7. FROM GENES TO STONES: EXPLORING GENETIC COMPONENTS IN CALCIUM OXALATE UROLITHIASIS

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Introduction. Calcium oxalate urolithiasis, characterized by the formation of kidney stones, presents a significant health burden worldwide. While environmental and dietary factors contribute to stone formation, recent studies highlight the pivotal role of genetic components in predisposing individuals to this condition. This research aims to explore the genetic underpinnings associated with calcium oxalate urolithiasis, elucidating the intricate interplay between genes and stone formation.

Aim of study. The primary objective of this study is to investigate the genetic factors involved in the pathogenesis of calcium oxalate urolithiasis. We aimed to identify specific genetic markers, pathways, and variations associated with stone formation. This exploration intends to provide a deeper understanding of the genetic landscape underlying this condition, potentially paving the way for targeted preventive strategies and therapeutic interventions.

Methods and materials. This abstract drew from research articles retrieved between 2015 and 2023 using keywords like "genetics," "Calcium oxalate urolithiasis," and "inheritance." Initially, 54 primary sources were identified, and 12 were chosen for analysis, forming the basis of this review's exploration into the genetic facets of calcium oxalate urolithiasis inheritance.

Results. Our investigations on urolithiasis revealed a spectrum of genetic variations within key genes involved in oxalate metabolism, calcium regulation, and renal transport mechanisms. Related to calcium oxalate urolithiasis there were identified the following genes: AGXT (involved in oxalate metabolism), SLC26A (oxalate transport in the kidney), 1CLCN5 (calcium transport), SLC34A1 (renal transport of calcium and phosphate), GRHPR (glyoxylate metabolism pathway), HOGA1 (metabolic pathway of hydroxyproline). Significantly higher frequencies of specific SNPs in these genes were observed in the urolithiasis cohort compared to the control group. Information related to mutations in these genes can help to identify drugs for personalized treatment.

Conclusion. The findings from this study underscore the substantial role of genetic components in calcium oxalate urolithiasis etiology. Identifying genetic variations and pathways associated with stone formation provides crucial insights into the underlying mechanisms. These discoveries hold promise for the development of targeted interventions, personalized treatment strategies, and improved risk assessment tools for individuals susceptible to calcium oxalate urolithiasis.

Keywords. Genetics, Calcium oxalate urolithiasis and inheritance.