Hong Kong J Radiol. 2014;17:194-7 | DOI: 10.12809/hkjr1412139

CASE REPORT

Intraspinal Primitive Neuroectodermal Tumour with Haemorrhage

OC Leung[1], A Lo[2], KW Yang[2]

[1]Department of Radiology, Tuen Mun Hospital, Tuen Mun, Hong Kong; [2]Department of Radiology and Organ Imaging, United Christian Hospital, Kwun Tong, Hong Kong

ABSTRACT

Intraspinal primitive neuroectodermal tumour is rare in the paediatric population and particularly rare when it is purely intramedullary. This report describes a 16-month-old girl who presented with acute onset of torticollis and lower limb weakness. Subsequent imaging, operative findings, and histopathological examination confirmed the diagnosis of an intraspinal primitive neuroectodermal tumour.

Key Words: *Astrocytoma; Magnetic resonance imaging; Neuroectodermal tumors, primitive; Spinal cord neoplasms*

中文摘要

椎管內原始神經外胚層腫瘤伴出血

梁安祥、羅煦寧、楊國偉

椎管內原始神經外胚層腫瘤在兒童中很罕見，純粹發生於髓內更為罕有。本文報告一名16個月大的女童病例，病發時出現急性斜頸和下肢無力。隨後的影像、手術和病理結果證實病人患有椎管內原始神經外胚層腫瘤。

INTRODUCTION

Primitive neuroectodermal tumour (PNET) was initially described by Hart and Earle[1] in 1973 in malignant tumours containing poorly differentiated neuroepithelial cells, regardless of location or cell types. The most famous type of PNET is medulloblastoma, which is typically located at the infratentorial region at roof of the fourth ventricle. In 1993, the World Health Organization classification recommended use of the term "PNET" to describe medulloblastoma and other central nervous system (CNS) tumours that are histologically similar.[2] However, medulloblastoma is still the most commonly used term to describe infratentorial PNETs that arise at the midline, while a PNET arising from other parts of the CNS is usually described as a CNS PNET.

Intraspinal PNET is rarely seen and could be intramedullary, intradural but extramedullary, or extradural. A purely intramedullary PNET is extremely rare; less than 30 cases have been reported in English literature[3] and to our knowledge, this is the first patient reported in Hong Kong.

CASE REPORT

In January 2012, a 16-month-old girl was admitted to the Accident and Emergency Department of United

Correspondence: *Dr KW Yang, Department of Radiology and Organ Imaging, United Christian Hospital, 130 Hip Wo Street, Kwun Tong, Kowloon, Hong Kong.*
Tel: (852) 3513 5323; Fax: (852) 3513 5524; Email: yangkw@ha.org.hk

Submitted: 24 Sep 2012; Accepted: 4 Jun 2013.

Christian Hospital because of decreased oral intake and acute onset of torticollis for 2 days, without recent history of neck injury. On physical examination, she was afebrile but irritable. Her neck was stiff and persistently turned to the right side. Rigidity of the neck was noted on passive turning. No abnormal neck mass could be palpated. Ultrasonography (USG) of the neck did not show any abnormal collection, lymph node, or sternocleidomastoid tumour. However, the day after admission, she was noted to have bilateral lower limb weakness and could not stand even with support.

Urgent magnetic resonance imaging (MRI) was arranged on the same day, which showed an ill-defined intramedullary mass with contrast enhancement from C1 to T1 segments of the spinal cord with cord expansion, and there was evidence of haemorrhage at the inferior aspect of the lesion (Figures 1 to 4). MRI of the brain was unremarkable. The initial radiological diagnosis was astrocytoma. C1 laminectomy and C3-C7 open-door laminotomy were performed, together with excision and debulking of the intramedullary tumour.

Pathological analysis confirmed a small round cell tumour with immunohistochemical staining positive for

