Table 1. Clinical and MRI findings in patients affected by galactosemia.

Pt.	Sex	Diagnosis	Age at MRI	Clinical presentation at time of MRI.	Clinical phase at presentation	MRI findings
1	M	Heterozygosity for GALT gene mutations (c.563A>G and c.684G>T).	7 months	Macrocephaly, hypotonia, failure to thrive, hepato-splenomegaly, cataracts, renal tubulopathy.	SUBACUTE	Diffuse bilateral supratentorial edema involving WM and deep GM (striatus and Thalamus), "double cap sign", thinning of corpus callosum, and cyst-like lesion in temporal poles. MRS: galactitol doublet at 3.6-3.74.
2	M	Heterozygosity for GALT gene mutations (c.563A>G and c.684G>T).	7 months	Macrocephaly, hypotonia, failure to thrive, hepato-splenomegaly, cataracts, renal tubulopathy.	SUBACUTE	Diffuse supratentorial edema involving WM and deep GM (striatus and Thalamus), "double cap sign", thinning of corpus callosum, and cyst-like lesion in temporal poles. MRS: galactitol doublet at 3.6-3.74.
3	M	Heterozygosity for GALT gene mutation (c.563A>G).	13 days	Lethargy, poor feeding, hypotonia, jaundice and acute liver failure.	ACUTE	Diffuse WM edema and areas of diffusion restriction in the temporo-occipital cortex and thalami. MRS: galactitol doublet at 3.6-3.74.
4	F	Reduced GALT activity on erythrocytes.	6 years	Focal seizures, vomiting, hypotonia and reduced level of consciousness.	CHRONIC	Patchy areas of WM abnormality in the subcortical and periventricular regions. Thinning of carpus callosum. Follow-up at 13 years of age shows the persistence of peritrigonal and subcortical WM abnormalities associated with ventricular enlargement and mild cerebellar atrophy.
5	M	Homozygosity for GALT gene mutation (c.563A>G).	7 years	Status dystonicus, developmental delay with failure to thrive and grow.	CHRONIC	T2 hyperintensity in globus pallidi.
6	M	Reduced GALT activity on erythrocytes.	14 years	Autism spectrum disorder and mild cognitive impairment with dyslexia, dyscalculia, dyspraxia. Mild cataracts	CHRONIC	No abnormalities.
7	M	Homozygosity for GALT gene mutation (c.563A>G).	17 days	Prematurity, lethargy, hypotonia, metabolic acidosis, hyperammonemia and sepsis.	ACUTE	Diffuse supratentorial brain edema with diffuse WM abnormality and diffusion restriction. Follow-up at 5 years of age demonstrates the presence of encephalomalacic changes in the parietal lobes with ventricular dilatation and corpus callosum thinning.
8	M	Homozygosity for GALT gene mutation (c.563A>G).	2.11 years	Mild developmental delay.	CHRONIC	Delayed myelination. Follow-up at 7 years of age shows persistent delayed myelination.

9	M	Homozygosity for GALT gene mutation (c.563A>G).	6 years	Mild mental retardation.	CHRONIC	Delayed myelination and thinning of the corpus callosum.
10	M	Homozygosity for GALK gene mutation (c.82C>A).	25 days	Lethargy, hypotonia, metabolic acidosis, jaundice, vomiting.	ACUTE	Diffuse supratentorial brain edema. MRS: galactitol doublet at 3.6-3.74.
11	M	Reduced GALT activity on erythrocytes.	5 years	Mild mental retardation more pronounced for language aspects.	CHRONIC	Delayed myelination, thinning of the corpus callosum and focal malacic changed in the right cerebellar hemisphere. MRS: normal.
12	M	Reduced GALT activity on erythrocytes.	5 years	Psychomotor developmental delay, cataracts, hepato-splenomegaly, seizures.	CHRONIC	Very small lesion in the left caudothalamic notch.
13	M	Reduced GALT activity on erythrocytes.	14 years	Psychomotor developmental delay, cataracts, hepato-splenomegaly.	CHRONIC	Patchy areas of WM abnormality in the subcortical and periventricular regions. Thinning of carpus callosum. Cerebellar atrophy.
14	F	Reduced GALT activity on erythrocytes.	10 years	Psychomotor developmental delay, cataracts, hepato-splenomegaly, renal tubulopathy.	CHRONIC	Delayed myelination in the supratentorial white matter, thinning of the corpus callosum, supratentorial and cerebellar atrophy with ventricular enlargement.
15	M	Reduced GALT activity on erythrocytes.	8 months	Psychomotor developmental delay, cataracts, hepato-splenomegaly.	SUBACUTE	Diffuse supratentorial white matter alteration, “double cap sign” and thinning of corpus callosum, cyst-like lesion in temporal poles. MRS: galactitol doublet at 3.6-3.74. Follow-up at 2 years of age demonstrates patchy areas of periventricular supratentorial white matter abnormalities and thinning of corpus callosum. MRS: normal.
16	M	Reduced GALT activity on erythrocytes.	8 months	Psychomotor developmental delay, cataracts, hepato-splenomegaly.	SUBACUTE	Diffuse supratentorial white matter alteration, “double cap sign” and thinning of corpus callosum, cyst-like lesion in temporal poles. MRS: galactitol doublet at 3.6-3.74.
17	M	Reduced GALT activity on erythrocytes.	18 months	Psychomotor developmental delay, seizures, hepato-splenomegaly.	SUBACUTE	Diffuse supratentorial white matter alteration, “double cap sign”, thinning of corpus callosum and cyst-like lesion in temporal poles.

WM: white matter; GM: grey matter; GALT: galactose-1-Phosphate uridyltransferase; GALK: galactokinase; MRS: magnetic resonance spectroscopy