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Could APC gene screening be useful in children with hepatoblastoma? Early onset of adenocarcinoma in a child with familial adenomatous polyposis and hepatoblastoma

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ABSTRACT

Familial adenomatous polyposis is an inherited disorder characterized by the development of hundreds of colorectal adenomas during adolescence, which in many cases will transform into colorectal cancer by the fourth decade of life, along with the development of various malignant tumors including hepatoblastoma. We report on a female patient with a de novo interstitial deletion of 5q21.3-q23.3, encompassing the APC gene, associated with adenomatous polyposis and early colorectal cancer, hepatoblastoma, epidermoid cysts, mental retardation, several mild dysmorphic signs and lower limb venous thrombosis.

Key words: adenocarcinoma, attenuated familial adenomatous polyposis, hepatoblastoma, 5q deletion, APC gene.

The authors declare no conflict of interest.

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