

Medullary Sponge Kidney is a rare disorder characterized by the formation of cystic malformations in the collecting ducts and the tubular structures within the kidneys (tubules) that collect urine. One or both kidneys may be affected. The initial symptoms of this disorder may include blood in the urine (hematuria), calcium stone formation in the kidneys (nephrolithiasis) or infection. The exact cause of Medullary Sponge Kidney is not known. The exact cause of Medullary Sponge Kidney is not known and most cases occur sporadically for no apparent reason. Some cases are thought to run in families (familial) and may be inherited as an autosomal dominant genetic trait. However, this inheritance pattern has not been proven. Some studies have suggested there may be a possible relationship between overactivity of the parathyroid gland (Hyperparathyroidism) and Medullary Sponge Kidney. Medullary Sponge Kidney is a rare disorder that affects slightly more women than men. It is thought to occur in 1 in 1,000 to 5,000 people in the United States. Although the symptoms of Medullary Sponge Kidney may begin at any age, they usually develop during adolescence or in adults between the ages of 30 and 50 years. Approximately 13 percent of all people who develop kidney stones are eventually diagnosed with Medullary Sponge Kidney. Medullary Sponge Kidney may also develop in people with Beckwith-Wiedemann Syndrome. (For more information on Beckwith-Wiedemann Syndrome, see the related disorders section of this report.)