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CASE REPORT

Hyperlipidemia Resulting in Abnormal Density and Signal Intensity of Blood in a Neonate with Lipoprotein Lipase Deficiency

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SUMMARY: We present the imaging findings in an 8-week-old infant with LPL deficiency. Due to markedly increased lipoproteins in the serum, abnormal hypodensity and abnormal T1-weighted hyperintensity were identified in the dural venous sinuses and medullary veins.

ABBREVIATIONS: Apo C-II = apolipoprotein C-II; FCS = familial chylomicronemia syndrome; LPL= lipoprotein lipase; SSS = superior sagittal sinus; VLDL = very-low-density lipoprotein

FCS results from either LPL or apo C-II deficiency.[1] Irrespective of whether the enzyme (LPL) or its cofactor (apo C-II) is deficient, the result is accumulation of lipoproteins in serum due to inadequate clearance. We report a rare case of LPL deficiency in an infant who had abnormal neuroimaging findings.

Case Report

An 8-week-old previously healthy term male infant was taken to an emergency department after the patient's mother noticed that the skin was pale and "odorous like an alcoholic" for 2 days. The patient had decreased oral intake, increased sleepiness, and a subjective fever, which the mother treated with acetaminophen. He appeared "gassy" and in pain before bowel movements. The urine and stool outputs were normal. There were no respiratory symptoms. The core body temperature was 38.8°C. The venous blood was noted to be pink with a milkshake consistency. Initially, the laboratory was unable to process the blood sample because of its unusual consistency. The patient was transferred to our institution and admitted to the intensive care unit for suspected sepsis and shock. The venous blood withdrawn at our institution was also pink.

Noncontrast head CT (Fig 1*A*, -*B*) showed diffuse hypoattenuation of the dural venous sinuses, internal cerebral veins, and the vein of Galen. The measured attenuation in the dural venous sinuses was approximately −15 HU. Linear hypodensities were present in the frontal white matter bilaterally.

MR imaging of the brain, performed a day later, showed abnormal lack of normal signal-intensity voids in the dural venous sinuses, particularly in the SSS (Fig 1*C*). There was markedly increased T1 signal intensity in the anterior aspect of the SSS. A fluid-fluid level was appreciated within the SSS near the vertex. Corresponding to the frontal linear hypodensities seen on CT, linear T1-weighted hyperintensities in the frontal lobes represented deep medullary veins (Fig 1*D*). The brain parenchyma was of normal appearance, except for subtle increased T2 signal intensity in the frontal white matter. There were no abnormal extra-axial fluid collections. Diffusion-weighted images showed no abnormalities.

The lipid panel was markedly abnormal. The concentrations of triglycerides (37,248 mg/dL; normal, 0–150 mg/dL), chylomicrons (36,836 mg/dL; normal, 0 mg/dL), and cholesterol (1056 mg/dL; normal, 0–200 mg/dL) confirmed the diagnosis of FCS. Diagnosis of LPL deficiency was subsequently made by quantification of LPL activity in the plasma after intravenous heparin injection. The patient was placed on a fat-free diet, and the level of triglycerides on discharge decreased to 1615 mg/dL 3 weeks later. The patient remained on a fat-free diet and showed a normal development. At 2 years of age, the serum triglyceride concentration was 905 mg/dL (normal, 30–86 mg/dL). There were no neurologic complications. No imaging follow-up was performed.

Discussion

Chylomicrons are responsible for transport of all dietary lipids into the circulation. Clearance of chylomicrons from the blood is rapid: The half-life of disappearance is <1 hour. LPL is located on the walls of the capillaries in many organs and tissues, including the heart, adipose tissue, spleen, lung, and renal medulla. LPL is not found in blood but is released after injection of heparin and can be quantified thereafter. Phospholipids and apo C-II are cofactors for LPL activity. LPL is the rate-limiting enzyme for hydrolysis and removal of triglycerides from chylomicrons and VLDLs.[2]

Patients with FCS may present with acute pancreatitis when the triglyceride level is >1000 mg/dL. The massive elevation of plasma triglycerides may be clinically silent. On physical examination, eruptive xanthomas may be seen. Two different genetic defects may result in FCS: LPL deficiency and apo C-II deficiency. Mutations in either the *LPL* gene or the *apo C-II* cause functional inability to hydrolyze chylomicrons and subsequent hyperchylomicronemia. Of these 2 genetic defects, LPL deficiency is more common, with approximately 1 in 1 million persons being affected.[1] In FCS, in addition to elevation of triglyceride levels, the cholesterol level is also increased because cholesterol is transported in chylomicrons.

Wilson et al[3] reported 2 siblings with FCS due to apo C-II deficiency. The older sibling presented with massively elevated triglyceride levels at 5 weeks of age. MR imaging showed multiple linear and globular foci of T1 shortening in the brain, corresponding to intravascular and extravascular fat. Despite dietary intervention and normalization of the abnormal lipid panel, this patient subsequently developed macrocystic en-

