CASE REPORT | OLGU SUNUMU

Hepatoblastoma in a child with neurofibromatosis type I

Nörofibromatozis tip I'de hepatoblastom olgusu

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A major hallmark of neurofibromatosis type I (NFI) is the development of benign tumors. Hepatoblastomas associated with NFI is reported rarely in the literature. We present here a one year-old girl with both NFI and hepatoblastoma. Hepatoblastoma can be associated with NFI. Abdominal ultrasound is not included within the diagnostic criteria or surveillance of NFI. As NFI can be associated with hepatoblastoma and other abdominal tumors, a careful physical examination and an abdominal USG if necessary should be considered.

Keywords: Hepatoblastoma; neurofibromatosis type I.

Nörofibromatozis tip I'in (NFI) en önemli bulgularından biri selim tümörlerdir. Bu hastalıkta hepatoblastom görülmesi literatürde çok nadir olarak bildirilmiştir. Burada Nörofibromatozis tip I ve hepatoblastomu olan bir yaşında bir kız çocuğu sunulmaktadır. Hepatoblastom nörofibromatozis tip I ile birlikte görülebilir. Nörofibromatozis tip I'de hepatoblastom ve diğer batın içi tümörler görülebileceğinden dikkatli bir fizik bakı ve gerekirse batın ultrasonografi yapılması gereklidir.

Anahtar sözcükler: Hepatoblastom; nörofibromatozis tip I.

Neurofibromatosis (NF) type I is one of the most common genetic disorders in the general population with approximately 1 in 3500 people.[1] Diagnostic criteria consists of; two or more of the following seven features; 1- Six or more cafe au lait sposts (1.5 cm or larger in postpubertal individulas, 0.5 cm or larger in prepubertal individulas), 2- Two or more neurofibromas of any type or one or more plexiform neurofibromas 3- Freckling of armpits or groin 4- Optic glioma (tumor of the optic pathway), 5- Two or more Lisch nodules (benign iris hamartomas), 6- A distinctive bony lesion (dsyplasia of sphenoid bone, dysplasia or thinning of long bone cortex) 7- First degree relative with NF.[2]

A population study from Japan of 26.084 children younger than 15 years revealed a six to eight fold increased incidence of cancer in NF1 patients. [3] Malignant peripheral nerve sheath tumors represent the most common neoplasm in approximately 5–10% of individulas with NFI.[4] Children with NFI have an increased risk of Wilm's tumor and myeloid leukemias, including acute myeloblastic leukemia, myelodysplasia and myeloproliferative syndromes.[5,6] In addition neuroblastoma has been reported in patients with NFI.[7] Other malignancies occur less frequently in patients with NF1, including pheochromocytoma, rhabdomyosarcoma and brain tumors other than optic gliomas.[8] There is only one case report of NFI associated with hepatoma and one with hepatoblastoma in the literature.[9,10]

CASE REPORT

A one year old girl living in a European country who was recently diagnosed with NFI presented to our child neurology clinic for a second-opinion. She was born full-term with a birth weight of 3730

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grams, with no difficulties. She sat up at the age of 8 months and started to walk at the age of 11 months. She spoke 3–4 words at the time of examination. She was a healthy 1 year-old girl with no complaints related with her abdomen. In her examination she had 15 cafe au lait spots more than 0.5 cm in diameter. She had no axillary or inguinal freckling, no osseus lesions or cutaneous neurofibromas. An abdominal mass was palpated at the right upper quadrant. She had the genetic confirmation as well because she did not fulfill the requirement for a definitive NFI. Her ophthalmological examination did not show any findings. MRI of the brain was performed and did not show any abnormalities. Her abdominal USG showed a solitary non-homogeneous mass with peripheral hypoechoic rim in the posterior right lobe of the liver in segment 7. It was confirmed with abdominal MRI (Fig 1) that showed 56x40 mm T2 hyperintense, T1 hypointense mass with diffusion restriction and with heterogeneous contrast enhancement suggestive of a malignant lesion. The patient's family decided to have surgery in the the country they currently live. The pathology report revealed hepatoblastoma.

DISCUSSION

Although primary hepatic cancers are rare in children, they are the third most frequent abdominal solid tumor in children. Two major types of hepatic cancer in infants and children have been described, hepatoblastoma and hepatocellular carcinoma (hepatoma). Hepatoblastoma is usually found in children younger than the age of 4 years and hepatocellular carcinoma usually occurs in children older than 6 years.[9] Both hepatoma and hepatoblastoma were found in NFI patients.[9,10]

To establish the extent of disease in an individual diagnosed with NFI, the following evaluations are recommended: Personal medical history with particular attention to features of NFI, physical examination with particular attention to the skin, skeleton, cardiovascular system, and neurologic systems, ophthalmologic evaluation including slit lamp examination of the irides, developmental assessment in children, other studies only as indicated on the basis of clinically apparent signs or symptoms, genetics consultation.[11,12]

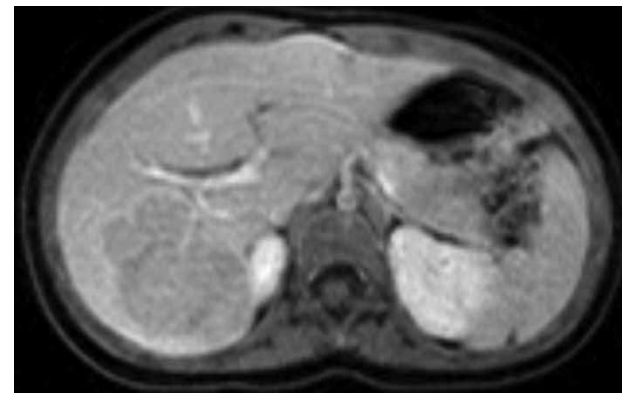

Fig. 1. 56x40 mm T1 hypointense mass in the right lobe segment 7, with diffusison restriction and heterogenous contrast enhancement.

The surveillance of NFI consists of annual physical examination by a physician who is familiar with the disease, annual ophthalmologic examination in early childhood, less frequent examination in older children and adults, regular developmental assessment by screening questionnaire (in childhood), regular blood pressure monitoring, other studies only as indicated on the basis of clinically apparent signs or symptoms, monitoring of those who have abnormalities of the central nervous system, skeletal system, or cardiovascular system by an appropriate specialist.[11,12] As NFI can associated with hepatoblastoma and other abdominal tumors, a careful physical examination and an abdominal USG if necessary, should be considered.

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