

Overview of the need for acute dialysis in genetic diseases

Genetik hastalıklarda akut diyaliz ihtiyacına genel bakış

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ABSTRACT

Dialysis is a renal replacement therapy used not only for patients with acute kidney injury but also for managing severe complications resulting from genetic pathologies. This letter focuses on acute dialysis applications, which, although not the primary treatment for genetic diseases, can be life-saving when necessary. Dialysis plays a critical role in conditions such as metabolic disorders that cause metabolic imbalances, including glycogen storage disease, acute kidney injury-inducing diseases such as polycystic kidney disease and Alport syndrome, severe electrolyte disturbances, hereditary cardiac diseases leading to excessive fluid overload, genetic diseases causing liver failure, as well as urea cycle disorders and organic acidemias requiring toxin removal. It is of significant importance in the treatment of end-stage renal disease and is employed when all other therapeutic options have failed. Issues such as equipment shortages, the risk of complications, and the necessity for specialized expertise should not be overlooked. Increasing awareness of these challenges can enhance clinical effectiveness and improve the survival outcomes of these patients.

ÖZ

Diyaliz, sadece akut böbrek hasarı olan hastalar için değil, aynı zamanda genetik patolojilerin yol açtığı ciddi komplikasyonları yönetmek için de kullanılan bir böbrek replasman tedavisidir. Bu mektubun odak noktası akut diyaliz uygulamaları olup, genetik hastalıkların birincil tedavisi olmasa da gerektiğinde hayat kurtarıcı olmaktadır. Diyaliz glikojen depo hastalığı gibi metabolik dengesizlik yapan metabolik hastalıklarda, polikistik böbrek hastalığı ve Alport sendromu gibi akut böbrek hasarı yapan hastalıklarda, ciddi elektrolit dengesizliklerinde, aşırı sıvı yüklenmesine yol açan kalıtsal kalp hastalıklarında ve karaciğer yetmezliğine yol açan genetik hastalıklarda, üre döngüsü bozuklukları ve organik asidemilerde toksinlerin uzaklaştırılmasında hayati müdahale sunar. Son dönem böbrek hastalığının tedavisinde büyük öneme sahip olup, diğer tüm yöntemlerin başarısız olduğu durumlarda uygulanmaktadır. Belirli ekipman eksiklikleri, komplikasyon riski ve deneyim gerekliliği gibi sorunlar göz ardı edilmemeli, farkındalığı artırmak klinik etkinliği ve bu hastaların hayatta kalma şansını iyileştirebilir.

Dear Editor;

The management of genetic diseases is currently considered one of the most complex and challenging areas of modern medicine (Levey, 2012). Among the various treatment methods used to improve the survival rates of these patients, dialysis plays a vital role.

Acute dialysis is a renal replacement therapy applied in genetic diseases as well as in many primary or secondary conditions that can cause acute kidney injury, such as infections, rheumatological conditions, trauma, and surgical diseases. The main indication in all cases is acute kidney injury, which occurs when the kidneys are unable to perform their glomerular filtration function adequately. Acute dialysis treatment is required when, despite all supportive and conservative treatments, desired metabolic and/or clinical goals (such as euvolemia) cannot be achieved. While acute dialysis is not the primary treatment for genetic diseases, it can be life-saving when necessary. Different types of acute dialysis treatments are effective for various genetic/metabolic diseases. In some conditions, peritoneal dialysis is recommended, while in others, high-flow hemodialysis or continuous slow replacement therapies (such as hemofiltration) are found to be more effective.

In this letter, I aim to raise awareness by presenting literature that emphasizes the importance of acute dialysis treatment when necessary in genetic diseases.

Acute Kidney Injury (AKI) and Dialysis: Some genetic diseases can lead to acute kidney injury (AKI) by directly affecting kidney functions. Diseases such as polycystic kidney disease and Alport syndrome can cause a sudden loss of kidney function. In the treatment of AKI, dialysis provides a crucial intervention by removing waste products, excess fluid, and electrolytes from the body when the kidneys can no longer filter adequately (Ortega-Loubon et al., 2021).

Metabolic Disorders and Dialysis: Certain genetic metabolic disorders, such as glycogen storage disease, are inherited conditions caused by enzyme deficiencies or dysfunctions. These can lead to clinical manifestations such as hypoglycemia, lactic acidosis, and organomegaly. They disrupt energy metabolism and cause structural damage to affected organs. As a result of metabolic imbalances, dialysis may be required and can provide significant improvements in the patient's overall clinical condition.

Electrolyte Imbalances and Dialysis: Genetic diseases such as cystic fibrosis and certain inherited renal tubulopathies can cause severe electrolyte imbalances in the body. These diseases often present with abnormalities in sodium, potassium, and blood gases. Although

fluid-electrolyte therapy is typically sufficient for management, persistent abnormal levels of electrolytes like potassium, calcium, and phosphate can lead to life-threatening conditions such as cardiac arrhythmias and muscle dysfunctions. Dialysis can quickly correct these electrolyte imbalances, potentially saving and stabilizing the patient's life.

Fluid Overload and Dialysis: Certain genetic diseases that lead to hereditary heart conditions or liver failure can cause excessive fluid accumulation in the body. This can result in severe complications such as pulmonary edema, making breathing difficult for patients. Dialysis is required when, despite all supportive treatments, the desired euvolemic state is not achieved. By rapidly removing excess fluid from the body, dialysis prevents such complications and improves the patient's overall condition.

Toxin Removal and Dialysis: Some genetic diseases lead to the accumulation of specific toxins in the body, necessitating dialysis to manage acute metabolic crises. For instance, conditions like urea cycle disorders and organic acidemias can cause rapid accumulation of such toxins in the blood. Dialysis facilitates the rapid clearance of these toxins, preventing neurological and systemic damage. In these situations, dialysis acts as a critical bridging therapy until the patient's condition is stabilized, improving their quality of life.

CONCLUSION

In conclusion, while acute dialysis is not the primary and sole treatment for patients with acute kidney injury due to genetic or underlying conditions, it can be life-saving when needed. Acute dialysis plays a critical role in the management of genetic diseases, offering vital intervention in cases of renal failure, metabolic crises, electrolyte imbalances, fluid overload, and toxin accumulation. Therefore, increasing awareness of the role of dialysis in the management of genetic diseases is crucial.

Regardless of the cause, attention must be given to the indications for acute dialysis in conditions such as acute kidney injury, intoxication, hypervolemia, and electrolyte imbalances. Initially, necessary conservative treatments, appropriate fluid-electrolyte therapy, and volume management should be provided. However, when acute dialysis is required, various parameters such as the patient's age, the center's experience, and technical facilities directly influence the success of the treatment and related complications. Not every dialysis method is applicable in every center or for every patient. There are significant risks including technical failure, infection, thrombosis, bleeding, and surgical complications that should not be overlooked.

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