Teaching NeuroImages: Galactitol peak and fatal cerebral edema in classic galactosemia

Too much sugar in the brain

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Figure 1 Conventional MRI and diffusion-weighted imaging (DWI) with apparent diffusion coefficient (ADC) map

(A) Sagittal and (B) axial turbo-spin-echo T2-weighted images, (C) DWI, and (D) ADC. (A) Massive edema causes cerebellar tonsils to descend into the foramen magnum. (B) Brain and cerebellum appear diffusely swollen with reduced gray/white matter differentiation. (C) DWI and (D) ADC maps show large posterior areas of restricted diffusion consistent with cytotoxic edema.

A newborn girl with genetically confirmed galactosemia (compound heterozygous for p.ARG51Trp [c.151C>T] and p.GLN188ARG [c.563A>G] mutations in galactose-1-phosphate uridylyltransferase [*GALT*] gene) presented with jaundice, hypoglycemia, and liver failure. A galactose-free diet was started but she became comatose. Brain MRI with spectroscopy showed diffuse edema (figure 1, A–D) and galactitol peak (figure 2). Despite cerebral edema therapy, she died at 21 days of cerebral herniation.

Galactosemia due to *GALT* deficiency presents with hepatopathy, cataracts, and occasionally intracranial hypertension, generally resolving with a galactose-free diet. Fatal cerebral edema is exceptional¹ and might be related to galactitol accumulation, an osmotic metabolite impermeable to membranes, synthesized by galactose through alternative pathways.²

AUTHOR CONTRIBUTIONS

Dr. Martinelli designed the study and contributed to interpretation of the data and to the drafting of the manuscript. Dr. Bernardi, Dr. Napolitano,

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Figure 2 Magnetic resonance (MR) spectroscopy

Single voxel short echo time (TE 30 ms) localized into right parietal white matter. LC-Model fitted MR spectrum shows presence of both galactitol and lactate. The in vivo concentrations of galactitol (prominent peak at 3.7 ppm) and lactate obtained from MR spectroscopy were 21.3 ± 1.0 and 6.4 ± 0.6 mmol/kg, respectively.

and Dr. Colafati contributed to acquisition and interpretation of the data and revised the manuscript. Dr. Dionisi-Vici supervised the study, contributed to interpretation of the data, and revised the manuscript.

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DISCLOSURE

The authors report no disclosures relevant to the manuscript. Go to Neurology.org for full disclosures.

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