CASE REPORT

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Usefulness of per-rectal portal scintigraphy with Tc-99m pertechnetate for galactosemia in infants

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Galactosemia discovered by newborn screening is rarely caused by enzyme deficiency. It has recently been reported that among patients without enzyme deficiency portosystemic shunting may be a cause of galactosemia in some patients. We did per-rectal portal scintigraphy in patients with such galactosemia detected during screening of newborns to examine the usefulness of this method for the diagnosis of portosystemic shunts via the inferior mesenteric vein. The subjects were eight neonates with galactosemia without enzyme deficiency detected during screening. A solution containing technetium-99m pertechnetate was instilled into the rectum, and serial scintigrams were taken while radioactivity curves for the liver and heart were recorded sequentially. The per-rectal portal shunt index was determined by calculating the ratio for counts of the liver to counts for the heart integrated for 24 seconds immediately after the appearance of the liver time-activity curve. A portosystemic shunt was detected in both of the patients with a shunt index of 30% or more, but not in the six patients with a shunt index less than 30%. The blood galactose levels of these six patients later entered the reference range. This method is noninvasive and there is little exposure to the radionuclide. It seemed to be useful for the diagnosis of portosystemic shunt in newborns with galactosemia without enzyme deficiency.

Key words: galactosemia, portosystemic shunt, per-rectal portal scintigraphy