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Data briefing:

Reflex testing for Lynch syndrome in people diagnosed with bowel cancer under the age of 50

Introduction

Lynch syndrome is an inherited condition that predisposes individuals to bowel and other cancers. It is caused by an alteration in a gene called a mismatch repair gene. These genes identify and repair errors that can occur in the replication of DNA. Alteration in one of these genes can lead to the development of tumours.

Lynch syndrome is estimated to cause over 1,000 cases of bowel cancer in the UK every year[i], many of them in people under the age of 50. Individuals that have Lynch syndrome have a lifetime risk of developing bowel cancer of approximately 35 per cent. Yet fewer than 5 per cent of individuals with the condition have been diagnosed in the UK[ii].

A simple screening test can be carried out to identify which bowel cancer patients are most likely to have Lynch syndrome. If this test is positive individuals are referred for genetic testing. Performing this type of screening test for Lynch syndrome on everyone diagnosed with bowel cancer under the age of 50 will help identify people at greater risk of recurrence and any family members who may also have the condition and be at risk of bowel cancer. Lynch syndrome can also affect treatment options offered to those diagnosed with bowel cancer so screening should be routinely performed at diagnosis.

A recent NHS study[iii] found that this kind of screening for Lynch syndrome in all patients diagnosed with bowel cancer under the age of 70 fulfils cost-effectiveness criteria for approval by NICE. The Royal College of Pathologists (RCPath) cancer dataset for the reporting of bowel cancer, published in July 2014, acknowledges the strong evidence base for performing Mismatch Repair Immunohistochemistry (MMR IHC) on all bowel cancer tumours. However given the resource implication of doing this the RCPath recommends performing this test on patients diagnosed with bowel cancer under the age of 50 at time of diagnosis. This is because Lynch syndrome is more common in patients under the age of 50. MMR IHC is now a core data set item for pathology and is a required reflex test for this group of young patients.

Method

Following the inclusion of MMR IHC testing for Lynch syndrome on all patients under the age of 50 diagnosed with bowel cancer in the RCPath cancer dataset, Bowel Cancer UK submitted a freedom of information (FOI) request to every trust in England, health board in Scotland and Wales and health and social care trust in Northern Ireland to establish the number of trusts/health boards that were implementing the core dataset requirement[iv]. The request was sent by email in November 2014 and asked the following questions:

1. *Are all patients diagnosed with bowel cancer under 50 years of age tested for Lynch syndrome at diagnosis within your trust?*

2. *Is Lynch syndrome testing carried out using MMR immunohistochemistry? If not, which method is being used to test for Lynch syndrome?*

3. *If testing for Lynch syndrome is not carried out, are you intending to implement the recommendation for Lynch syndrome testing? If so, by when?*

4. If *no, please could you tell us why you will not be implementing the guidelines?*

As this was an FOI request, respondents were required to respond within 20 working days.

Findings

In England, a 78 per cent response rate was achieved. There were 12 trusts to which the request was not deemed applicable to. These were specialist hospitals, such as the Liverpool Heart and Chest NHS Foundation Trust or the Moorfields Eye Hospital NHS Foundation Trust. A further 21 trusts did not respond within the 20 working days deadline for FOI requests.

In Scotland and Northern Ireland a 100 per cent response rate was achieved. Only one health board in Wales did not respond.

Country	**Contacted**	**Responded**	**Not applicable**	**Did not respond**
England	159	125	12	22
Scotland	14	14	-	-
Wales	7	6	-	1
Northern Ireland	5	5	-	-

In England, 49 per cent of NHS trusts that responded screen bowel cancer patients under the age of 50 for possible Lynch syndrome and 51 per cent said they currently do not. More than half of health boards in Wales do not screen patients. In Scotland half of health boards currently do not follow the RCPath cancer dataset. All health and social care trusts in Northern Ireland responded to say that they perform the test to identify possible Lynch syndrome patients.

Number of trusts which screen patients diagnosed with bowel cancer under 50 years of age for Lynch syndrome	**Yes**	**No**
a. England	61	64
b. Scotland	7	7
c. Wales	2	4
d. Northern Ireland	5	0

1. Trusts that do not screen all bowel cancer cases under 50 for Lynch syndrome

In England, of those trusts which did not screen patients under 50 for Lynch syndrome at the time of the request, 28 (45 per cent) were aiming to implement the recommendation or were finalising business cases and/or logistics. There were 13 trusts in which the recommendations were under discussion or due to be discussed and 10 trusts stated that they had no plans to implement screening for Lynch syndrome for under 50s.

Trusts which do not currently perform Lynch syndrome screening on bowel cancer patients under age 50:		
a.	Intend to implement this recommendation	28
b.	In discussion on whether/how to implement	13
c.	No plans to implement the recommendations	10
d.	No response	14

In the trusts that either were in discussion or had no plans to implement the recommendation, the main reason why they had not implemented testing was related to funding. Responses included:

> *"An operational meeting took place for the guidelines to be implemented, but this is on-going as there is a funding issue. This remains unresolved."*
>
> *"New testing requires funding. The lack of funding has been raised as an issue within the Bowel Cancer MDT, with the Chair of the Hospital Cancer Committee and will be discussed at the next Pathology Clinical Network Group and Bowel Cancer Clinical Network Group meeting."*

One trust mentioned the potential **impact on patients** that such a screening service could have, in terms of families and patients misunderstanding what the test means and what should be done with the information. Another trust stated they were **awaiting NICE guidance** before they would comply with the recommendation.

