SUPPLEMENTARY APPENDIX

The clinical and laboratory evaluation of familial hemophagocytic lymphohistiocytosis and the importance of hepatic and spinal cord involvement: a single center experience

Burcin Beken,[1] Selin Aytac,[2] Gunay Balta,[2] Baris Kuskonmaz,[2] Duygu Uckan,[2] Sule Unal,[2] Mualla Cetin[2] and Fatma Gumruk[2]

[1]Hacettepe University Department of Pediatrics, and [2]Hacettepe University Department of Pediatric Hematology, Ankara, Turkey

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Correspondence: burcinbeken@gmail.com

Online Supplementary Table 1S. Characteristics of patients with spinal cord involvement.

Patient	Age of diagnosis (year/ gender)	Mutation	HF in bone marrow at diagnosis	Neurological findings at diagnosis	CSF findings at diagnosis			Cranial MRI findings at diagnosis	Treatment protocol	Cranial MRI findings after treatment	Relapse	Cranial MRI findins during relapse	Spinal MRI findings during relapse	HSCT	Prognosis
					Protein (mg/dl)	Cell	HF								
1	12/M	PRF1	-	Headache, delirium	86	+	-	Bilateral patchy lesions in infra- and supratentorial compartments. Cerebellitis-like lesions. Demyelinating lesions in brainstem, hemorrhagic lesions in deep white matter.	HLH-2004	Regression of hemorrhagic lesions.	+ (BM+CNS)	Progression in potine and supratentorial lesions in terms of size and contrast uptake.	Two different focuses with T2 signal increase in spinal cord C7-T9 vertebral levels. Findings compatible with arachnoiditis in cauda equina fibers.	+	Exitus
2	14/F	UNC13D	-	Seizure	50	+	-	Contrast-holding nodules in brainstem, right frontal, posterior parietal and left oxipital regions.	HLH-2004	Minimal regression of right frontal region lesions.	+ (BM+CNS)	Punctat-curvilineer stainings consistent with perivascular pattern, located in brainstem, thalamus and bilateral cerebellar hemishphere. Three parenchimal lesions with hemorrhagic component in right frontal and bilateral oxipital regions.	Lesions with intramedular contrast uptake in cervicomedullar junction, cervical and toracal spinal cord.	+	Exitus
3	9.5/M	UNC13D	+	Intracranial hypertension	32	+	-	Ring shaped and lineer micronodular contrast uptake in right cerebellar hemisphere medial region. T2 hyperintense signal changes in bilateral periventricular deep white matter, centrum semiovale and corona radiata.	HLH-2004	-	+ (BM+CNS)	Progression of the lesions in cerebellum, brainstem, cerebellar white matter and cortex. Obliteration of basal cisterns and foramen magnum due to edema of cerebellum and brainstem. Narrowing of the 4th ventricle.	Diffuse, active inflamation-infiltration findings in brainstem, cerebellum and entire spinal cord up to the T6 vertebral level.	+	Exitus
4	7.5/F	STX11	+	Left hemiparesis	77	+	+	Widespread,probable inflamatuar lesions in bilateral cerebellar and cerebral white matter. Right temporoparietooxipital cytotoxic edema. Focal T2 signal changes in cervical spinal cord. Widespread contrast uptake in cervical spinal, infra- and supratentorial pial regions.	HLH-2004	Total regression of brainstem, cerebellum and periventricular white matter lesions. Contrast uptake in bilateral 3rd and 5th cranial nerves.	+ (BM+CNS)	Lesions with T2 signal increase in lesions of right cerebral hemisphere white matter extending vermis and cerebellum.	Increased pial contrast uptake aroud cervical spinal cord. T2 hyperintense lesion without contrast uptake in posterior of medulla oblangata. Tonsiller herniation.	+	Exitus
5	11/F	Not detected	+	Seizure, cranial nevre palsy	102	+	-	Lesions in brainstem, bilateral cerebellar hemisphere and periventricular white matter.	HLH-2004	-	+ (BM+CNS)	Severe inflamation and edema in cerebellar parenchima and brainstem. Progression of lesions in deep serebral white matter, corticomedullar junction and bilateral internal capsule posterior cruses.	Focal T2 signal increases in entire spinal cord. Contrast-holding lesions in middle and lower toracal spinal segments. Diffuse contrast uptake in cauda equina.	--	Exitus

BM; bone marrow, CNS; central nervous system, CSF; cerebrospinal fluid, HF; hemophagocytosis, HLH; hemophagocytic lymphohistiocytosis, HSCT; hematopetic stem cell transplantation, MRI; magnetic resonans imaging, PRF; perforin, STX; syntaxin, UNC13D; coding gene of Munch 13-4 protein.