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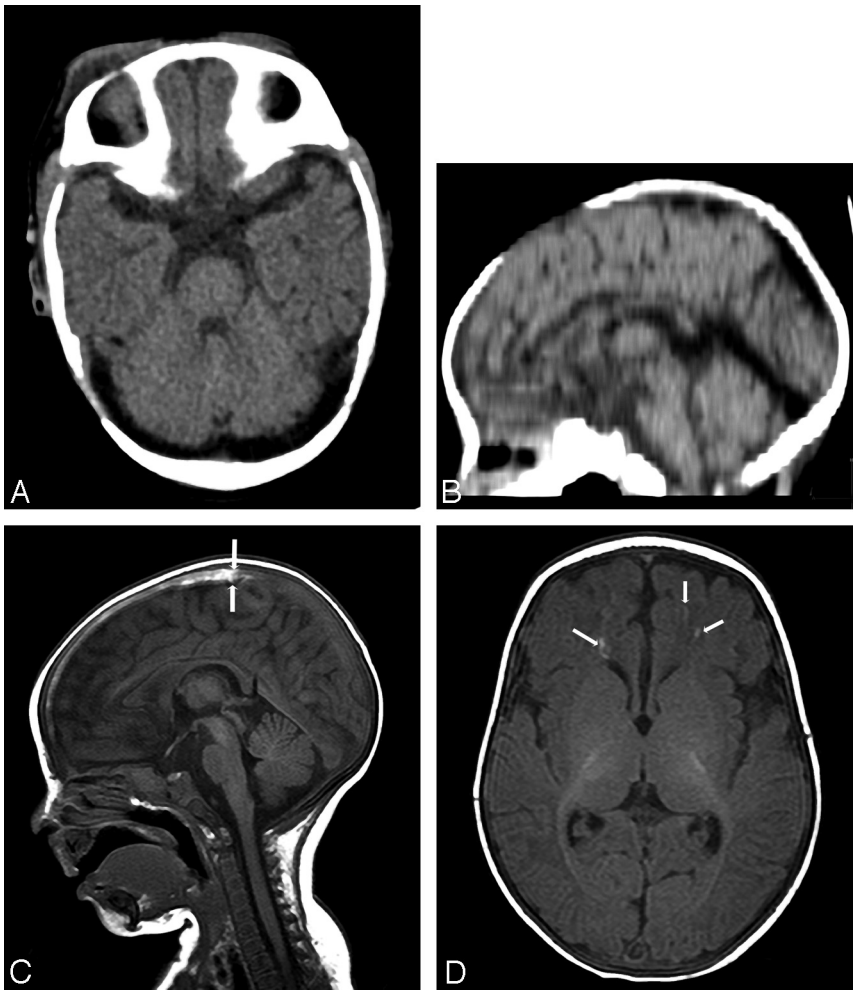

Fig 1. *A,* Axial noncontrast CT image shows decreased attenuation of the transverse sinuses. The density of the transverse sinuses is −15 HU. *B,* Reformatted midsagittal CT image demonstrates hypoattenuation in the superior sagittal sinus, internal cerebral veins, vein of Galen, straight sinus, and torcula. *C,* Sagittal T1-weighted (TR/TE, 500/12 ms; NEX, 4) image shows lack of normal signal void in the superior sagittal sinus. The anterior part of the superior sagittal sinus is hyperintense, and a fluid-fluid level (*arrows*) is present near the vertex. *D,* Axial T1-weighted (TR/TE, 500/12 ms; NEX, 4) image displays linear T1 shortening (*arrows*) in the anterior frontal white matter, corresponding to fat in the deep medullary veins.

cephalomalacia, cerebral atrophy, and subdural collections and became neurologically impaired. The younger sibling, reportedly, had less pronounced imaging findings; fat signal intensity was limited to the intravascular compartment, as was the case in our patient. This child had a normal neurologic outcome following dietary intervention.

CT findings in our patient were noteworthy because it is almost impossible to record negative Hounsfield units in the dural venous sinuses in the absence of marked lipemia. In fact, in the neonate and infant, the attenuation of dural venous sinuses is generally higher than it is in older children, due to hemoconcentration. In severe anemia, the dural venous sinus attenuation may decrease and approach that of CSF attenuation.

On MR imaging, lack of normal signal voids in the dural venous sinuses may raise the possibility of thrombosis. In the absence of a CT scan, this could potentially have caused diagnostic difficulty. In a dural venous sinus, observing a fluid-fluid level, particularly with a T1-weighted hyperintense nondependent component, is very unusual. We hypothesize that this T1 shortening of the anterior half of SSS and anterior deep medullary veins is due to the increased fat concentration in these vessels compared to the posterior venous vasculature. The relatively slow flow in the veins and dural venous sinuses may allow for this "fatty supernatant" due to the lesser specific gravity of fat. There is probably a threshold in plasma fat concentration to generate T1 shortening. Otherwise all the venous structures would have demonstrated increased T1 signal.

FCS or other hyperlipidemic conditions should be entertained when abnormal low attenuation is identified in the vasculature on CT. The presence of fat should be considered as a possibility in addition to venous thrombosis, when T1 shortening is present in the dural venous sinuses and cerebral veins.

References

1. Tershakovec AM, Rader DJ. **Disorders of lipoprotein metabolism and transport**. In: Behrman RE, Kliegman RM, Jenson HB, eds. *Nelson Textbook of Pediatrics*. Philadelphia: Saunders; 2004:445–59
2. Botham KM, Mayes PA. **Lipid transport and storage.** In: Murray RK, Granner DK, Rodwell VW, eds. *Harper's Illustrated Biochemistry*. New York: McGraw-Hill; 2006:217–29
3. Wilson CJ, Priore Oliva C, Maggi F, et al. **Apolipoprotein C-II deficiency presenting as a lipid encephalopathy in infancy.** *Ann Neurol* 2003;53:807–10