Case Report

Malignant rhabdoid tumor—The great mimicker: Two case reports

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Abstract

Malignant rhabdoid tumors (MRT) represent a very rare group of tumors in infants that have an aggressive clinical behavior, are refractory to therapy, and have a high mortality rate. We present two cases that demonstrate the importance of early diagnosis for further treatment. The first case was a 4 and a half month old boy that presented with an enlarged liver and was treated as hepatoblastoma with metastatic changes in the lung parenchyma after the diagnosis. The second case was a female neonate presenting with hardening on the right side of the abdomen which was treated as MRT of the liver after radiological and clinical evaluation. In both cases, the initial histopathologic diagnosis was hepatoblastoma, but later analyses confirmed the diagnosis of MRT. From our experience, imaging findings can be a valuable tool in directing further evaluation since even histopathologic differentiation from the small cell undifferentiated hepatoblastoma is a real challenge.

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Introduction

Malignant rhabdoid tumors (MRT) represent less than 5% of malignant liver tumors in infants, have an aggressive clinical behavior, are refractory to chemotherapy and radiation, and have a high mortality rate [1,2]. Patients usually appear with a fever, abdominal distension, very elevated LDH (lactate dehydrogenase) and normal or lower AFP (alpha fetoprotein) levels at the time of diagnosis. One of the greatest challenges for successful treatment is a correct diagnosis, especially differentiation of MRT from the small cell undifferentiated hepatoblastoma (SCUD-HB) that has different treatment regimens [3]. Opposite to hepatoblastoma which affects children most often during the first 18 months of life, MRT affects infants during first 8 months of life [4]. We present two cases during a period of 5 years, showing diagnostic pitfalls in differentiating MRT from SCUD-HB and the importance of early diagnosis for further treatment.

Case report

First case

Four and a half month old boy was hospitalized in the regional hospital due to hematemesis and melena. Proximal GI endoscopy showed normal findings. Physical examination determined that the liver was significantly enlarged on palpation. Total and conjugated bilirubin, AST (aspartate aminotransferase), ALT (alanine aminotransferase), albumin,
and coagulation status were normal. Abdominal ultrasound showed a heterogeneous, predominantly hypoechoic lesion that appeared to be in the caudal part of the liver, below the portal vein, medially from the gallbladder.

After that, the patient was transferred to our hospital where CT (Fig. 1) and MRI (Fig. 2) of the abdomen showed an expansive, heterogeneous lesion in the liver segment IVb, with signs of necrosis and without hemorrhage. It was considered to be hepatoblastoma with at least 3 smaller lesions in the liver segment V. AFP-alpha fetoprotein was slightly elevated, VMA (vanillylmandelic acid) and HVA (homovanillic acid) were negative. Initial radiological examination detected metastatic changes in the lung parenchyma and was followed by a partial resection of the primary tumor from the surface of the right hepatic lobe. Pathohistologic (PH) finding confirmed the diagnosis of hepatoblastoma with metastatic changes on the omentum. Chemotherapy was initiated according to the appropriate protocol, but the diagnostic evaluation after the third cycle showed a progression of the metastatic changes in the lungs, while the tumor in the liver regressed. Due to the lack of appropriate therapeutic response, PH samples were sent for reevaluation to an external laboratory. The new PH finding suggested MRT, leading to a different chemotherapy treatment.

Radical surgery was performed on the primary tumor in the abdomen, which showed significant progression in the meantime. In surgery, the tumor appeared to be of retroperitoneal origin, infiltrating anterior abdominal wall, parts of small and large bowel, bladder, common bile duct, and surrounding liver parenchyma. Chemotherapy for rhabdoid tumor was continued; however, the metastatic changes in the right lung progressed further. The patient passed away at the age of one as the disease progressed.
During the hospitalization, she had good appetite, gained weight properly, did not have a fever. Laboratory findings showed AFP-alpha fetoprotein of 2158.5 IU/mL, beta HCG was not elevated. MRI showed a tumor in the fifth and sixth liver segment and the PH results confirmed the diagnosis of fetal hepatoblastoma. At the site of the biopsy, a tumor-like lesion developed in the front abdominal wall during next few days.

The patient was then transferred to our hospital where laboratory findings showed that white blood cells count was 21.0 $10^9$/L, hemoglobin 144.0 g/L and platelet count 556.0 $10^9$/L. Ultrasound (Fig. 3) was repeated and MRI examination (Fig. 4) showed heterointense mass on T2W, with hyperintense cystic component and small solid component. Signs of hemorrhage were present in the cystic component on T1W and after administration of contrast the tumor was found to be inhomogeneous, with heterointense opacification and hyperintense capsule. There were signs of two more masses in the abdominal and chest wall with the same characteristics as the one in the liver. No other metastases were detected on the abdominal MRI or on the chest CT scan.

