



Together to stop bowel cancer

Time to Test



Introduction

Lynch syndrome

Each year approximately 1,200 cases of bowel cancer in the United Kingdom (UK) are caused by Lynch syndrome, making it the most common form of hereditary bowel cancer. It is also responsible for causing over 1,000 cases of other cancers, including womb, ovarian, endometrial, and brain cancers. As it is an inherited condition there is a 50:50 chance of passing the condition on through generations, which means whole families can be devastated by bowel cancer. An estimated 175,000 people have Lynch syndrome in the UKⁱ, but fewer than 5% of individuals know they have the conditionⁱⁱ. This has been attributed to the lack of molecular diagnostic testing, which can identify Lynch syndrome, being carried out across the UK.

The value and role of molecular diagnostics

Our genes have a major role to play in the development of cancer and as such, if we want to improve survival and outcomes, it is important to understand the genetic make-up of the disease. This is crucial as we know that cancer is increasingly being defined not by which body part has been affected, but by the specific gene changes that have occurred in a particular cancer. Molecular diagnostic tests can identify these changes in genes and so provide critical insights into the development of cancer.

One of these gene changes involves defects in the mismatch repair (MMR) genes. These genes are responsible for recognising and repairing damage to DNA. When there is a fault in one or more of the MMR genes, mistakes in DNA are not corrected. Over time, the accumulation of mistakes in DNA can lead to the development of tumour cells. Around 15% of people with bowel cancer are estimated to have defects to these genes and 3% of these have the genetic condition Lynch syndromeⁱⁱⁱ, which can increase the lifetime risk of bowel cancer to up to 80%^{iv}.

Testing for Lynch syndrome

Molecular diagnostic tests can identify Lynch syndrome in patients who have been diagnosed with bowel cancer. If changes to the MMR genes are detected, further genetic testing is recommended to confirm a Lynch syndrome diagnosis.

The two molecular tests that can be used to identify features of Lynch syndrome are:

- **Microsatellite instability (MSI):** microsatellites are repetitive sequences of DNA. When the MMR genes stop working properly mistakes in copying the DNA correctly can occur. This test looks for those errors, which could be a sign of Lynch syndrome.
- **Immunohistochemistry (IHC):** mutations in MMR genes can prevent the development of proteins in tumour cells. This test identifies whether there is a loss of one or more of these proteins, which suggests a defect in one of the MMR genes.

As microsatellite instability and MMR deficiency can also occur in tumours that are not linked to Lynch syndrome, further testing for changes in the BRAF V600E gene and MLH1 protein expression is necessary, as these changes are rarely seen in tumours from people with Lynch syndrome. If no changes are identified, further genetic testing is required to definitively diagnose Lynch syndrome^v.

Diagnostic testing in the UK

Clinical guidance has evolved to expand testing from those identified as having strong family history of bowel cancer, to testing specific age groups and more recently to testing all bowel cancer patients. However as health policy in the four nations is devolved, there is some variation in clinical practice and the implementation of guidelines on who should be tested for MMR defects.

England: In 2017, following analysis of clinical and cost effectiveness, the National Institute for Health and Care Excellence (NICE) published diagnostic guidance, *"DG27: Molecular testing strategies for Lynch syndrome in people with colorectal cancer"*^{vi}, recommending that everyone diagnosed with bowel cancer is tested for molecular features of Lynch syndrome automatically, at the time of diagnosis. However diagnostic guidelines are not a mandatory requirement.

Scotland: In Scotland, the Scottish Molecular Pathology Consortium (MPC) is responsible for carrying out strategic reviews of the organisation and development of molecular pathology services. Following a review, the MPC recommended that bowel cancer patients under the age of 60 are referred for testing. This is implemented through a national approach in which four regional centres – Aberdeen, Dundee, Edinburgh and Glasgow – carry out testing for all Scottish hospitals. In 2017 the MPC agreed it would be appropriate to adopt NICE DG27 in Scotland and requested that the consortium laboratories assessed the impact of adopting the recommendation.

Wales: NICE guidelines are applicable in Wales, however, there is also no mandatory requirement to implement recommendations. Health boards in Wales have a process to ensure guidelines are appropriately disseminated to inform clinical practice and service planning. There is consensus across clinical teams and the All Wales Medical Genetics Service to develop the service for Lynch syndrome testing to ensure the guidance can be implemented across Welsh health boards. A working group has been established, which is comprised of surgeons, pathologists, oncologists and geneticists. This group will make recommendations on a draft pathway for the analysis of MSI in all bowel cancer patients and funding options to deliver the service are being explored.

