Practice Parameters For The Identification And Testing Of Patients At Risk For Dominantly Inherited Colorectal Cancer

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It should be recognized that these guidelines should not be deemed inclusive of all proper methods of care or exclusive of methods of care reasonably directed to obtaining the same results. The ultimate judgment regarding the propriety of any specific procedure must be made by the physician in light of all of the circumstances presented by the individual patient.

Inherited colorectal cancer includes a group of syndromes in which predisposition to the disease is based on a germline mutation that may be transmitted from parent to child. The risk of colorectal cancer is very high in patients affected by such a mutation. Guidelines addressing the recognition, testing, and screening of families and patients with the syndromes of familial adenomatous polyposis and hereditary non-polyposis colorectal cancer are presented and discussed:

Take a family history. This is the first step in recognizing families possibly affected by inherited colorectal cancer.

1. Document a suspicious pedigree; a family tree based on the recollection of family members is not solid enough evidence. Request medical records to confirm diagnosis.
2. Identify criteria for genetic testing. Familial adenomatous polyposis is easily recognized clinically when patients present with more than 100 colorectal adenomas. Fewer adenomas are needed for a diagnosis when a patient is part of an established kindred. The Amsterdam Criteria are a way of clinically identifying families with hereditary non-polyposis colorectal cancer, where a mismatch repair gene mutation can be detected. Testing for microsatellite instability in tumors is a screen for families with hereditary non-polyposis colorectal cancer where the clinical pattern of the disease is suggestive but not strong enough to fulfill Amsterdam Criteria.
3. Offer surveillance to families not meeting the above criteria for genetic testing. Families with more than two first-degree relatives affected with colorectal cancer, especially if one is affected at a young age (<45 years), need to be offered endoscopic surveillance even if genetic testing is not indicated.
4. Adhere to all protocols for genetic testing. Institutional review board approval, informed consent, and pretest and posttest counseling are the key elements of genetic testing for inherited colorectal cancer.

Summary: It is hoped that these guidelines will assist in the recognition and management of patients affected by syndromes of inherited colorectal cancer.