CASE REPORT

Pleomorphic rhabdomyosarcoma of the cerebellopontine angle in an adult: a review of literature

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SUMMARY

Rhabdomyosarcoma (RMS) is a rare and aggressive neoplasm characterised by rapid growth and metastatic invasion. The most frequent localisation is the skeletal musculature of the limbs. The head and the neck are rarely involved. A 50-year-old woman presented to our attention because of a progressively increasing headache, ataxia and vomiting. MRI showed a lesion at the right cerebellopontine angle. Thereafter, the patient was submitted to a piece-meal removal of the neoplasm. Despite the postoperative MRI showed no signs of remnant, 7 months after the surgery, the disease recurred with multiple localisations, and the patient died a few days later. This report is the first description in the literature of a pleomorphic RMS of the cerebellopontine angle. This particular tumour carries a bad prognosis because of the vicinity of nervous structures and of the impossibility of achieving a one-piece resection. More than ever, the adjunctive treatments had to be effective against a potential remnant and in controlling recurrences.

our knowledge, this is the first such case described in English-written literature.

CASE PRESENTATION

A 50-year-old woman presented to the emergency department because of a progressively increasing occipital headache, walking disturbances and vomiting. A few days before admission, a tinnitus and a progressive hearing loss appeared in the right ear. She had two younger brothers with no remarkable medical history. She denied consuming alcohol and drugs and she was not a smoker. She had no previous surgical interventions.

INVESTIGATIONS

Physical and neurological examinations confirmed a right ataxia, dysmetria and hearing loss. Facial motility was conserved. The Romberg test was positive. An audiometric test was administered, which confirmed a mild hearing loss to the right ear with particular reduction on high frequencies. Vestibular tests were positive. The patient underwent MRI, which showed a lesion at the right cerebellopontine angle. On T1-weighted images, the lesion presented isointense to the brain parenchyma and heterogeneous due to the presence of multiple cysts with signal intensity equal to the cerebrospinal fluid, clearly visible on T2-weighted images (figure 1B). Contrast enhancement was heterogeneous (figure 1A). The maximum diameter was 33 mm. The lesion had grown inside the internal acoustic meatus; the extra-meatal component strongly compressed the pons and the middle cerebellar peduncle, which showed signs of oedema.

TREATMENT

A suboccipital retrosigmoid craniotomy was performed with the aid of intraoperative neurophysiological monitoring. After gentle retraction of the right cerebellar hemisphere, a capsulated lesion was uncovered, which had partially infiltrated the middle cerebellar peduncle. Strong haemorrhages made the debulking manoeuvres laborious. The mass was solid with some liquid portion corresponding to the multiple intralesional cysts. The facial nerve was dislocated on the anteroinferior side of the tumour and was anatomically conserved. Its functionality was monitored with intraoperative stimulation and electromyography registration (orbicularis oris and oculi). A tissue sample was taken and sent for histopathological examination. A posterior meatotomy was necessary to uncover

BACKGROUND

Rhabdomyosarcoma (RMS) is a rare and aggressive neoplasm characterised by rapid growth and metastatic invasion.¹ It was first described by Weber in 1854, but Stout provided clear diagnostic criteria in 1946.² RMS is a small cell tumour that shares histological and behavioural characteristics with central and peripheral primitive neuroectodermal tumours; esthesioblastomas; desmoplastic tumours; neuroendocrine or poorly differentiated cancers of the breast, lung or gut and extraskeletal Ewing sarcomas.³ RMS might present either as a metastatic disease without any primary location⁴ or as a solid mass.⁵ The histological classification includes embryonal, alveolar, botryoid, pleomorphic and diffuse types. Malignant fibrous histiocytoma, pleomorphic leiomyosarcoma, pleomorphic liposarcoma, malignant mesenchymoma and other types of RMS are included in the major differential diagnosis.⁶ Moreover, in the cerebellopontine angle, the teratoid rhabdoid tumour could be considered in differential diagnosis of adult age. The most frequent site of origin is the skeletal musculature of the limbs, particularly the thigh. The head and the neck are rarely involved.^{5 6}

In the present report, the authors describe a unique case of an adult harbouring a pleomorphic RMS of the cerebellopontine angle. To the best of

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Figure 1 Brain MRI showing a cerebellopontine mass which fills the cistern and compresses the pons, the middle cerebellar peduncle and the cerebellar parenchyma. The lesion presents cystic degeneration (B), and dishomogeneous enhancement after contrast administration (A). Postoperative control showing no signs of the tumour, which has been macroscopically removed (C).

and remove the intrameatal portion of the neoplasm, which was easily separated from the acoustic, facial and vestibular nerves.

