Occult leptomeningeal large cell medulloblastoma in an adult.

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OBJECTIVE AND IMPORTANCE: Large cell medulloblastoma is an uncommon malignancy of childhood that often pursues an aggressive clinical course. We report the first case of this entity in an adult that proved to be an unsuspected primary leptomeningeal tumor.

CLINICAL PRESENTATION: A 30-year-old man complained of worsening neck pain over the course of 3 months. Neck pain increased a few days prior to admission and a cervical spine CT revealed tonsillar herniation. Cervical spine MRI performed the day prior to admission confirmed the diagnosis of Chiari I malformation and C3-4 disk herniation without spinal cord compression. On the day of admission, the patient became unresponsive and resuscitative measures were unsuccessful. Postmortem examination of the brain was notable for necrotic cerebellar tonsils, but demonstrated no evidence of an intraparenchymal mass lesion. Microscopic examination of the cerebellum revealed discohesive neoplastic cells, which showed characteristic dot-like immunoreactivity for synaptophysin, diagnostic of large cell medulloblastoma within the subarachnoid space.

CONCLUSIONS: Our experience with this unique case illustrates the challenges of diagnosing a primary leptomeningeal neoplasm. This case also underscores the importance of maintaining a high degree of suspicion for leptomeningeal neoplasms in patients who present with imaging studies suspicious for Chiari I malformation.

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