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Commentary

Recommending Liver Transplant as an Exception for Chanarin-Dorfman Syndrome

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DESCRIPTION

Chanarin-Dorfman Syndrome (CDS) is a rare autosomal recessive genetic disorder that affects multiple systems in the body. It is characterized by the accumulation of neutral lipids, particularly triglycerides, in various organs including the liver, muscle, and skin. CDS is caused by mutations in the ABHD5 gene, which encodes a protein called α/β-hydrolase domain-containing protein 5 (ABHD5). ABHD5 is important in the breakdown of stored triglycerides into free fatty acids, which can then be used for energy production.

CDS was first described in 1975 by two physicians, Dr. Chanarin and Dr. Dorfman, who noticed a unique presentation of a patient with ichthyosis, hepatomegaly, and lipid droplets in peripheral blood cells. Since then, only around 100 cases have been reported worldwide, making it an exceptionally rare condition. CDS is more commonly found in populations with high levels of consanguinity, such as in the Middle East, North Africa, and South Asia.

The clinical manifestations of CDS are variable, and no two patients are exactly alike. The most common symptoms are dry, scaly skin (ichthyosis), hepatomegaly, and muscle weakness. Other symptoms that may be present include cataracts, short stature, developmental delay, intellectual disability, and seizures. The severity of the symptoms can vary widely, even among siblings with the same genetic mutation.

One of the most severe complications of CDS is liver disease. The accumulation of lipids in the liver can lead to cirrhosis, liver failure, and even Hepatocellular Carcinoma (HCC). Liver transplantation is the only effective treatment for end-stage liver disease in CDS patients. However, due to the rarity of the condition and the associated difficulties in diagnosis and management, there have been very few reported cases of liver transplantation in CDS patients.

In a recent case report published in the journal Liver Transplantation, doctors from the King Faisal Specialist Hospital and Research Centre in Riyadh, Saudi Arabia, describe the successful liver transplantation of a 7-year-old boy with CDS. The patient had been diagnosed with CDS at the age of 2, and had developed end-stage liver disease with cirrhosis and portal hypertension. The doctors had initially attempted to manage his liver disease with medication and a low-fat diet, but his condition continued to deteriorate, and he was eventually listed for liver transplantation.

The transplantation was performed using a liver from a deceased donor, and the patient's postoperative course was uneventful. He was discharged from the hospital on postoperative day 10, and has been doing well with no signs of rejection or recurrence of the disease. The doctors note that the successful transplantation in this patient provides evidence that liver transplantation can be a viable treatment option for end-stage liver disease in CDS patients.

Liver transplantation in CDS patients presents several unique challenges. One of the main concerns is the accumulation of lipids in the transplanted liver. This can lead to early graft dysfunction, and may increase the risk of post-transplant complications such as infection and rejection. To minimize the risk of these complications, it is important to carefully select donors with healthy livers, and to closely monitor lipid levels in the recipient both before and after transplantation.

Another concern is the potential for recurrence of the disease in the transplanted liver. While CDS is caused by a genetic mutation, it is not a primary liver disease, meaning that the disease can affect other organs in addition to the liver. Therefore, there is a risk that the disease could recur in the transplanted liver, even if the patient receives a liver from a healthy donor

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