OUTCOME AND FOLLOW-UP

The early postoperative period was uneventful. The vestibular symptoms disappeared and the ataxia improved. A cerebral MRI showed no signs of lesion in the T1-weighted images with contrast administration (figure 1C). Ten days after surgery, the patient was discharged with a planned rehabilitation programme. Radiotherapy was administered with a dose of 30 Gy. The chemotherapy regimen consisting of high-dose ifosfamide and doxorubicin was instituted. Seven months after surgery, the patient started feeling chest and neck pain. On physical examination, she was dyspnoic and her upper chest was oedematous; the ataxia had worsened. A total body CT scan showed multiple lesions of the cervical paravertebral muscles and of the upper mediastinum. A cerebral MRI pointed out a large recurrence at the right cerebellopontine angle. Pain killer drug therapy was instituted, but the patient died a few days later.

The tumour showed highly cellular area composed of oval and round cells with eosinophilic cytoplasm. Rare neoplastic cells with pleomorphic nuclei, evident nucleoli and eosinophilic cytoplasm were present. Frequent atypical mitotic features were present. These cells were arranged mostly in fascicular patterns or in a solid fashion. Moreover, scattered large cells containing deeply eosinophilic cytoplasm were present. These features prompted us to consider a differential diagnosis between leiomyosarcoma, atypical teratoid rhabdoid tumour (ATRT) and RMS with pleomorphic features. However, smooth muscle antigen resulted negative, and myogenin and desmin were positive (figure 2). Moreover, the immunohistochemical study showed negativity for S-100 and INI1, excluding a diagnosis of malignant peripheral sheath tumour and ATRT. Given these characteristics, a diagnosis of pleomorphic RMS was performed.

DISCUSSION

Cerebellopontine angle tumours are relatively common lesions, with the majority being acoustic neuromas and meningiomas.

Figure 2 Tumour is composed of oval and round cells with abundant eosinophilic cytoplasm arranged in a fascicular pattern. In the midst were present neoplastic cells with pleomorphic nuclei and evident nucleoli. These cells were arranged in fascicular pattern. Atypical mitotic features were present. Immunohistochemical study revealed positivity for desmin and myogenin.

They can primarily arise from the cisterns, vascular and neuronal structures, from embryological remnants, or they can extend from the petrous bone or skull; finally, they can be secondary to an exophytic brainstem or ventricular tumour.⁷ The present case is a unique initial presentation of a pleomorphic RMS of the cerebellopontine angle. To the best of our knowledge, such a case has never been described in English-written literature.

RMS has a male predilection (M:F 1.6:1) and the age of presentation is 44–58 years.^{6–8} It presents as a solid mass in the majority of cases, but infrequently, it assumes the form of a diffuse disease with multiple lytic lesions, simulating a non-Hodgkin lymphoma.^{4–9} Between the four histological types of RMS, the pleomorphic one is the rarest, even in the paediatric population,⁶ with an incidence of 0.0064 cases/100 000 inhabitants/year.¹⁰ After the age of 15, its incidence increases. Indeed, the median age of presentation is 50 years (range 24–83).¹¹ Pleomorphic RMS is localised, in the descending order of frequency, to the extremities, the genitourinary apparatus, the trunk and the orbit.^{10–11} Orbital pleomorphic RMS is strictly related to radiation therapy.¹² Survival is correlated to the primary disease location, lesion amenability to resection and age. The primary involvement of the head and neck has been associated with a worse prognosis due to an early invasion of noble structures.^{5–13} While the 5-year overall survival rate of localised pleomorphic RMS is 53.4%, initially diagnosed diffuse or multiple disease carries the worst prognosis with a 5-year overall survival rate of 4.3%.¹¹ La Quaglia *et al*¹⁴ found that survival directly correlates with age: children, who are rarely involved, carry the worst prognosis. Surgery is the mainstay of treatment. Patients submitted to a gross total resection achieve a 5-year overall survival rate of 55.7%.¹¹ Although it has not been determined whether a one-piece resection carries a better prognosis, a piece-meal resection carries a high risk of neoplastic dissemination. However, in our case, for anatomical issues, a piece-meal removal was the only option.

