

On-line Table: Summary of clinical and neuroradiologic findings in 10 patients with HCC

Patient/Sex	1/M	2/M	3/M	4/F	5/M	6/M	7/F	8/M	9/M	10/M
Clinical findings										
Congenital cataract	+	+	+	+	+	+	+	+	+	+
Acquisition of supported walking	24 m	18 m	12 m	14 m	12 m	24 m	14 m	13 m	14 m	24 m
Loss of supported walking	NE	8 y	8 y	NA	9 y	9 y	8 y	NE	NE	-
Pyramidal signs	+	+	+	+	+	+	+	+	+	+
Cerebellar signs	+	+	+	+	+	+	+	+	+	-
Mental retardation	Mild	Mild	Moderate	Mild	Moderate	Mild	Moderate	Mild	Mild	Mild
Seizures	+	-	-	-	+	-	-	-	+	-
Neuroradiologic findings										
Age at MRI	2 y	3 y	26 y	6 y	NT	7 y	8 y	20 m	20 m	16 y
Age at CT	NT	3 y	NT	NT	23 y	7 y	NT	NT	NT	NT
Supratentorial WM Hypomyelination	Diffuse	Diffuse	Diffuse	Diffuse	Not evaluable	Diffuse	Diffuse	Partly spared subcortical WM	Partly spared subcortical WM	Diffuse
Other WM findings	Increased water content; deep periventricular WM; A-P gradient	Increased water content (liquefaction); deep periventricular WM; A-P gradient	Diffuse WM atrophy/glios	Localized areas of increased water content; deep frontal lobes	WM atrophy; hypodense (gliotic) periventricular WM	None	Increased water content (liquefaction); frontoparietal, posterior, periventricular WM; A-P gradient	None	None	Diffuse WM atrophy/glios
Corpus callosum	Thin splenium	Thin	Thin	Thin	NE	Thin	Thin	Normal	Normal	Thin
Cerebellum	Hypomyelinated white core; normal medullary laminae	Hypomyelinated white core; normal medullary laminae	Cortical atrophy; hypomyelinated white core; normal medullary laminae	Hypomyelinated white core; normal medullary laminae	NE	Hypomyelinated white core; normal medullary laminae	Hypomyelinated white core; normal medullary laminae	Normal	Normal	Normal
Brain stem	Normal	Hyperintense corticospinal tracts	Pons hypoplasia; hyperintense transverse fibers	Pons hypoplasia	Pons hypoplasia	Pons hypoplasia; hyperintense corticospinal tracts	Pons hypoplasia	Hyperintense substantia nigra	Normal	Pons hypoplasia
Age at follow-up MRI	NT	8 and 16 y	NT	NT	NT	NT	15 y	NT	NT	17 y
Changes on follow-up MRI	NA	Supratentorial WM atrophy	NA	NA	NA	NA	Supratentorial WM atrophy	NA	NA	Unchanged
MRS findings (age)	NT	↓ Cho/Cr (16 y)	NT	NA	NT	NT	↓ Cho/Cr (15 y)	↑ mins/Cr, ↑ Cho/Cr, ↑ lipids (20 m)	↑ mins/Cr, ↑ Cho/Cr, ↑ lipids (20 m)	↑ mins, ↑ Cr, ↓ NAA (17 y)

Note:—+ indicates present; -, absent; y, years; m, months; WM, white matter; NT, not tested; NE, not evaluable; MRI, magnetic resonance imaging; MRS, MR spectroscopy; A-P, anteroposterior; NA, not available; Cho, choline; Cr, creatine; mins, myo-inositol; NAA, N-acetylaspartate; ↑, increased; ↓, decreased.