> *"The reasons for not screening all bowel cancer cases for Lynch syndrome are multiple, and include the cost containment as well as the unplanned impact on wellbeing that can arise out of any screening program."*

In Wales, just two out of the four health boards that do not routinely test patients under 50 stated they have plans to implement the cancer dataset. In Scotland following the publication of the RCPath cancer dataset guideline and recent publications outlining the economic benefits of testing for Lynch syndrome, the National Services Division Scotland and the Molecular Pathology Consortium are now looking at ways to deliver testing for Lynch syndrome across Scotland. This group will provide guidance and a nationally agreed protocol for implementation. Although a timescale has not yet been agreed, we believe this approach is a model of good practice.

2. Trusts that screen all bowel cancer cases under 50 for Lynch syndrome

The majority of trusts that do carry out screening for Lynch syndrome in England use MMR immunohistochemistry to perform the test. Four of the health boards in Scotland that responded to the questions stated they used MMR IHC, only one health board in Wales and all health and social care trusts in Northern Ireland performed MMR IHC to identify possible cases of Lynch syndrome. A number of trusts, in addition to IHC, also perform other tests such as microsatellite instability, *BRAF* mutation analysis and next generation sequencing.

Is Lynch syndrome testing carried out using MMR immunohistochemistry?	**England**	**Scotland**	**Wales**	**NI**
a. MMR immunohistochemistry	57	4	1	5
b. MSI	2	-	-	-
c. Unknown /DNR	2	3	1	0

3. Approaches to testing

Although screening for Lynch syndrome is being carried out in a number of trusts and health boards in the UK there is considerable variation in the pathway. The following four approaches were identified:

a. Reflex testing
b. MDT discussion ⇨IHC
c. MDT discussion ⇨Genetics ⇨ IHC
d. MDT discussion ⇨GP ⇨Genetics ⇨IHC

> *"All patients diagnosed with bowel cancer are discussed on the local lower gastro-intestinal multidisciplinary meetings (MDT) and those aged under 50 years (and selected others as outlined in the RCPath 2014 bowel cancer reporting dataset) have IHC."*

> *"Patients diagnosed with bowel cancer under the age of 50 are referred to our local genetics service."*
>
> *"MMR testing would normally be requested by the geneticist (referral advised at MDT). The Trust does not request testing directly as the Trust believes that proper counselling is needed for genetic testing and its implications."*
>
> *"Bowel MDT recommends that all patients diagnosed with bowel cancer under the age of 50 years are referred to the Oxford Genetics Service through their GP. This is communicated to the GPs by letter following the MDT. The Genetics department has then subsequently requested tissue to be sent for MMR testing after an interview with the patient and with patient consent."*

Conclusion

Our findings show that the RCPath guidelines may have already had some impact on the application of IHC testing for Lynch syndrome in patients diagnosed under the age of 50. However there is considerable variation in the approach to testing. Many trusts/health boards do not yet have in place a comprehensive, systematic reflex service as mandated in the RCPath reporting data set. A national approach to testing, as is being developed in Scotland, would provide the consistency needed to ensure that all trusts/health boards in England and Wales are performing the required reflex test for Lynch syndrome.

It is crucial that these patients are identified and go on to have genetic testing to receive a definitive diagnosis of Lynch syndrome. By systematically testing everyone under the age of 50 in this way, we can also identify families who may also be at risk of Lynch syndrome and ensure they can access regular colonoscopies. Regular colonoscopy in patients with Lynch syndrome reduces their mortality from bowel cancer by an estimated 72 per cent.

We are further concerned by the number of CCGs that did not hold any information on whether their providers are compliant with RCPath cancer dataset and perform IHC in patients under 50 with bowel cancer. As this is part of national British Society of Gastroenterology and RCPath guidance for colorectal teams, we believe screening for Lynch syndrome should be commissioned by CCGs from their local hospital trusts.

Recommendations

We believe the following recommendations will improve screening for Lynch syndrome and lead to the identification and surveillance of affected patients:

1. NHS England and Wales should adopt a similar approach to Scotland and establish a nationwide initiative to ensure a consistent, systematic approach to screening for Lynch syndrome as mandated by the Royal College of Pathologists.

2. All CCGs must commission to reflect the RCPath cancer dataset thus ensuring providers are compliant with this cancer dataset.

3. Accreditation of pathology departments should be linked to compliance with the core minimum dataset which may be used as a metric.

This data briefing was prepared by Bowel Cancer UK in February 2015. For further infomation, contact Asha Kaur, Policy Manager, at asha.kaur@bowelcanceruk.org.uk.

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[i] Screening for the Lynch Syndrome, Hampel et al, NEJM (2005) http://www.nejm.org/doi/full/10.1056/NEJMoa043146

[ii] UK colorectal cancer patients are inadequately assessed for Lynch syndrome, Adelson et al, Frontline Gastroenterology (2013), http://fg.bmj.com/content/early/2013/08/09/flgastro-2013-100345

[iii] A systematic review and economic evaluation of diagnostic strategies for Lynch syndrome, Snowsill et al (2014) http://www.journalslibrary.nihr.ac.uk/__data/assets/pdf_file/0005/125978/FullReport-hta18580.pdf

[iv] In November 2014 a request was sent to all 211 Clinical Commissioning Groups in England for the same information requested from NHS Acute Trusts. Out of 173 responses 78 per cent of CCGs said they did not hold any of the information requested. A number of trusts stated that they did not specifically commission for diagnosis of Lynch syndrome or do not specify in contracting which diagnoses should be performed and that they rely on clinical judgement within secondary care. The Department of Health Pathology Quality Assurance Review (2014) specified that the "commissioning of pathology services is, in principle, no different from commissioning of other clinical services."

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