Urea Cycle Disorders Consortium Newsletter

Winter 2024/2025

Dear Study Participants,

We want to take a moment to express our heartfelt gratitude for your invaluable participation in Urea Cycle Disorders Consortium (UCDC) research. Your contributions are vital to advancing our understanding of urea cycle disorders and improving care for individuals and families affected by these conditions.

Thanks to your support, we are thrilled to share several research accomplishments from 2024, including new papers published that highlight key findings made possible by your involvement. These publications reflect the power of collaboration between researchers and participants like you, and we are excited to continue building on these advancements.

As we look back on 2024, we also set our sights on the opportunities that lie ahead in 2025. Together, we can continue to make meaningful strides in advancing our mission to improve the lives of people with UCD.

From all of us at the UCDC, we wish you and your loved ones a happy, healthy new year!

Warm regards,
The UCDC Team

What's in this issue?

The following articles summarize recent research by the Urea Cycle Disorders Consortium (UCDC). Click "Read Full Summary" to open the full summary and access the original scientific paper by clicking the full summary title.
If you are interested in learning more about the UCDC and our research, please visit our website **here**.

1 Using Phenylbutyrate Metabolite Testing to Improve Urea Cycle Disorder Treatment

Testing phenylbutyrate metabolites can help manage urea cycle disorders (UCDs). The paper highlights that younger patients and those on higher doses may need closer monitoring. While ammonia and amino acid levels remain key markers, metabolite testing adds valuable insights into treatment safety and effectiveness.

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2 Quality of Life Challenges in Urea Cycle Disorders

Urea cycle disorders (UCDs) affect quality of life for kids and adults. Kids face more challenges with physical and emotional health, while adults report worse mental health. The findings emphasize the need for better mental healthcare and tools tailored to UCD patients.

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3 Impact of L-Citrulline/Arginine Supplementation After Liver Transplantation in Urea Cycle Disorders

L-citrulline/arginine supplementation might not improve hospital stays, growth, or brain function after liver transplants in people with urea cycle disorders. More research is needed to find better treatments for these patients.

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4 Health Risks for Females with Ornithine Transcarbamylase Deficiency (OTCD)

Females with Ornithine Transcarbamylase Deficiency (OTCD) might face health risks, even if they do not show severe symptoms. The paper aims to create a tool to predict which females are at higher risk for complications, helping doctors provide better care and prevent problems in the future.

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5 Liver Transplantation and Health Outcomes in Urea Cycle Disorders

Liver transplantation in UCD patients helps stabilize metabolism and improve growth but does not seem to affect cognitive function compared to medical management. The paper highlights the need for further research into transplant-related outcomes and the long-term effects on brain health.

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6 Understanding Seizures and Epilepsy in Urea Cycle Disorders

High ammonia levels in UCD patients can trigger seizures and increase the risk of epilepsy, especially in those with late-onset seizures. The findings suggest the need for regular EEG monitoring to detect both visible and hidden (subclinical) seizures, which can help prevent brain damage and improve patient care.

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How are we doing?
Leave your feedback by taking this quick survey.

Urea Cycle Disorders Consortium Website