Unusual pattern of metastatic disease in a patient in their early childhood with group 4 medulloblastoma

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DESCRIPTION

A patient in their early childhood with a history of speech and fine motor delay presented with ataxia over several weeks. Neurological examination revealed only mild difficulty with tandem straight-line gait. MRI of the brain demonstrated a fourth ventricular mass with reduced diffusivity extending into the right foramen of Luschka associated with obstructive hydrocephalus (figure 1A). Spinal MRI showed an apparent extramedullary intradural mass at C1 and C2 with similar signal characteristics to the brain lesion concerning for metastatic disease without evidence of leptomeningeal disease (figure 1B). Expansion of the central canal was appreciated from C1 to C3, likely representing a cord syrinx associated with altered cerebral spinal flow from the brain and cervical lesions. On neurosurgical exploration, the spinal mass was intramedullary and was completely resected at the same time as gross total resection of the posterior fossa tumour. Pathology of the spinal tumour revealed sheets of small round blue cell neoplasm with pleomorphic and hyperchromic nuclei, nuclear moulding, high nuclear to cytoplasmic ratios and inconspicuous nucleoli (figure 2A,B). Histopathology of both the spinal and posterior fossa tumour confirmed a diagnosis of medulloblastoma, WHO grade IV. Both the posterior fossa and spinal tumour were classified as group 4 by methylation analysis and were P53 wildtype, non-MYC amplified. The patient was treated with craniospinal proton radiation and adjuvant chemotherapy and has been in remission for 5 years post-therapy.

Medulloblastoma is the most common primary malignant brain tumour in the paediatric population, with an approximately 10-fold higher incidence than in adults.[1] The clinical presentation is often non-specific and includes headache, vomiting and gait disturbances which are frequent with other posterior fossa tumours.[2 3] Medulloblastomas are classified into four molecular subgroups based on gene expression profiling, including *wingless (WNT), sonic hedgehog (SHH)*, group 3 and group 4.[4] Molecular subgroups 3 and 4 have the highest rates of metastases, and medulloblastoma has a propensity for leptomeningeal seeding and drop metastasis through the cerebrospinal fluid.[1 5 6] Intramedullary metastasis of medulloblastoma is rare; however, increased NOTCH1 pathway expression in spinal metastasis has been linked to group 3 medulloblastoma cells.[5 7] Tumour seeding via the central canal and widening from hydrocephalus may represent a mechanism of spread.[8 9] Other potential mechanisms include hematogenous dissemination or extension of a deposit from the subarachnoid space into the spinal cord.[9] To our knowledge, only six paediatric cases of intramedullary metastasis of medulloblastoma have been reported.[8–13] Four cases demonstrated signs of spinal cord compression, such as urinary incontinence or progressive muscle weakness.[8–11] Intramedullary metastasis was usually discovered after

Figure 1 Neuroimaging features of medulloblastoma and intramedullary spinal cord metastasis. (A) Post-gadolinium T1-weighted MRI sagittal sequence at diagnosis demonstrates a T1 hypointense posterior fossa mass with linear enhancement and distinct C2–C3 enhancing mass lesion (arrow) consistent with metastasis. (B) Axial post-gadolinium T1-weighted MRI sequence of the cervical metastatic lesion had the appearance of an extramedullary intradural tumour (arrow).

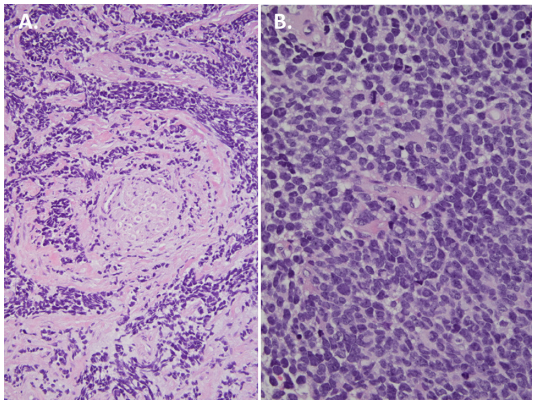

Figure 2 Pathological features of intramedullary cervical metastasis. (A) H&E-stained section of the spinal cord mass revealed an area of small round blue cell tumour cells surrounding a nerve associated with crush artefact (200×). (B) On higher power magnification (400×), an adjacent area reveals a more well-defined small blue cell tumour consistent with a histological diagnosis of classic medulloblastoma.

the intracranial lesion or at initial diagnosis if there were signs of spinal cord compression.[8–12] Additionally, none of the reported lesions involved the spinal cord at the C1–C2 level. Cervical myelopathy at the C1–C2 level from compressive lesions may present with impaired thermoception and nociception, perceptual dysfunction and upper limb muscle weakness.[14]

Our case represents an unusual pattern of intramedullary metastatic disease of the cervical cord in group 4 medulloblastoma without clinical correlates of spinal cord involvement.

Learning points

- Medulloblastoma metastases usually occur as leptomeningeal spread along the craniospinal axis; however, intramedullary metastases are a rare occurrence.
- Group 4 medulloblastomas may present with spinal metastatic disease.
- Patients with intramedullary metastases of medulloblastoma may not always present with clinical correlates of spinal cord compression or involvement.

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Case reports provide a valuable learning resource for the scientific community and can indicate areas of interest for future research. They should not be used in isolation to guide treatment choices or public health policy.

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