History
9 month old patient with a history of VP shunt placement for hydrocephalus

Diagnosis
Macrocephaly capillary malformation aka Macrocephaly cutis marmorata telangiectatic congenita

Additional Clinical
Initial MRI at one month of age was obtained with no evidence of cerebellar herniation. The patient then acquired cerebellar herniation on their second MRI with secondary need for a shunt placement. Images from the third MRI show numerous findings reported with the above syndrome as well as worsening CTH.

Discussion

Various neuroimaging findings are nicely delineated in the reference article. Findings include: Ventriculomegaly with or without obstruction, Cavum septum pellucidum, Migrational anomalies, cerebellar tonsillar herniation (acquired), brain asymmetry, white matter abnormalities (usually only seen after 6 months of age), large head circumference, enlarged dural venous sinuses, large corpus callosum, large optic nerves or optic nerve sheaths.

Patients who did have spine MRI in the below study did not have evidence of tethering or syrinx. The cause of cerebellar tonsillar herniation is presumed to be cerebellar overgrowth with a possible secondary contribution from abnormal venous sinuses. Patients with the syndrome require follow up imaging as CTH is likely to worsen over time and can be surprisingly rapid.

In a patient with this syndrome white matter abnormalities should not be pursued as evidence of metabolic disease unless clinical symptoms develop that are atypical of the syndrome.

Findings
Extensive white matter signal abnormality
Cavum septum pellucidum
Acquired cerebellar herniation with altered brainstem morphology/compression and fourth ventricular narrowing (acquired due to rapid brain growth)
Polymicrogyria
Prominent corpus callosum
Large optic nerves

Reference