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Pathology

Medullomyoblastoma: A Case Report and Literature Review of a very Rare Variant of Medulloblastoma

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Abstract **Case Report**

Medullomyoblastoma is an extremely rare tumor, corresponding to a variant of medulloblastoma with rhabdomyoblastic differentiation. Both tumor populations are sharing the same genetic alterations. We report a case of a 10 year-old girl, admitted for symptoms of intracranial hypertension associated with a cerebellar syndrome. Magnet resonance imaging of the brain was performed, and revealed a lesion of the cerebellar vermis. A wide excision was performed and the histopathological examination objectified a densely round cells proliferation coexisting with nests of rhabdomyoblastic cells that express desmin and myogenin in immunohistochemistry, making the diagnosis of medullomyoblastoma.

Keywords: Medulloblastoma; Medullomyoblastoma; Cerebellum; Immunohistochemistry.

INTRODUCTION

Medulloblastoma is a primary neuroectodermal cerebellar embryonic tumor, which can classically include aspects of lymph node, glial or melanocyte neuronal differentiation. We report a very rare case of medulloblastoma with muscle differentiation, called medullomyoblastoma.

OBSERVATION

A 10-year-old girl, without a history, admitted for symptoms of intracranial hypertension that has been evolving for a month associated with a discreet cerebellar syndrome.

Brain MRI had revealed a large process of the vermian posterior brain fossa, in T2 hypersignal, T1 hyposignal, increasing after gadolinium injection (Fig 1).

The patient benefited from a wide tumor removal leaving in place a small part that infiltrates the lower corner of the floor of the 4th ventricle.

Anatomopathological examination of the lesion showed a densely cellular proliferation, consisting of sheets of small roughly round cells (Fig 2), with a high nucleocytoplasmic ratio, with a rounded hyperchromatic nucleus, without visible nucleolus, giving rise to many mitoses. Rare rosettes were observed (Fig 3).

To this classic aspect of medulloblastoma was added a fairly abundant component of large cells (Fig 4), sometimes elongated, showing an eosinophilic cytoplasm, with a rounded or elongated nucleus, with a discreetly thickened nuclear membrane, slightly densified chromatin and a prominent nucleolus (Fig 5). This cell population showed in the immunohistochemical study an expression of desmine (Fig 6) and myogenin (Fig 7). The diagnosis of medulloblastoma with myogenic differentiation is made.

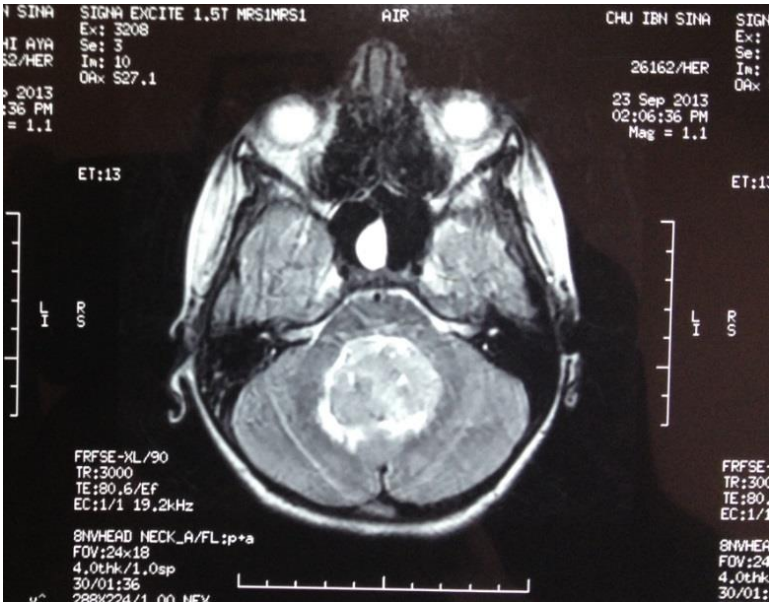

Figure 1: Cerebral MRI showing a large vermian process increasing after gadolinium injection

