Testing strategies for Lynch syndrome in people with endometrial cancer

Lynch syndrome (also known as hereditary non-polyposis colorectal cancer [HNPCC]) is an inherited genetic condition that is associated with an increased risk of several cancers, including endometrial and colorectal cancer. NICE diagnostic guidance 27 recommends using immunohistochemistry- or microsatellite instability-based strategies to test for Lynch syndrome in people diagnosed with colorectal cancer. Currently, testing for Lynch syndrome in people diagnosed with endometrial cancer is not done, or only occurs for subgroups of people with an identified risk factor for the condition; for example, age at diagnosis or a family history of Lynch syndrome-related cancers. Endometrial cancer can be the first cancer to occur in people with Lynch syndrome and the optimal testing strategy may be different to testing strategies used in colorectal cancer. Identifying the syndrome at the point of endometrial cancer diagnosis could prevent the occurrence of further cancers in people with Lynch syndrome (such as colorectal cancer), through increased surveillance and strategies aimed at reducing risk. It may also help to identify family members with Lynch syndrome and help to reduce the incidence of primary cancers and increase early detection if cancer occurs.

Initial testing to identify tumours with deficient DNA mismatch repair (which indicates potential Lynch syndrome) can be done with microsatellite instability testing or immunohistochemistry. The use of subsequent tests to help rule out sporadic cancer (such as *MLH1* promoter hypermethylation testing) will also be considered.

The NICE diagnostics assessment programme will assess the clinical and cost-effectiveness of molecular testing strategies for Lynch syndrome in people with endometrial cancer in order to make recommendations on their use in the NHS.