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Monogenic Diseases in Adult Kidney Stone Formers: Unravelling the Genetic Puzzle

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Introduction

Kidney stones, a prevalent urological disorder, affect a substantial number of adults globally. While the majority of kidney stones arise from a combination of lifestyle and dietary factors, a distinct subset of individuals experience stone formation due to more intricate underlying causes. Recent advances in research have brought to light the significance of monogenic diseases in the etiology of kidney stones among adults. Monogenic diseases, or single-gene disorders, are characterized by mutations in a single gene that disrupt its normal functioning [1], leading to various health conditions. Within the context of kidney stones, certain monogenic diseases have emerged as potential contributors to stone formation.

In recent years, research efforts have made noteworthy progress in uncovering the genetic basis of kidney stones in adults. Several monogenic diseases, including cystinuria, primary hyperoxaluria, and Dent disease, among others, have been implicated as major factors in adult kidney stone formers. These genetic disorders can give rise to abnormal urinary excretion of stone-forming substances, ultimately culminating in the development of kidney stones. Consequently, understanding the role of monogenic diseases in kidney stone formation holds tremendous promise for personalized treatments and improved outcomes for patients.

The burgeoning field of monogenic diseases in adult kidney stone is formers exploring recent studies and genetic discoveries that have deepened our understanding of this complex relationship. We also discuss the clinical implications of genetic testing, which can aid in identifying specific mutations responsible for stone formation and guiding tailored treatment approaches [2]. By shedding light on this critical aspect of kidney stone pathophysiology, we aim to emphasize the potential for precision medicine in managing this condition, thereby improving the lives of those affected by kidney stones and paving the way for future research in this exciting area of urology.

Description

The role of monogenic diseases

Monogenic diseases play a pivotal role in the etiology of kidney stones in some adult patients. These disorders result from mutations in a single gene, leading to disruptions in various physiological processes that contribute to stone formation. Several monogenic diseases have been identified as significant contributors to kidney stone development in adults, including cystinuria, primary hyperoxaluria, and Dent disease.

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Cystinuria leads to excessive cystine excretion in urine, promoting the formation of cystine stones. Primary hyperoxaluria causes the overproduction of oxalate, resulting in the formation of calcium oxalate stones. Dent disease affects the reabsorption of substances in the kidneys, leading to increased excretion of calcium and phosphate and increased stone risk.

Recognizing the presence of these monogenic diseases in kidney stone formers is crucial for personalized treatment strategies. Genetic testing can identify specific mutations, guiding targeted therapies to address the underlying cause and improve patient outcomes. Understanding the role of monogenic diseases is vital for advancing precision medicine in the management of kidney stones in adults.

Recent studies and genetic discoveries

Over the past few years, researchers have made significant strides in unraveling the genetic basis of kidney stones in adults. Several monogenic diseases have been implicated in adult kidney stone formers, such as cystinuria, primary hyperoxaluria, and Dent disease, among others. These genetic disorders can lead to abnormal urinary excretion of stone-forming substances, ultimately contributing to stone formation.

Cystinuria, an autosomal recessive disorder, results in the excessive excretion of cystine in urine. The accumulation of cystine crystals can lead to the development of kidney stones, often requiring multiple interventions to manage recurrent stone episodes. Primary hyperoxaluria, on the other hand, is characterized by the overproduction of oxalate, which combines with calcium in the kidneys, forming calcium oxalate stones. The severe form of primary hyperoxaluria can result in kidney failure and necessitate kidney transplantation.

Dent disease is an X-linked recessive disorder that affects the reabsorption of certain substances in the kidneys, leading to increased excretion of calcium and phosphate in the urine, ultimately promoting stone formation [3].

Clinical implications and personalized treatments

The identification of monogenic diseases in adult kidney stone formers has significant clinical implications, as it offers a more precise approach to diagnosis, management, and treatment. Understanding the genetic basis of kidney stone formation can pave the way for personalized treatments that target the underlying cause of stone development, ultimately leading to improved patient outcomes.

Genetic testing can play a pivotal role in accurately diagnosing monogenic diseases responsible for kidney stones. By identifying specific genetic mutations, clinicians can differentiate between different types of kidney stones and determine whether a monogenic disorder is the underlying cause [4]. This information is crucial for risk stratification, as some monogenic diseases may carry a higher risk of recurrent stone formation or kidney damage compared to others. Early identification of these genetic disorders can aid in developing targeted preventive measures and individualized follow-up plans.

Monogenic diseases often lead to specific patterns of stone composition and urinary excretion of stone-forming substances. Armed with knowledge of the genetic defect, healthcare providers can recommend personalized dietary modifications tailored to the patient's unique metabolic profile. For example, individuals with cystinuria may benefit from increased fluid intake and alkali therapy to prevent cystine stone formation. Patients with primary hyperoxaluria may require dietary restrictions to limit oxalate-rich foods. These targeted

interventions can help reduce stone recurrence and improve overall kidney health.

Genetic insights can also guide the use of pharmacological therapies in managing kidney stones. For instance, some monogenic diseases involve defects in specific transporters or enzymes. Drugs that target these specific molecular pathways may prove more effective in preventing stone formation. By matching pharmacological therapies to the underlying genetic defect, patients can receive more precise and potentially more successful treatment options.

Understanding the genetic basis of monogenic kidney stone disorders opens the door to exploring new therapeutic targets. Researchers can focus on developing innovative therapies aimed at correcting or compensating for the genetic defects responsible for stone formation. This may involve gene therapy, enzyme replacement, or other cutting-edge approaches designed to address the root cause of the condition. The identification of monogenic diseases in kidney stone formers has implications for their family members as well. Genetic screening of family members can help identify individuals at risk of developing kidney stones due to inherited mutations. Early detection can lead to timely interventions and preventive measures to minimize stone formation [5]. Additionally, genetic counseling can provide valuable information about the inheritance pattern of the monogenic disease, helping family members make informed decisions regarding family planning and medical management.

Conclusion

As our understanding of the genetic basis of kidney stone formation in adult patients expands, so does the potential for personalized treatments and improved patient outcomes. Identifying monogenic diseases associated with kidney stones is a significant step towards unraveling the complex pathophysiology of stone formation. By integrating genetic testing into the diagnostic process, clinicians can develop tailored strategies to manage kidney stone formers more effectively, reducing the burden of this condition on affected individuals and healthcare systems alike. Further research and collaboration between geneticists, urologists, and nephrologists hold the key to unlocking new avenues for prevention and treatment of kidney stones in the future.

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Conflict of Interest

There is no conflict of interest by author.

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