalTrials.gov

• ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22Frasier+syndrome%22

Other Diagnosis and Management Resources

• MedlinePlus Encyclopedia: Focal Segmental Glomerulosclerosis
  https://medlineplus.gov/ency/article/000478.htm

• MedlinePlus Encyclopedia: Nephrotic Syndrome
  https://medlineplus.gov/ency/article/000490.htm

Additional Information & Resources

Health Information from MedlinePlus

• Encyclopedia: Ambiguous Genitalia
  https://medlineplus.gov/ency/article/003269.htm

• Encyclopedia: Focal Segmental Glomerulosclerosis
  https://medlineplus.gov/ency/article/000478.htm

• Encyclopedia: Nephrotic Syndrome
  https://medlineplus.gov/ency/article/000490.htm

• Health Topic: Kidney Diseases
  https://medlineplus.gov/kidneydiseases.html

• Health Topic: Kidney Failure
  https://medlineplus.gov/kidneyfailure.html
Genetic and Rare Diseases Information Center

- Frasier syndrome

Additional NIH Resources

- National Institute of Diabetes and Digestive and Kidney Diseases: Glomerular Diseases
  https://www.niddk.nih.gov/health-information/kidney-disease/glomerular-diseases

Educational Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology: Frasier Syndrome
  http://atlasgeneticsoncology.org/Kprones/FrasierID10035.html
- Johns Hopkins Medicine: Glomerulosclerosis
  https://www.hopkinsmedicine.org/health/conditions-and-diseases/glomerulosclerosis
- MalaCards: frasier syndrome
  https://www.malacards.org/card/frasier_syndrome
- Merck Manual Consumer Version: Nephrotic Syndrome
- Orphanet: Frasier syndrome
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=347

Patient Support and Advocacy Resources

- March of Dimes: Genital and Urinary Tract Defects
- National Kidney Foundation: Focal Glomerulosclerosis
  https://www.kidney.org/atoz/content/focal
- University of Kansas Resource List: Kidney/Urological Conditions
  http://www.kumc.edu/gec/support/kidney.html
- University of Kansas Resource List: Sexuality and Sexual Differentiation Syndromes
  http://www.kumc.edu/gec/support/ambig.html

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28Frasier+syndrome%5BTIAB%5D%29+AND+english%5BLa%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D
Catalog of Genes and Diseases from OMIM

- **FRASIER SYNDROME**
  http://omim.org/entry/136680

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19169475

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20497763

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16927106

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