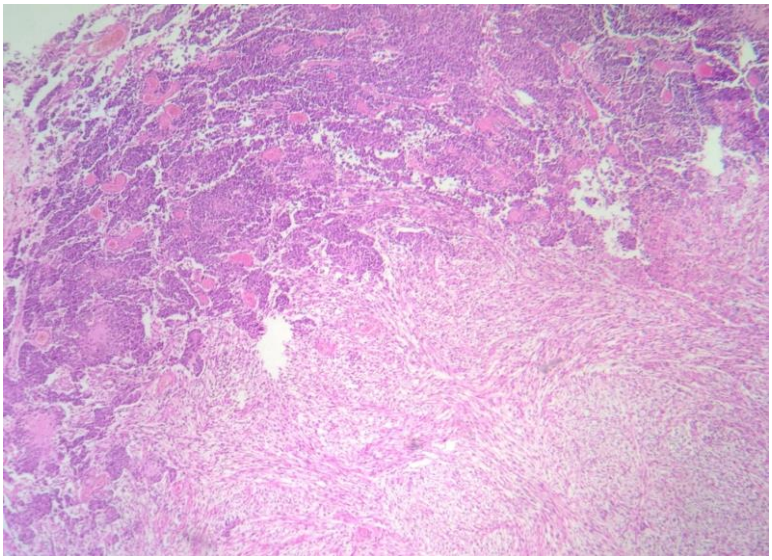

Figure 2: Double-component tumor proliferation (HE, x50)

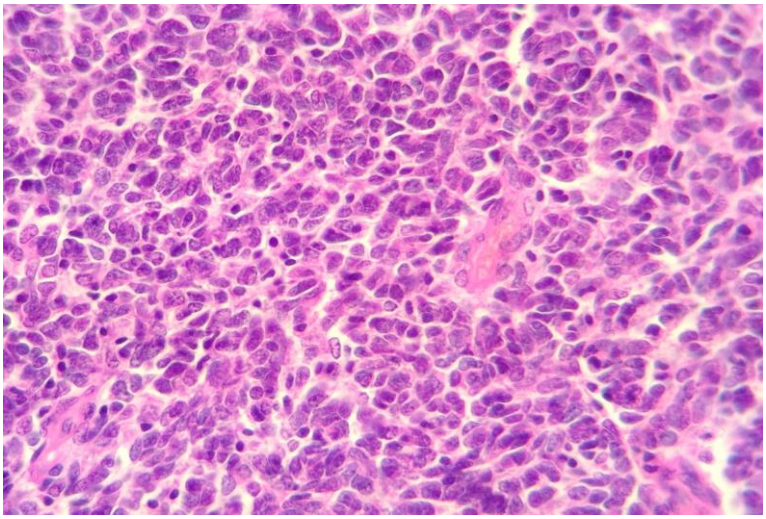

Figure 3: The first component is densely cellular, consisting of sheets of small round cells, sometimes arranged in rosettes, corresponding to a classic medulloblastoma (HE, x400)

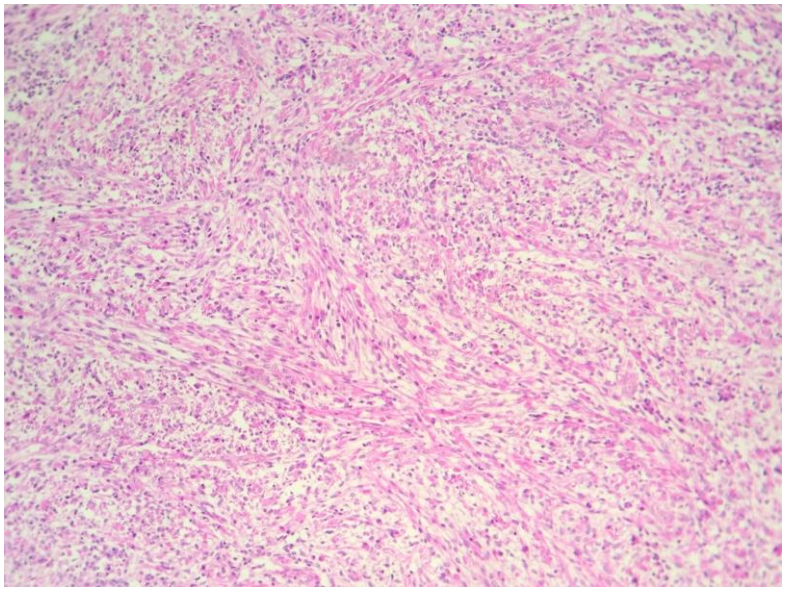

Figure 4: The second component, rhabdomyoblastic, is fusocellular in appearance (HE, x200)

