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Titel des Beitrags:
[Molecular pathology in hereditary colorectal cancer. Recommendations of the Collaborative German Study Group on hereditary colorectal cancer funded by the German Cancer Aid (Deutsche Krebshilfe)]

Abstract:
Although twin studies indicate that inherited genetic factors contribute to about 35% of colorectal cancers (CRC), the exact genetic background has currently been elucidated in only 5-10% of cases. These comprise several hereditary cancer predisposition syndromes that present with a high number of syn- or metachronous neoplasms within an affected person and/or family. Many of these tumors exhibit typical histopathological changes. In general, one should discriminate between cancer syndromes associated with adenomatous and non-adenomatous (i.e., hamartomatous) polyps, the latter being quite rare. The patient’s age often serves as a substantial hint to hereditary cancer. The next step of diagnostic work-up includes analysis of microsatellite instability (MSI) together with immunohistochemical detection of a loss of expression in one of the most frequently affected mismatch repair genes (MSH2, MSH6; MLH1, PMS2). Finally, the molecular demonstration of a gene mutation in the blood or germline is the most expensive and tedious procedure.
This requires a signed informed consent from the patient after appropriate genetic counseling.

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