Renal coloboma syndrome

Renal coloboma syndrome (also known as papillorenal syndrome) is a condition that primarily affects kidney (renal) and eye development. People with this condition typically have kidneys that are small and underdeveloped (hypoplastic), which can lead to end-stage renal disease (ESRD). This serious disease occurs when the kidneys are no longer able to filter fluids and waste products from the body effectively. It has been estimated that approximately ten percent of children with hypoplastic kidneys may have renal coloboma syndrome. The kidney problems can affect one or both kidneys.

Additionally, people with renal coloboma syndrome may have a malformation in the optic nerve, a structure that carries information from the eye to the brain. Optic nerve malformations are sometimes associated with a gap or hole (coloboma) in the light-sensitive tissue at the back of the eye (the retina). The vision problems caused by these abnormalities can vary depending on the size and location of the malformation. Some people have no visual problems, while others may have severely impaired vision.

Less common features of renal coloboma syndrome include backflow of urine from the bladder (vesicoureteral reflux), multiple kidney cysts, loose joints, and mild hearing loss.

Frequency

The prevalence of renal coloboma syndrome is unknown; at least 60 cases have been reported in the scientific literature.

Causes

Renal coloboma syndrome is caused by mutations in the PAX2 gene. The PAX2 gene provides instructions for making a protein that is involved in the early development of the eyes, ears, brain and spinal cord (central nervous system), kidneys, and genital tract. The PAX2 protein attaches (binds) to specific regions of DNA and regulates the activity of other genes. On the basis of this role, the PAX2 protein is called a transcription factor. After birth, the PAX2 protein is thought to protect against cell death during periods of cellular stress.

Mutations in the PAX2 gene lead to the production of a nonfunctional PAX2 protein that is unable to aid in development, causing incomplete formation of certain tissues. Why the kidneys and eyes are specifically affected by PAX2 gene mutations is unclear.

Approximately half of those affected with renal coloboma syndrome do not have an identified mutation in the PAX2 gene. In these cases, the cause of the disorder is unknown.
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
• coloboma of optic nerve with renal disease
• coloboma-ureteral-renal syndrome
• ONCR
• optic coloboma, vescicoureteral reflux, and renal anomalies
• optic nerve coloboma renal syndrome
• papillorenal syndrome
• RCS
• renal-coloboma syndrome

Diagnosis & Management
Genetic Testing Information
• What is genetic testing?
  /primer/testing/genetictesting
• Genetic Testing Registry: Renal coloboma syndrome

Other Diagnosis and Management Resources
• GeneReview: PAX2-Related Disorder
  https://www.ncbi.nlm.nih.gov/books/NBK1451

Additional Information & Resources
Health Information from MedlinePlus
• Health Topic: Kidney Diseases
  https://medlineplus.gov/kidneydiseases.html
• Health Topic: Optic Nerve Disorders
  https://medlineplus.gov/opticnervedisorders.html

Genetic and Rare Diseases Information Center
• Renal coloboma syndrome
Additional NIH Resources

• National Eye Institute: Coloboma

• National Institute of Diabetes and Digestive and Kidney Diseases: Vesicoureteral Reflux

Educational Resources

• MalaCards: papillorenal syndrome
  https://www.malacards.org/card/papillorenal_syndrome

• Merck Manual Consumer Version: Kidneys

• Orphanet: Renal coloboma syndrome
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=1475

Patient Support and Advocacy Resources

• MACS: Microphthalmia, Anophthalmia and Coloboma Support (UK)
  https://macs.org.uk/

• The Kidney and Urology Foundation of America
  http://www.kidneyurology.org/

Clinical Information from GeneReviews

• PAX2-Related Disorder
  https://www.ncbi.nlm.nih.gov/books/NBK1451

Scientific Articles on PubMed

• PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28renal+coloboma+sindrome%5BTIAB%5D%29+OR+%28papillorenal+sindrome%5BTIAB%5D%29+OR+%28renal-coloboma+sindrome%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2880+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

• PAPILLORENAL SYNDROME
  http://omim.org/entry/120330
Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11093271

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20301624

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17541647

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11581073

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/14566649

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16971658

Reprinted from Genetics Home Reference: 

Reviewed: July 2008
Published: July 7, 2020

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services