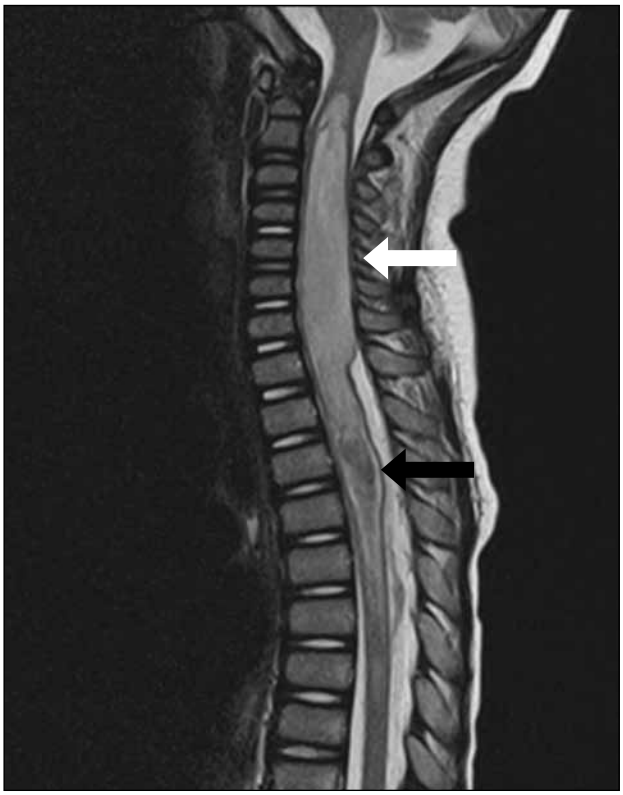

Figure 2. A T2-weighted sagittal magnetic resonance image shows an ill-defined hyperintense lesion at the cervical and upper thoracic cord (white arrow). Hypointensities at the inferior aspect of the lesion could represent previous haemorrhage (black arrow).

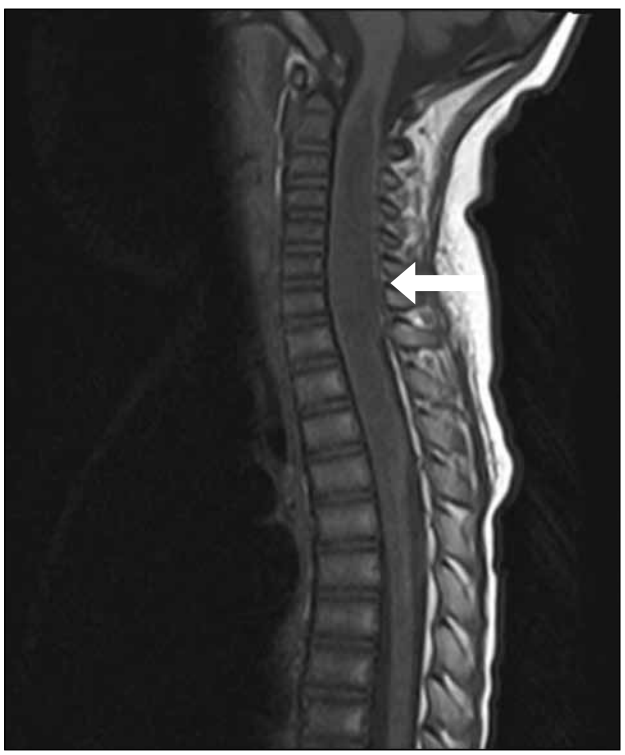

Figure 1. A T1-weighted sagittal magnetic resonance image shows an ill-defined hypointense lesion at the cervical and upper thoracic cord with cord expansion (arrow).

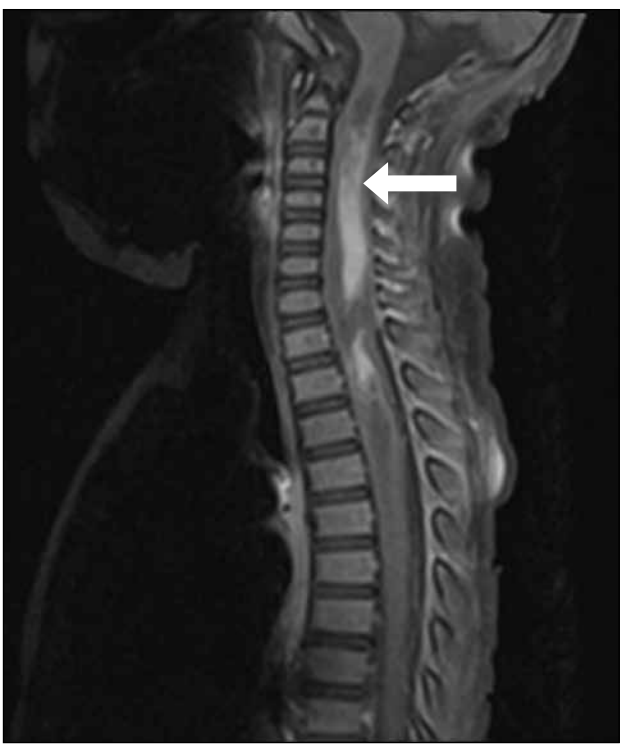

Figure 3. A contrast T1-weighted sagittal magnetic resonance image shows heterogeneous contrast enhancement of the cord lesion (arrow).

