Molecular testing for Lynch syndrome in people with colorectal cancer: systematic reviews and economic evaluation

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Plain English summary

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The DNA mismatch repair system corrects errors in DNA replication that occur when cells in the body divide. Without the mismatch repair system, errors in DNA multiply and this can lead to cancer developing, especially in the bowel and in the female reproductive organs.

Most people are born with two working copies of the genes responsible for the mismatch repair system, but some people inherit a faulty (mutated) copy of one of the genes. As they have only one working copy they are more likely to lose the function in the mismatch repair system and subsequently develop cancer. This leads to patterns of cancer in a family, which is known as Lynch syndrome.

Around two in five people with Lynch syndrome will develop bowel cancer during their lifetime and around one in three women with Lynch syndrome will develop womb cancer. People with Lynch syndrome are also more likely to develop ovarian cancer and a number of other cancers.

The aim of this study was to find out whether it would be clinically effective (i.e. good for patients and their families) and cost-effective (i.e. a good use of limited NHS resources) to screen bowel cancer patients for Lynch syndrome by testing their tumours. If tests on the tumours show signs of Lynch syndrome they can be offered genetic testing to search for the mutated gene responsible. Family members can then be offered a blood test to see if they have the mutated gene. The benefit of knowing that someone has Lynch syndrome is that surveillance can be offered, such as colonoscopy, to reduce the risk of cancer in the future.

Published evidence shows that tumour tests can help to identify Lynch syndrome. Although it has not been proven in practice, a mathematical model suggests that screening would be clinically effective and cost-effective.

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This report

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