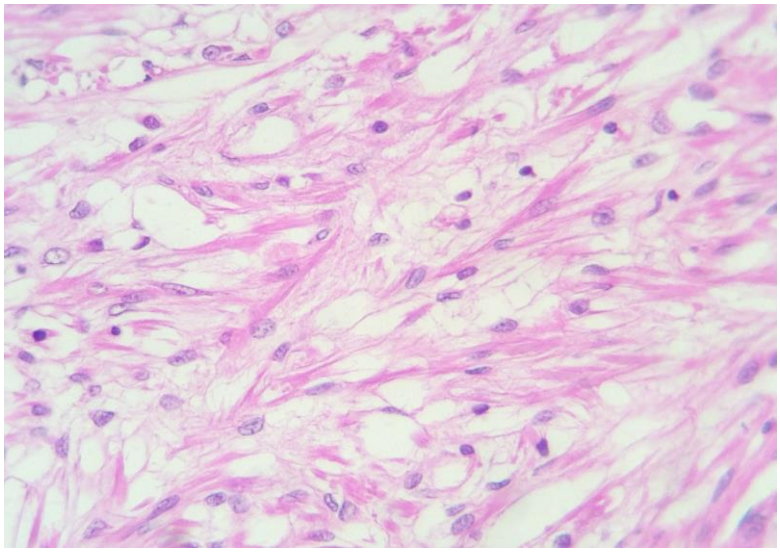

Figure 5: Rhabdomyoblastic cells are elongated, showing a striated eosinophilic cytoplasm, with a rounded or elongated nucleus, sometimes nucleolated (HE, x400)

