Hyperuricosuria (HUU) in Dalmatians is a well described hereditary defect that was identified in 1916. All purebred Dalmatians tested have this change in urinary metabolism. The breakdown of dietary purines causes most dogs to excrete allantoin in their urine. Dalmatians, however, produce and excrete high levels of uric acid. This constant hyperuricosuria can result in the formation of urinary urate stones. About 20 percent of male Dalmatians require the surgical removal of urate stones, followed by lifelong medical treatment. The only other mammals that excrete uric acid in their urine are humans and some primates. Gout is the common end point of a group of disorders that cause hyperuricemia and hyperuricosuria in humans. Dalmatians are the only non-primate mammalian model for HUU.

A backcross was performed between a Dalmatian and a Pointer in order to more thoroughly investigate the mutation that causes HUU in Dalmatians. F1 animals were bred back to Dalmatians, and only offspring with low levels of uric acid in their urine were used for future breeding. The breeding program has been going on for nine generations, producing dogs that exhibit the phenotypic characteristics of the Dalmatian breed standard and carry the part of the Pointer’s genome around the HUU mutation. These dogs segregate HUU and their DNA was used in a linkage study.

Genotyping 153 different microsatellite markers spanning the 38 canine chromosomes enabled us to narrow down a region of linkage disequilibrium within the backcross family. The analysis of additional markers within the region enabled us to identify four candidate genes. These genes were sequenced in an effort to detect a causative mutation. A missense mutation in the SLC2A9 gene has been identified as the cause of huu in Dalmatians as well as other breeds, including Bulldogs and Black Russian Terriers. A DNA test for the mutation is available through the UC Davis Veterinary Genetics Laboratory: http://www.vgl.ucdavis.edu/services/dog.php