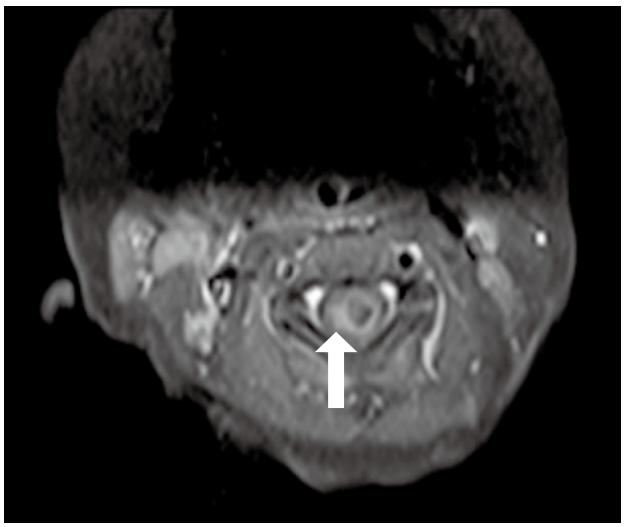

Figure 4. A contrast T1-weighted axial magnetic resonance image shows heterogeneous contrast enhancement of the cord lesion, which is purely intramedullary (arrow).

both neuronal differentiation (synaptophysin and NB84 +ve) and glial differentiation (focal GFAP +ve). These features were consistent with CNS PNET.

Chemotherapy was commenced and well-tolerated. Subsequent MRI showed persistent cervical cord expansion with evidence of cystic changes. Residual minimal contrast enhancement at the inferior aspect could represent postoperative change or residual tumour. The baby girl remained active, and only slightly increased muscle tone was noted at left lower limb across the knee and ankle joints. Further courses of chemotherapy will be given.

DISCUSSION

PNETs are a group of malignant tumours composed of highly undifferentiated neuroepithelial cells. The commonest location is in the cerebellum, but it can also be found in other parts of the CNS, including the cerebral hemispheres, pineal gland, brainstem, and spinal cord.

Intraspinal tumours are uncommon in paediatric patients, with the most ones being astrocytomas and ependymomas. The former are more common in young children and the latter in young adults. Drop metastases from primary intracranial tumours disseminating via cerebrospinal fluid account for the majority of spinal PNETs. Primary intraspinal PNET is a rare entity, and less than 30 patients with intramedullary PNET have been reported in the English literature. Intramedullary primary PNET can present in both children and adults and has no gender predilection. Most preferentially involve the thoracic region. To date, no distinctive radiological features have been established.

Our patient was admitted for acute onset of torticollis, and initial investigations were conducted to rule out common osseous and soft tissue causes of torticollis. However, plain radiographs and USG of the neck did not review any significant lesion. Subsequently, she was noted to have bilateral lower limb weakness. Urgent MRI showed an infiltrative and purely intramedullary cervical cord tumour, with imaging characteristics similar to astrocytoma. However, there was evidence of haemorrhage at the inferior aspect of the tumour, which was not a typical feature of astrocytomas, nor was it reported previously in intraspinal PNETs.

The diagnosis of PNET depends upon histopathological and immunobiochemical assessment, because of its rather non-specific imaging characteristics. No optimal treatment for intraspinal PNET has been established and in our patient and other reported cases, the primary treatment was surgery followed by chemoradiotherapy.[4-6] However, the clinical outcome is usually poor, and most patients usually die from the disease. On going to press with this report, our patient had survived 8 months post-surgery and had received eight cycles of chemotherapy which had all been well-tolerated. She walked independently with a mildly limping gait due to left-sided weakness, but otherwise she was clinically well. A follow-up MR study 6 months after initial surgery revealed that most of the intraspinal mass disappeared and was replaced by cystic areas. Residual contrast enhancement at the inferior aspect of the lesion could represent post-treatment change or residual tumour. Further follow-up MR studies will be required to evaluate the outcome of this lesion.

In conclusion, intraspinal PNET is a rare spinal cord tumour that has never been reported in Hong Kong. Our case was particularly unique as it was an intramedullary tumour in the cervical region and the first reported to have tumoural haemorrhage. Its MR imaging features were akin to an astrocytoma. Histopathological and immunobiochemical assessments were necessary to make a definitive diagnosis.

REFERENCES

1. Hart MN, Earle KM. Primitive neuroectodermal tumors of the brain in children. Cancer. 1973;32:890-7. crossref

2. Kleihues P, Burger PC, Scheithauer BW. The new WHO classification of brain tumours. Brain Pathol. 1993;3:255-68. cross ref
3. Mulholland CB, Barkhoudarian G, Cornford ME, McBride DQ. Intraspinal primitive neuroectodermal tumor in a man with neurofibromatosis type 1: Case report and review of the literature. Surg Neurol Int. 2011;2:155. cross ref
4. Ellis JA, Rothrock RJ, Moise G, McCormick PC 2nd, Tanji K, Canoll P, et al. Primitive neuroectodermal tumors of the spine: a comprehensive review with illustrative clinical cases. Neurosurg Focus. 2011;30:E1. cross ref
5. Han IH, Kuh SU, Chin DK, Kim KS, Jin BH, Cho YE. Surgical treatment of primary spinal tumors in the conus medullaris. J Korean Nerosurg Soc. 2008;44:72-7. cross ref
6. Kumar R, Reddy SJ, Wani AA, Pal L. Primary spinal primitive neuroectodermal tumor: case series and review of the literature. Pediatr Neurosurg. 2007;43:1-6. cross ref