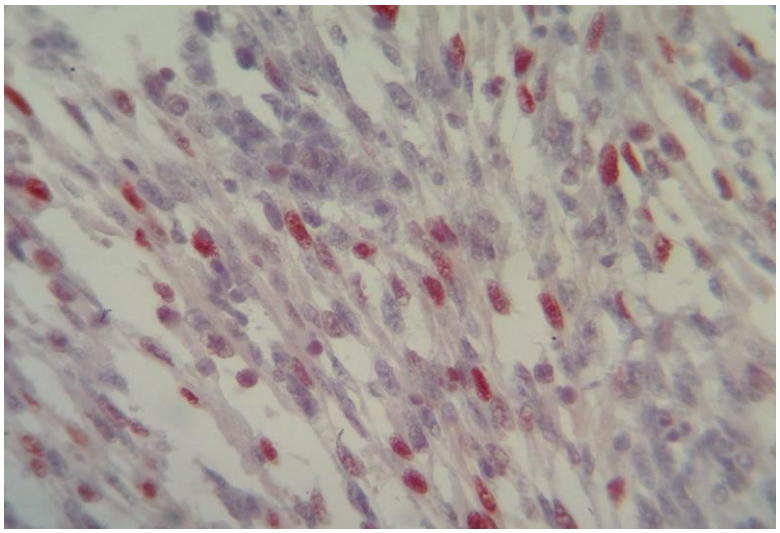

Figure 6: Expression of desmine by the rhabdomyoblastic component

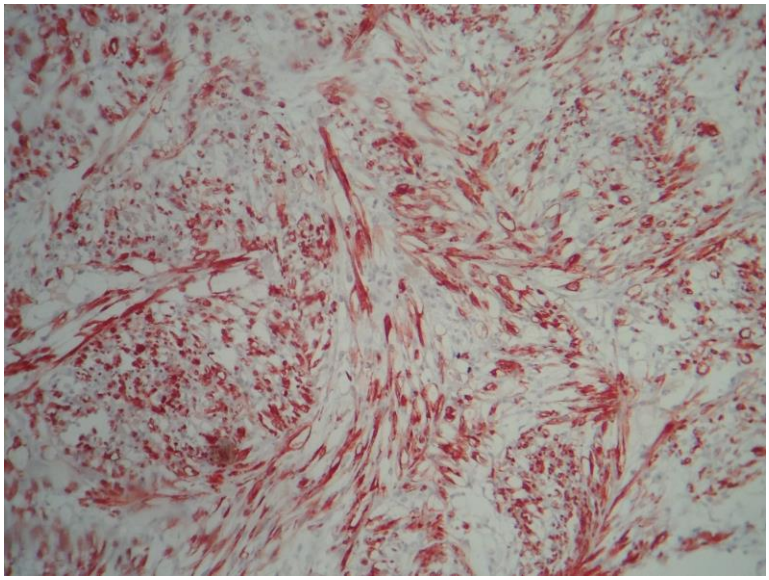

Figure 7: Expression of myogenin by the rhabdomyoblastic component

DISCUSSION

Although medulloblastoma is the most common embryonic tumor of neuroepithelial tissue, medullomyoblastoma (MMB), a variant of medulloblastoma with rhabdomyoblastic elements, is an extremely rare clinical-pathological entity, of which only about forty cases have been reported so far in the literature.

The MMB was first described by Marinesco and Goldstein in 1933 [1-3].

The average age at diagnosis is 4.5 years, with 90% of cases occurring before the age of ten [3]. The male/female sex ratio is changing from 2 to 4.5 [3, 4].

Clinically, medullomyoblastoma is usually revealed by cerebellar syndrome or intracranial hypertension syndrome [3]. The prognosis would be overall pejorative with an average survival of about two years [3].

The radiological aspect is variable and not very specific. Cerebral CT shows a lesion of the cerebellar vermis, most often very limited and spontaneously hyperdense, rarely of cystic appearance, sometimes with hypodensal necrotic changes, increasing after injection of contrast medium. The MRI signal is variable, hypo- or iso-intense on T1 weighted sequences, hypo- or hyperintense in T2 [3, 5-7].

Macroscopically, medullomyoblastoma appears as a soft, brittle, reddish or greyish-white tumor, sometimes lobulate in appearance [7-9].

Histopathological examination makes it possible to focus the diagnosis of medullomyoblastoma on the detection of an aspect of classical medulloblastoma or one of its large cell/anaplastic or nodular/desmoplastic variants, associated with a contingent of rhabdomyoblastic differentiation cells of varying degrees, dispersed or grouped into nodules. These cells are either fusiform, with an eosinophilic cytoplasm sometimes with clearly visible transverse striations or, more rounded in shape, with an abundant eosinophilic cytoplasm, with sometimes multiple nuclei, containing an eosinophilic nucleolus [3, 7-9].

Immunohistochemically, the neuroblastic component typically expresses NSE (neuron specific enolase) and synaptophysine, in varying ways neurofilament, chromogranin and GFAP. Rhabdomyoblastic cells variably express desmine, myoglobin, myogenin, smooth muscle actin, myosin, NSE and vimentin [6, 7, 9, 10].

Molecularly, MMB is characterized by a gain of chromosome 17q (isochromosome 17q), monosomy 17 or amplification of the c-myc gene [2,3].

These genetic abnormalities are found in both populations, neuroblastic and rhabdomyoblastic, suggesting that they derive from the same tumor clone.

The main differential diagnosis to evoke is the atypical rhabdoid and teratoid tumor, negative for myogenin and whose prognosis is much worse [10, 11].

Other differential diagnoses are immature teratoma and medullaepithelioma, which can rarely contain cartilaginous, bone or striated muscle heterologous contingents, associating with characteristic neuroepithelial structures [12].

Radical surgical treatment presents difficulties because medullomyoblastoma is often difficult to dissect from cerebellar hemispheres as is the case for our patient.

Medulllomyoblastomas are rapidly progressive tumors, local recurrence and metastases are common [13, 14].

Treatment with radiation therapy and platinum salt chemotherapy after radical surgery seems to improve the prognosis [3, 14, 15].

CONCLUSION

In conclusion, medullomyoblastoma is an extremely rare variant of medulloblastoma, which has cellular elements with rhabdomyoblastic differentiation. Both tumor populations would derive from the same clone. It should be distinguished mainly from atypical rhabdoid and teratoid tumor, immature teratoma and medullaepithelioma, the immunohistochemical study that can correct the diagnosis in difficult cases. His prognosis remains very poor. Treatment is based on surgery coupled with radiochemotherapy.

Conflicts of Interest: The authors do not declare any conflict of interest.

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