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Liver transplant may arrest neurological damage in rare, progressive form of autism

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Summary: A patient with a rare metabolic disease, lathosterolosis, that causes liver failure and autistic behavior experienced significant improvements in both her physical and mental health after receiving a liver transplant, according to a new case report. Lathosterolosis, a rare disease caused by a defect in cholesterol synthesis, is characterized by multiple congenital anomalies, mental retardation, and progressive liver disease.

FULL STORY

A patient with a rare metabolic disease that causes liver failure and autistic behavior experienced significant improvements in both her physical and mental health after receiving a liver transplant, according to a new case report published in the *American Journal of Transplantation*. The report's findings suggest an unexpected link between metabolic conditions and some forms of autism, and they point to the importance of a healthy liver for normal brain function.

Lathosterolosis, a rare disease caused by a defect in cholesterol synthesis, is characterized by multiple congenital anomalies, mental retardation, and progressive liver disease.

In the case report, Pier Luigi Calvo, MD, of the Cittá della Salute e della Scienza in Italy, and his colleagues describe the only patient with the disease who is alive today.

When she was diagnosed at 2 years of age, she exhibited autistic behavior, she was unable to walk unaided, and her sight was impaired by cataracts.

By 7 years of age she developed end-stage liver disease and received a liver transplant.

One year after the transplant, cholesterol synthesis was normal and she gradually began to walk independently and to interact with her caregivers.

After five years, mental deterioration stopped and the patient had returned to her everyday life, albeit with limitations.

The investigators hypothesize that, while established structural defects in the patient's brain could not be reversed, a degree of residual plasticity in her central nervous system may have benefited from the normalization of cholesterol levels that occurred after liver transplantation.

"We were dealing with a unique case -- literally, as the child is the only known surviving patient with the condition -- so it is difficult drawing inferences of broader significance," said Dr. Calvo. However, the findings suggest that timely liver transplantation might arrest the progression of neurological damage caused by diseases related to problems with cholesterol production. "We described this case on its own merits and also as a possible model for other, more common defects of cholesterol biosynthesis," said Dr. Calvo.

Story Source:

Materials provided by **Wiley**. *Note: Content may be edited for style and length.*

Journal Reference:

1. P. L. Calvo, A. Brunati, M. Spada, R. Romagnoli, G. Corso, G. Parenti, M. Rossi, M. Baldi, G. Carbonaro, E. David, A. Pucci, A. Amoroso, M. Salizzoni. **Liver Transplantation in Defects of Cholesterol Biosynthesis: The Case of Lathosterolosis**. *American Journal of Transplantation*, 2014; DOI: 10.1111/ajt.12645

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