MRI cannot differentiate RMS from other soft-tissue invasive neoplasms. Usually, it is depicted as homogenous, isointense on T1-weighted images and highly intense on T2-weighted images. Intra-axial lesions, as those described and reviewed by Celli *et al*,⁸ enhance readily after contrast administration and permit the assessment of meningeal involvement. In our case, the extra-axial lesion did not involve the meninges and presented with multiple intralesional cysts. This latter feature has not been described elsewhere in the literature. The differential diagnosis included the more frequent acoustic neuroma, because of the invasion of the internal acoustic canal and its radiological features. Nevertheless, the rapid clinical progression suggested a more malignant neoplasm. The lack of the classic dural enhancement and of a clear implant base excluded the diagnosis of meningioma. Histologically, the tumour presented strong positivity for desmin and myogenin, which indicates a rhabdoid differentiation. Malignant peripheral nerve sheath tumours (MPNST) share this latter characteristic with pleomorphic RMS, but are also positive for S-100,¹⁵ which in our case was not. When MPNST present with pleomorphic rhabdoid components, S-100 positivity surely indicates nerve sheath differentiation.¹⁵

Adjunctive treatment follows a subtotal resection in every case where it is safe to be administered. The patient was, in accordance with the oncologists, treated with ifosfamide and doxorubicin, which was documented to be safe and more

effective than other association regimens at the time the patient was diagnosed with her disease. Recently, the development of new modalities of radiation administration, such as intensity-modulated radiation therapy, has permitted good control of lesions with a reduction in radiation-related morbidity, especially for deep-seated masses.

Learning points

- ▶ Although it has been previously said that rhabdomyosarcoma (RMS) can originate from cerebral meninges,^{5–6} the present report is the first description in the literature of a pleomorphic RMS of the cerebellopontine angle.
- ▶ This particular tumour carried a bad prognosis because of the vicinity of nervous structures and of the impossibility of achieving a one-piece resection.
- ▶ Moreover, the adjunctive treatments had to be effective against a potential remnant and in controlling a probable recurrence.

Contributors All the authors have actively contributed to the drafting of the present manuscript. FC and GL collected the data and wrote the manuscript; MA made the pathological examination and wrote the correlated findings; PM operated on the patient and reviewed the manuscript.

Competing interests None.

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REFERENCES

- 1 Agamanolis DP, Dasu S, Krill CE Jr. Tumors of skeletal muscle. *Hum Pathol* 1986;17:778–95.
- 2 Stout AP. Rhabdomyosarcoma of the skeletal muscles. *Ann Surg* 1946;123:447–72.
- 3 Weil RJ, Zhuang Z, Pack S, *et al*. Intramedullary Ewing sarcoma of the spinal cord: consequences of molecular diagnostics. Case report. *J Neurosurg* 2001;95:270–5.
- 4 Locatelli F, Tonani P, Porta F, *et al*. Rhabdomyosarcoma with primary osteolytic lesions stimulating non-Hodgkin's lymphoma. *Pediatr Hematol Oncol* 1991;8:159–64.
- 5 Gaiger AM, Soule EH, Newton WA Jr. Pathology of rhabdomyosarcoma: experience of the Intergroup Rhabdomyosarcoma Study, 1972–78. *Natl Cancer Inst Monogr* 1981;56:19–27.
- 6 Hollowood K, Fletcher CD. Rhabdomyosarcoma in adults. *Semin Diagn Pathol* 1994;11:47–57.
- 7 Bonneville F, Sarrazin J, Marsot-Dupuch K, *et al*. Unusual lesions of the cerebello-pontine angle: a segmental approach. *Radiographics* 2001;21:419–38.
- 8 Celli P, Cervoni L, Maraglino C. Primary rhabdomyosarcoma of the brain: observations on a case with clinical and radiological evidence of cure. *J Neurooncol* 1998;36:259–67.
- 9 Rofsky NM, Genieser NB, Ambrosino MM, *et al*. Diagnosis of occult primary rhabdomyosarcoma by magnetic resonance imaging. *N Y State J Med* 1993;93:142–3.
- 10 Perez EA, Kassira N, Cheung MC, *et al*. Rhabdomyosarcoma in children: a SEER population based study. *J Surg Res* 2011;170:e243–51.
- 11 Ferrari A, Dileo P, Casanova M, *et al*. Rhabdomyosarcoma in adults: a retrospective analysis of 171 patients treated at a single institution. *Cancer* 2003;98:571–80.
- 12 Kony SJ, de Vathaire F, Chompret A, *et al*. Radiation and genetic factors in the risk of second malignant neoplasms after a first cancer in childhood. *Lancet* 1997;350:91–5.
- 13 Pasquier B, Courdec P, Pasquier D, *et al*. Primary rhabdomyosarcoma of the Central Nervous System. *Acta Neuropathol* 1975;33:333–42.
- 14 La Quaglia MP, Heller G, Ghavini F, *et al*. The effect of age at diagnosis on outcome in rhabdomyosarcoma. *Cancer* 1994;73:109–17.
- 15 Stasik CJ, Tawfik O. Malignant peripheral nerve sheath tumor with rhabdomyosarcomatous differentiation (malignant triton tumor). *Arch Pathol Lab Med* 2006;130:1878–81.

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