

MEETING ABSTRACT



The case of the youngest infant with hepatoblastoma with APC gene mutation

Maryna Krawczuk-Rybak^{1*}, Anna Jakubiuk-Tomaszuk², Elżbieta Skiba¹, Andrzej Pławski³

From Annual Conference on Hereditary Cancers 2011 Szczecin, Poland. 17-18 November 2011

Hepatoblastoma is a rare early malignant liver neoplasm occurring in infants and children. Some cases of hepatoblastoma are associated with genetic conditions such as trisomies of chromosomes 18, Beckwith-Wiedemann syndrome and familial adenomatous polyposis (FAP). The observed increase in the risk of hepatoblastoma in APC (adenomatous polyposis coli) gene mutation carriers is low, not exceeding 1%, which is associated with APC gene mutation located in the region of codons 459-1309, where most mutations identified in families affected by FAP in Poland are found. Tissues obtained from patients with hepatoblastoma but without the diagnosis of FAP show loss of heterozygocity at the locus of APC or MCC (mutated in colorectal cancers) genes. Hepatoblastoma growth is the result of the sequence of changes in genetic material. However, a major role can be ascribed to the Wnt pathway where somatic mutations have been observed in the genes of Bcathenin (CTNNB1, Cathenin (cadherin-associated protein), beta 1.88kD) and AXIN1 (Axis inhibitor 1). We present a case of familial hepatoblastoma in a 3-month-old infant with a constitutional mutation in the APC gene.

Author details

¹Department of Pediatric Oncology and Haematology, Medical University of Bialystok, Poland. ²Department of Pediatric Laboratory Diagnostics, Medical University of Bialystok, Poland. ³Institute of Human Genetics, Polish Academy of Sciences, Poznan, Poland.

Published: 20 April 2012

doi:10.1186/1897-4287-10-S3-A18

Cite this article as: Krawczuk-Rybak *et al.*: **The case of the youngest infant with hepatoblastoma with APC gene mutation**. *Hereditary Cancer in Clinical Practice* 2012 **10**(Suppl 3):A18.

¹Department of Pediatric Oncology and Haematology, Medical University of Bialystok, Poland

Full list of author information is available at the end of the article

Submit your next manuscript to BioMed Central and take full advantage of:

- Convenient online submission
- Thorough peer review
- No space constraints or color figure charges
- Immediate publication on acceptance
- Inclusion in PubMed, CAS, Scopus and Google Scholar
- Research which is freely available for redistribution

) Bio Med Central

Submit your manuscript at www.biomedcentral.com/submit



© 2012 Krawczuk-Rybak et al; licensee BioMed Central Ltd. This is an Open Access article distributed under the terms of the Creative Commons Attribution License (http://creativecommons.org/licenses/by/2.0), which permits unrestricted use, distribution, and reproduction in any medium, provided the original work is properly cited.