The mass in the front abdominal wall was extirpated and sent for PH analysis. Histologic and immunohistochemical characteristics were suggestive for rhabdoid tumor of the liver.

Resection of the fifth and sixth segment of the liver was done showing that the entire mass was infiltrating the falci-form ligament and front abdominal wall.

PH results showed a mixed tumor, consisted of small cell undifferentiated hepatoblastoma and MRT of the liver (Fig. 5).

For the definitive confirmation of the rhabdoid component, genetic testing for SMARCB1/INI1 needed to be done.

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Second case

One week after the birth of the patient, parents noticed the hardening on the right side of the abdomen. The primary care practice referred the girl to the specialized pediatric hospi-tal. During the hospitalization, she had good appetite, gained weight properly, did not have a fever. Laboratory findings showed AFP-alpha fetoprotein of 2158.5 IU/mL, beta HCG was not elevated. MRI showed a tumor in the fifth and sixth liver segment and the PH results confirmed the diagnosis of fetal hepatoblastoma. At the site of the biopsy, a tumor-like lesion developed in the front abdominal wall during next few days.

The patient was then transferred to our hospital where laboratory findings showed that white blood cells count was 21.0 $10^9$/L, hemoglobin 144.0 g/L and platelet count 556.0 $10^9$/L. Ultrasound (Fig. 3) was repeated and MRI examination (Fig. 4) showed heterointense mass on T2W, with hyperintense cystic component and small solid component. Signs of hemorrhage were present in the cystic component on T1W and after administration of contrast the tumor was found to be inhomogeneous, with heterointense opacification and hyperintense capsule. There were signs of two more masses in the abdominal and chest wall with the same characteristics as the one in the liver. No other metastases were detected on the abdominal MRI or on the chest CT scan.

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samples were sent to a foreign institution and analysis showed loss of SMARCB1 staining in the MRT. Chemotherapy was applied according to the protocol from European Rhabdoid registry (V2 2010). A year after the initial administration in the regional hospital, the patient is alive and well.

Discussion

INI1-negative immunoreactivity and the presence of rhabdoid morphology are the current basis of histologic diagnosis of MRT. When the MRT lacks classic rhabdoid morphology it is often misclassified as SCUD-HB if additional test for SMARCB1 deletion is not done [5]. Case reports of misdiagnosed MRTs that were treated as hepatoblastomas leading to the disease progression and patient’s death [6]. In a recently published paper, Fazlollahi et al. suggest that the term INI1-negative hepatoblastoma should be eliminated and liver tumors with small cell or rhabdoid morphology that are negative for hepatocellular markers (Hep-par and Arginase) and INI1-negative should be diagnosed as MRT [7].

Garces-Inigo et al. have shown that MRT does not exhibit specific features on MRI, but it usually appears as hypointensity on T1W images and heterogeneous hyperintensity on T2W images [8]. Hepatoblastomas appear differently on imaging modalities depending on histologic type ranging from relatively homogenous epithelial type to heterogeneous masses with calcifications and areas of hemorrhage and necrosis in mixed type [9]. Calcifications are found in more than 50% of hepatoblastomas and can be a useful feature for making a differential diagnosis, since they are rare in MRT [10].

There are not many reports describing specific characteristics of this tumor that can be detected using imaging. Similar to our case report, Kapral et al. showed relatively T2 hyperintense mass with a hemorrhage into the cystic component. The solid component of the mass was T1 hypointense and post-contrast images showed heterogeneous enhancement while the cystic component showed rim enhancement [11].

Since the tumor in the second case did not have typical imaging characteristics for hepatoblastoma, we started considering that a different diagnosis is possible. The first case we reported reminded us that even though it is very rare, MRT should be considered when discussing malignant liver lesion in children in the first year of life. Even though PH in the second case suggested hepatoblastoma, similarity of imaging appearance of these two tumors, led us to do further diagnostics in order to confirm MRT.

Conclusion

Being difficult to treat and having a low survival rate, especially with metastasis present at the time of diagnosis, early and accurate diagnosis of MRT is crucial. From our experience imaging findings can be a valuable tool in directing further evaluation and providing the best treatment options for this aggressive tumor.

Patient consent statement

Patient’s parents provided written informed consent for the publication of this report.

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