Liver failure in newborn – mind galactosemia

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Background:

Classic galactosemia is a rare autosomal recessive disease caused by galactose-1-phosphate uridylyltransferase (GALT) deficiency. GALT converts galactose to glucose, if deficient, galactose-1-phosphate and other metabolites accumulate in the body. The main signs of GALT deficiency are liver failure, susceptibility to E. coli sepsis, and cataracts during the neonatal period. Untreated disease causes early death. Restriction of galactose prevents life-threatening complications. Still, the majority of patients experience long-term chronic complications, such as delayed speech, movement disorders, and ovarian insufficiency in women.

Case presentation:

A male newborn developed jaundice on the third day of life. Phototherapy was started and conducted for five days, with satisfactory results. On the ninth day, septic-like deterioration occurred. Laboratory findings revealed high inflammatory markers and signs of acute liver failure. Metabolic disease was suspected, therefore enteral nutrition was replaced with an intravenous infusion of dextrose. The patient received antibiotics, immunoglobulins, and blood derivatives (fresh frozen plasma and erythrocyte concentrate). High galactose in blood and urine pointed to galactosemia. After four days of intensive care treatment, the patient recovered and enteral nutrition with galactose-free formula was started. Dried blood spot testing showed GALT activity less than 2% of the control value. The diagnosis was confirmed by GALT gene testing, revealing two common mutations p.Q188R and p.K285N. The patient recovered fully and had no complications at the age of four.

Conclusion:

Although rare, galactosemia is one of the common causes of neonatal liver failure. It is critical to consider this disease in newborns with pathological hyperbilirubinemia, vomiting, signs of liver failure, and sepsis-like conditions. Timely diagnosis and treatment will prevent life-threatening complications and adverse outcomes.

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