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.

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 osseous 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. Both hepatoma and hepatoblastoma were found in NFI patients.

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.

**REFERENCES**

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