

# VETERINARY TECHNICAL DATASHEET

Xanthinuria, Type 1a



Mutation Found In :Mixed breed dogs

## Disorder Type

- Urinary

## Disease Severity

- Moderate/severe

## Background

Xanthinuria is a hereditary disease that leads to excessive xanthine in the urine. Excessive xanthine can accumulate in the urinary tract causing formation of xanthine stones that can obstruct the urinary tract or kidneys. Different breeds suffer from different forms of xanthinuria.

## Key Signs

- Difficulties when passing urine
- Pain when passing urine
- Inflammation in bladder
- Hematuria
- Kidney failure

## Clinical Description

This disorder can cause formation of stones throughout the upper and lower urinary tracts, including the urethra, bladder, ureters, and kidneys. Clinical signs of urolithiasis in the lower urinary tract include pain while urinating, bloody urine, and blockage of the urinary tract. Patients with urinary stones are more susceptible to urinary tract infections. Blockage of the ureters and kidneys causes abdominal pain and kidney failure. A blocked urinary tract is a life-threatening condition that requires immediate veterinary care. The disorder can develop at any age, starting as early as when a pup is a few months of age. Males are more likely to form clinically relevant stones than females. Urine sampling and ultrasound examinations are used for diagnostics. These stones are radiolucent and thus not clearly visible on radiographs.

## Mode of Inheritance

- autosomal recessive

## Gene Name

- Confidential

## Next Steps

Management of the disease consists of increasing water intake, limiting purine intake, and keeping the urine alkaline. Special low-purine diets are available. Affected dogs must be carefully monitored and follow-up visits are needed. A blocked urinary tract is a life-threatening condition that usually requires surgical treatment.

## References

E. Furrow, N. Tate, K. Minor, J. Mickelson, K. Peterson, and J. Lulich. Three diverse mutations underlying canine xanthine urolithiasis. ACVIM Research Report, 2016