PATIENT-CENTERED OUTCOMES RESEARCH INSTITUTE
RESEARCH SUMMARY

PROJECT INFORMATION

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Comparing Treatment Options for Urea Cycle Disorders

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Organization
Children's Research Institute

What was the research about?

Urea cycle disorders, or UCDs, are rare health disorders in which the body can't get rid of ammonia. Ammonia is a substance the body produces as it breaks down protein in food. Ammonia builds up in the bloodstream as a result of UCDs. This build-up can lead to brain damage, disability, or death.

One way to treat UCDs is with medical management. This treatment involves a low-protein diet and medicine that helps the body break down ammonia. Another way is with liver transplant.

In this study, the research team looked at the medical histories of patients with severe UCDs. The team wanted to learn if the health of patients who had liver transplants and those on medical management differed over time. They also wanted to see how families and doctors made the difficult decision about which treatment to use.

What were the results?

The research team didn't find major differences in survival, mental ability, or quality of life between patients who had liver transplants and those on medical management. For patients who had liver transplant sooner, the team saw a trend toward higher family quality of life and better mental ability. But the data didn't show these effects for sure.

In interviews, the research team learned that families and doctors considered many personal, social, and medical factors when deciding on treatment.

What did the research team do?

The research team looked at medical data from 187 patients with UCD. All went to the hospital for UCD within 28 days of birth. The team collected medical histories from a UCD database and also from patients referred by study partners.

To make sure the study looked at patients who were comparable, the team matched liver transplant and medically managed patients based on their

- Decade of birth
- UCD type
- Method of diagnosis
- Type of symptoms at diagnosis
- Age when symptoms started
- Age at first health problem due to ammonia, and highest ammonia level during that health problem

All patients with the most severe UCDs had liver transplants, and all patients with the least severe UCDs had medical management. The research team excluded these patients because they couldn't be matched up. The final data set included 109 patients.

To learn about how they decided which treatment to choose, the research team also interviewed parents and healthcare professionals.

Patient advocates, researchers, and healthcare professionals provided feedback throughout the study.

What were the limits of the study?

The low number of matched patients in the study limited how well the research team could see if the two groups differed.

Future studies could continue to look into how treatments for UCDs affect patients' health.

How can people use the results?

Patients and their doctors can use these results when considering treatment for UCDs.

To learn more about this project, visit www.pcori.org/AhMew325.