Cystic liver metastases from extracranial primitive neuroectodermal tumour: a case report

Bharti K. Shah
Kieran McHugh

Abstract We report a rare case of cystic metastases to the liver from a peripheral primitive neuroectodermal tumour in a child. Ultrasound and CT appearances are described with a discussion on the histological findings and the differential diagnosis.

Introduction

Extracranial primitive neuroectodermal tumours (PNET) are a group of morphologically similar, highly malignant, small round-cell tumours. These are of presumed neural crest origin, arising outside the central and sympathetic nervous system [1, 2]. Metastases to the liver from these lesions are rare and cystic liver metastasis has not been described before.

Case report

A previously healthy girl presented at 7 months of age with left facial weakness. A CT brain scan at that time was normal and her facial weakness resolved in 2 weeks. At 14 months she was readmitted to the hospital with a 3-day history of right-sided facial weakness and a 4-week history of progressive peri-orbital swelling of the left eye and vomiting. She had had recurrent ear infections over the previous 5 weeks. The parents had also noticed an increased abdominal girth.

On examination, there was left-sided ptosis with depression of the globe and mild right-sided facial weakness, which was of lower motor neuron type. A maculopapular rash on her shoulder and napkin area was noted. Abdominal examination revealed massive, tender, smooth hepatomegaly. She had marked cervical lymphadenopathy and a few inguinal lymph nodes.

An MRI study showed a retrobulbar extracranial mass within the left orbit, depressing and distorting the globe. There was a further soft-tissue mass in the right infratemporal fossa, extending superiorly into the middle cranial fossa and elevating the temporal lobe (Fig. 1). Further masses were seen in the left petrous bone and in the right paravertebral region, extending beneath the right occipital bone and medially into the cervical canal. The masses were isointense to brain on T1-weighted images, heterogeneous with high intensity areas on T2-weighted images and showed variable enhancement after gadolinium administration. There was also evidence of right mastoiditis.

Ultrasound showed multiple anechoic cystic lesions with posterior enhancement in both lobes of the liver. A large cyst in the right lobe measured 6 × 8.4 × 7 cm and contained a mobile cystic lesion within measuring 5 × 3.7 × 5 cm (Fig. 2). The cysts in the left lobe contained some echogenic areas, but the rest of the cyst did not have any solid component.

Contrast-enhanced CT confirmed multiple cysts of varying sizes throughout the liver, the largest measuring 8 cm. A rim of calcification was noted posteriorly in the liver adjacent to one cyst (Fig. 3). One of the cysts was closely related to the porta hepatitis. The kidney and spleen were normal, as was a skeletal survey. At this juncture, the mother mentioned a 1-cm nodular swelling, present in the thigh for some time. A biopsy of the skull lesion was performed and a right thigh mass excised. Histology of both lesions showed infiltration by a malignant, small, round-cell tumour arranged in sheets and trabeculae and separated by broad fibrous septa. Immunohistochemistry showed positive cytoplasmic staining for cytokeratin, neuron-specific enolase and vimentin. There was strong membrane staining for MIC2. Staining for S-100, desmin, alpha-fetoprotein, LCA and PGP 9.5 were negative. The overall features were consistent with the diagnosis of PNET. The primary was thought to be the lesion in the right thigh.
Discussion

The differential diagnoses in this case based on the clinical and imaging findings, included Langerhan’s cell histiocytosis, metastatic neuroblastoma and hydatid disease. Less likely diagnoses were rhabdoid tumour of the liver and cystic hepatoblastoma. Due to the cystic nature of the liver lesions and because hydatid disease was a possibility, biopsy was deemed inappropriate. Biopsy of the skull lesion and excision of the right thigh mass were performed instead.

Malignant round-cell tumours of childhood are a composite group which include Ewing’s sarcoma, rhabdomyosarcoma, neuroblastoma and lymphoma [2, 3]. PNET is classified as a separate entity on the basis of its distinct clinical profile and ultrastructural, immunocytochemical and cytogenetic features [1]. Histologically, the diagnosis is suggested on light microscopy by the presence of Homer-Wright pseudorosettes. On electron microscopy, cytoplasmic process, neurosecretory granules and microtubules are observed. Immunohistochemistry shows neuronspecific enolase and MIC2 positivity, though neither is specific for this condition. Cytogenetic analysis shows a non-random balanced translocation between chromosomes 11 and 22 [t(11:22)(q24;q12)] [2].

PNETs are more common in the central nervous system. The sites of extracranial PNET, in decreasing order of frequency, are the chest, pelvis, and abdomen, including the retroperitoneum and neck. These tumours have an overall metastatic potential similar to other round-cell tumours. PNET tend to be more locally aggressive with lower incidence for bone involvement than the other tumours. The sites of distant metastasis are usually lung or bone, with bone marrow, lymph node and liver involvement being less common [2, 3].

In general, cystic hepatic lesions are uncommon in childhood and present a diagnostic dilemma. They may be congenital, acquired or neoplastic. Congenital cysts comprise mesenchymal hamartoma, epidermoid cysts, simple cysts and cysts in association with polycystic renal disease [4]. Acquired cysts may be infective or traumatic.

Amongst neoplastic lesions in children, hepatoblastomas with cystic change are not uncommon. On review of the English language literature, no reports of cystic metastasis to the liver from a peripheral PNET have

Fig. 1 Parasagittal, T1-W contrast-enhanced MRI showing a mass in the right infratemporal and paravertebral region (arrow)

Fig. 2a Ultrasound of the liver showing two cysts, the anterior one (arrow) containing a cystic lesion within

Fig. 2b Post-contrast CT of the abdomen showing multiple cysts in the liver, one anterior to the IVC with a rim of calcification (arrowhead)