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# Molecular screening for Lynch syndrome. population based approach using immunohistochemistry and methylation analysis

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Microsatellite instability (MSI) in colorectal cancer is one of the few prognostic markers and markers of cancer biology that have made it from bench to bedside. MSI tumors have a better prognosis and may respond differently to chemotherapy, but here we will focus on MSI and screening for the hereditary cancer syndrome, Lynch syndrome, which affects 2-5 % of all colorectal cancer patients.

MSI is variable length mutations in tumor DNA caused by deficiency of DNA mismatch repair. Dysfunction of this repair system is caused by inactivation of any of the repair enzymes MLH1, MSH2, MSH6, or PMS2. It can be measured either on the DNA level as MSI or on the protein level with loss of expression of the affected protein. Lynch syndrome is caused by hereditary mutations in any of the four mismatch repair genes. About 15 % of all colorectal cancers have MSI, but not more than one in three of these are caused by germline mutations. The rest is caused by a sporadic phenomenon, promoter hypermethylation of MLH1.

Based on an exploratory study and a validation study, we have established a strategy for molecular screening for Lynch Syndrome with initial immunohistochemistry and in the case of MLH1 deficiency also promoter methylation analysis. The strategy is now implemented in our region and will be followed prospectively.

Several obstacles and challenges have to be met to bring knowledge from the laboratory to the patients. Molecular screening for Lynch syndrome may serve as a template for how to do this successfully.

## Biography

Lars Henrik Jensen is a medical doctor from University of Aarhus. He completed his Ph.D in 2007 from University of Southern Denmark and has been an exchange visitor at University of Southern California. His primary areas of research are gastrointestinal cancers, clinical trials, and molecular markers.