DG-42 Auditing the testing of endometrial cancer for Lynch syndrome at Royal Devon University Healthcare NHS Foundation Trust

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Introduction

- Lynch syndrome (LS) is a hereditary cancer syndrome predisposing patients to develop endometrial, colorectal, and other cancers
- 2-5% of endometrial cancers (EC) can be attributed to LS
- EC is often the sentinel cancer in the patients with LS
- In 2020 NICE has published a stepwise testing strategy (designated DG42) for LS in EC including mismatch repair protein (MMR) immunohistochemistry, MLH1 promoter methylation and germline testing.
- Additionally, the BAGP has published guidance to facilitate standardised reporting of the testing in EC

Aims

The primary aim of this audit was

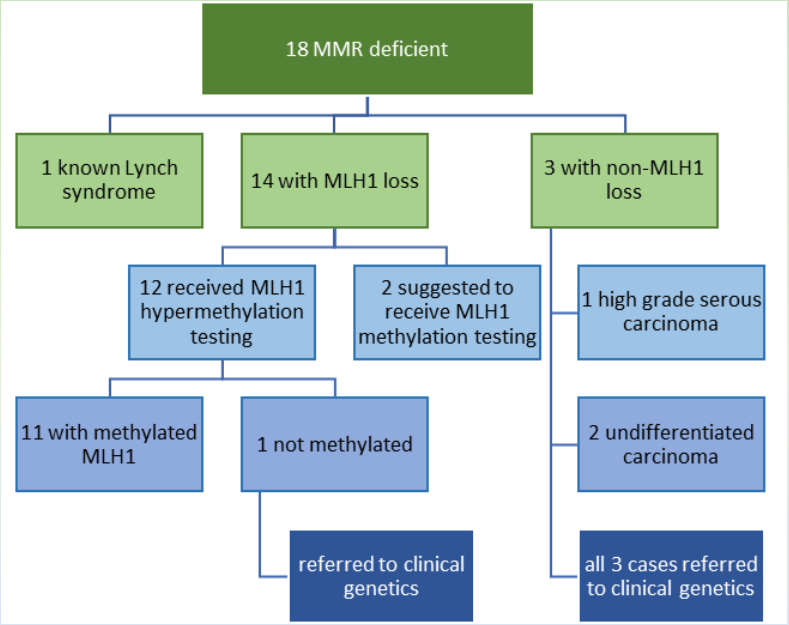
- To assess the departmental practice on LS testing pathway in EC in relation to DG42.

The secondary aims included

- Assessing the prevalence of MMR deficiencies in EC
- Assessing the prevalence of Lynch syndrome

Method

- Electronic search of the Royal Devon database of all newly diagnosed EC from October 2020 to December 2021
- Case analysis for testing for MMR deficiency, and its documentation in histology reports and MDT outcome proformas
- Analysis of cases with MMR deficiency for further testing as per the NICE protocol



Results

- 125 cases of newly diagnosed EC, 47 serous and 78 non-serous carcinoma
- 30% of cases were not tested for MMR deficiency, 32 of 37 cases were serous carcinomas
- 100% of histology reports contained MMR status, including the resection reports (MMR testing routinely done on biopsy)
- 20% of tested cases had MMR deficiency
  - 2 loss of MSH2/MSH6
    - 1 of which was known LS
  - 2 isolated MSH6 loss
  - 1 MLH1 loss without methylation
- 2 cases with no record of appropriate further testing
- 5 cases received clinical genetics referrals
  - 3 in progress
  - 1 sporadic
  - 1 lost to follow up

Discussion

- Noteworthy variability between MMR testing for serous and non-serous EC
- Thorough reporting of MMR status in histology reports
- Inconsistency in inclusion of MMR status in MDT proformas
- For those tested, NICE guidance on stepwise testing strategy for LS was followed

Recommendations

- Reflex testing should occur on all EC subtypes, including serous carcinomas and carcinosarcoma
- Documentation of MMR status could be improved in MDT proformas

References

1. National Institute for Health and Care Excellence. Testing strategies for Lynch syndrome in people with endometrial cancer London: NICE; 2020 Singh N, Wong R,
2. Tchrakian N, Allen S-G, Clarke B, Gilks B. Interpretation and Reporting Terminology for Mismatch Repair Protein Immunohistochemistry in Endometrial Cancer. The British Association of Gynaecological Pathologists; 2020.

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