NATIONAL INSTITUTE FOR HEALTH AND CARE EXCELLENCE

DIAGNOSTICS ASSESSMENT PROGRAMME

Equality impact assessment – Guidance development

Testing strategies for Lynch syndrome in people with endometrial cancer

Consultation

1.	Have the potential equality issues identified during the scoping process been addressed by the Committee, and, if so, how?

The following potential equality issues were identified during scoping:

- All people with cancer are covered under the disability provision of the Equality Act (2010) from the point of diagnosis. Information from tests in this assessment may influence decisions on fertility and conception. Pregnancy is a protected characteristic under the Equality Act.
- The specificity of MSI and IHC to detect potential Lynch syndrome associated endometrial cancer may decrease in older cohorts because the occurrence of somatic *MLH1* promoter hypermethylation increases with age (that is, a larger proportion of endometrial tumours with deficient MMR will be because of somatic, rather than inherited, causes with increased age).
- Clinical experts highlighted that endometrial cancer is often the first Lynch syndrome related cancer that occurs in women with the condition. Testing people at the point of endometrial cancer diagnosis will therefore provide an opportunity to identify the condition earlier and prevent subsequent Lynch syndrome related cancer.
- Clinical experts further commented that the numbers of variants of unknown significance that are identified may vary by ethnicity. People from ethnic groups in which few studies identifying mutations in Lynch syndrome associated genes have been done

are more likely to have a variant of unknown significance identified by testing.

The committee noted that endometrial cancer is often the first Lynch syndrome-associated cancer that women have and provides an opportunity to diagnose the condition before other Lynch syndrome-associated cancers, such as colorectal cancer, develop. This was highlighted in the draft guidance as a likely benefit of increased testing for Lynch syndrome after endometrial cancer (section 4.1 of the diagnostics consultation document).

The committee noted that the assessed strategy of straight to germline testing would result in an increased number of variants of uncertain significance being detected (that is, mutations in MMR genes for which it is not known if they cause Lynch syndrome or not) because initial tumour tests would not rule out non-pathogenic mutations (with no defective MMR) from proceeding to germline testing. Clinical experts also highlighted that for some ethnic groups fewer studies on Lynch syndrome causing mutations has been done. Therefore, for these groups there are likely to be more variants of uncertain significance would be identified by a direct to germline testing strategy (without the benefit of information from initial tumour tests). The committee noted that this may result in further testing of tumour tissue needing to be done for these groups to determine if the variant of uncertain significance is likely to be pathogenic (see section 4.8 of the diagnostics consultation document). This strategy (straight to germline testing) was not recommended by the committee.

No data on the accuracy of the tests by the age of the person tested were identified. The recommendation for use of IHC testing followed by *MLH1* promoter hypermethylation testing is for people of any age.

2.	Have any other potential equality issues been raised in the diagnostics assessment report, and, if so, how has the Committee addressed these?

No other potential equality issues were raised in the diagnostics assessment report.

3.	Have any other potential equality issues been identified by the Committee, and, if so, how has the Committee addressed these?

The committee noted that Lynch syndrome test results can have a substantial impact on people and so it is very important that patients and their carers have understood the full implications of a diagnosis of Lynch syndrome, for themselves and their families. This is especially important for people with a learning disability, who may need support from a carer to enable them to fully engage in discussions about testing and to provide informed consent. Recommendation 1.2 in the draft diagnostics guidance states that healthcare professionals should ensure that people are informed of the possible implications of test results for both themselves and their relatives and ensure that relevant support and information is available. Discussion of genetic testing should be done by a healthcare professional with appropriate training.

4.	Do the preliminary recommendations make it more difficult in practice for a specific group to access the technology compared with other groups? If so, what are the barriers to, or difficulties with, access for the specific group?

People with a learning disability may need support from a carer to enable them to fully engage in discussions about genetic testing and to provide informed consent. The importance of involving a carer in discussions has been highlighted in section 4.2 of the consultation document.

5.	Is there potential for the preliminary recommendations to have an adverse impact on people with disabilities because of something that is a consequence of the disability?

People with a learning disability may need support from a carer to enable them to fully engage in discussions about genetic testing and to provide informed consent. The importance of involving a carer in discussions has been highlighted in section 4.2 of the consultation document.

6.	Are there any recommendations or explanations that the Committee could make to remove or alleviate barriers to, or difficulties with, access identified in questions 4 or 5, or otherwise fulfil NICE's obligations to promote equality?

The recommendations specify that healthcare professionals should ensure that people are informed of the possible implications of test results for both themselves and their relatives and ensure that relevant support and information is available. This would include ensuring that people with a learning disability have a carer available to make sure they have support to allow them to fully engage in discussions about genetic testing and to provide informed consent for testing.

7.	Have the Committee's considerations of equality issues been described in the diagnostics consultation document, and, if so, where?

Committee consideration about the need for support for patients and their carers to understand the full implications of a diagnosis of Lynch syndrome, for themselves and their families, is described in section 4.2 and 1.2 of the diagnostics consultation document. This includes specific consideration that people with a learning disability may need support from a carer to enable them to fully engage in discussions about testing and to provide informed consent.

Section 4.2 of the diagnostics consultation document describes the committee's consideration that endometrial cancer may be the first Lynch syndrome associated cancer that people with the condition have.

Section 4.8 of the diagnostics consultation document describes the committee's consideration that people from ethnic groups in which few studies identifying mutations in Lynch syndrome associated genes have been done are more likely to have a variant of unknown significance identified by testing. This was noted as being a particular issue for this group if straight to germline testing was recommended (that is, with no testing of tumour tissue).

Approved by Associate Director (name): Rebecca Albrow

Date: 18/03/2020

Diagnostics guidance document

1.	Have any additional potential equality issues been raised during the consultation, and, if so, how has the Committee addressed these?
No additional potential equality issues were raised during the consultation.	

2.	If the recommendations have changed after consultation, are there any recommendations that make it more difficult in practice for a specific group to access the technology compared with other groups? If so, what are the barriers to, or difficulties with, access for the specific group?
The recommendations did not change after consultation.	

3.	If the recommendations have changed after consultation, is there potential for the preliminary recommendations to have an adverse impact on people with disabilities because of something that is a consequence of the disability?
The recommendations did not change after consultation.	

4.	If the recommendations have changed after consultation, are there any recommendations or explanations that the Committee could make to remove or alleviate barriers to, or difficulties with, access identified in questions 2 and 3, or otherwise fulfil NICE's obligations to promote equality?
The recommendations did not change after consultation.	

5.	Have the Committee's considerations of equality issues been described in the diagnostics guidance document, and, if so, where?

Committee consideration about the need for support for patients and their carers to understand the full implications of a diagnosis of Lynch syndrome, for themselves and their families, is described in section 4.2 and 1.2 of the diagnostics guidance document. This includes specific consideration that people with a learning disability may need support from a carer to enable them to fully engage in discussions about testing and to provide informed consent.

Section 4.1 of the diagnostics guidance document describes the committee's consideration that endometrial cancer may be the first Lynch syndrome associated cancer that people with the condition have.

Section 4.8 of the diagnostics guidance document describes the committee's consideration that people from ethnic groups in which few studies identifying mutations in Lynch syndrome associated genes have been done are more likely to have a variant of unknown significance identified by testing. This was noted as being a particular issue for this group if straight to germline testing was recommended (that is, with no testing of tumour tissue).

Approved Associate Director (name): Rebecca Albrow

Date: 26/08/2020