Northern Ireland: The Department of Health in Northern Ireland review NICE guidance and make decisions on whether it should be endorsed for use in the nation. In 2017 the Chief Medical Officer informed the charity that it is their intention to include universal testing to identify MMR deficiency as a potential service development for 2018/19. However this is

subject to funding. The political situation in Northern Ireland continues to postpone the introduction of universal testing, as there is no Health Minister for Northern Ireland.

The Royal College of Pathologists (RCPATH), which in 2014 recommended testing only bowel cancer patients under the age of 50, recently updated their guidance in December 2017 to recommend universal testing following the publication of NICE guidance. RCPATH guidance is applicable across the UK^{vii}.

Benefits of universal testing for Lynch syndrome

The value of molecular diagnostics spans the entire cancer pathway, from identifying a person's risk of developing cancer, to determining whether a patient has cancer due to a hereditary condition, or which treatments are safe and effective to use for a particular patient. As such it is an important decision-making aide in helping to personalise the diagnosis, treatment and care of an individual, and deciding what screening and monitoring a patient may need following their diagnosis of cancer.

Identifying people with Lynch syndrome has the following benefits:

- **Identifying more high risk individuals:** Testing bowel cancer patients for Lynch syndrome means we can identify those at greater risk of recurrence and also means that their family members can be offered testing to identify others at higher risk of developing bowel cancer and other cancers.
- **Enabling early diagnosis:** Once identified, individuals and their families can be placed in a screening or surveillance programme to receive regular colonoscopy. This has been found to reduce the risk of dying from bowel cancer by up to 72% because it can detect cancer at earlier stages when it is more treatable and the chance of survival is high^{viii}.
- **Informing treatment options and success:** Testing at the time of diagnosis is crucial as the condition can affect treatment options for bowel cancer. Identifying bowel cancer patients in this high risk group will ensure patients aren't offered treatment they won't respond to or may have adverse reactions to.

In addition to this, testing all bowel cancer patients for molecular features of Lynch syndrome is also a cost-effective use of NHS resources. This is because regular colonoscopy for this high risk group helps to ensure bowel cancers are detected at an earlier stage, which reduces the need for costly treatments. The tests costs just £200, less than half the price of a colonoscopy at £580^x, compared to an average cost of £25,000 for bowel cancer treatment. NICE also estimates that over 300 bowel cancers could be prevented each year through universal testing for Lynch syndrome^x. Furthermore, it can also deliver cost savings through avoiding the application of ineffectual therapies and potentially accelerating recovery.

Brief summary of the data from 2016

We conducted two Freedom of Information (FOI) requests in 2015 and 2016 to find out how many hospitals were following RCPATH guidance and discovered that patients diagnosed with bowel cancer were not routinely being tested for deficiencies in the MMR genes at the point of diagnosis of bowel cancer. The FOI results found that whilst the percentage of hospitals testing for Lynch syndrome in the under 50s had increased from 49% to 69% over the course of the year; there were still inconsistencies across the testing pathway with many not testing for the condition at diagnosis. In 2016, only 11% of hospitals were routinely carrying out molecular testing at diagnosis prior to treatment, and 56% performed the test as a reflex test (i.e. automatically at the time a pathologist makes the cancer diagnosis). The FOI identified funding, resource and capacity as being key barriers to implementation^{xi}.

As clinical guidance has moved in the direction of universal testing and away from selective testing, this report assesses whether this change had been replicated in clinical practice.

Methods

In January 2018, FOI requests were submitted to:

- Every hospital in England, Health Board in Scotland and Wales, and Health and Social Care Trust in Northern Ireland to determine whether Lynch syndrome testing is being offered to all bowel cancer patients
- Clinical Commissioning Groups (CCGs) in England to assess whether they are commissioning NICE DG27
- NHS England to provide further clarity on the commissioning responsibility for NICE DG27

Findings

Response rates

In England, 207 Clinical Commissioning Groups (CCGs) were sent the FOI request, and 204 (99%) responded. NHS England responded to the FOI request for further clarity on funding.

| Commissioners | Contacted | Responded | Did not respond |
|---------------|-----------|-----------|-----------------|
| CCGs | 207 | 204 | 3 |
| NHS England | - | Yes | - |

A total of 167 hospitals across the UK were sent the FOI request and 154 (92%) responded.

| Country | Contacted | Responded* | Did not respond |
|------------------|-----------|------------|-----------------|
| England | 141 | 130 | 11 |
| Scotland | 14 | 14 | 0 |
| Wales | 6 | 5 | 1 |
| Northern Ireland | 5 | 5 | 0 |
| Total | 167 | 154 | 14 |

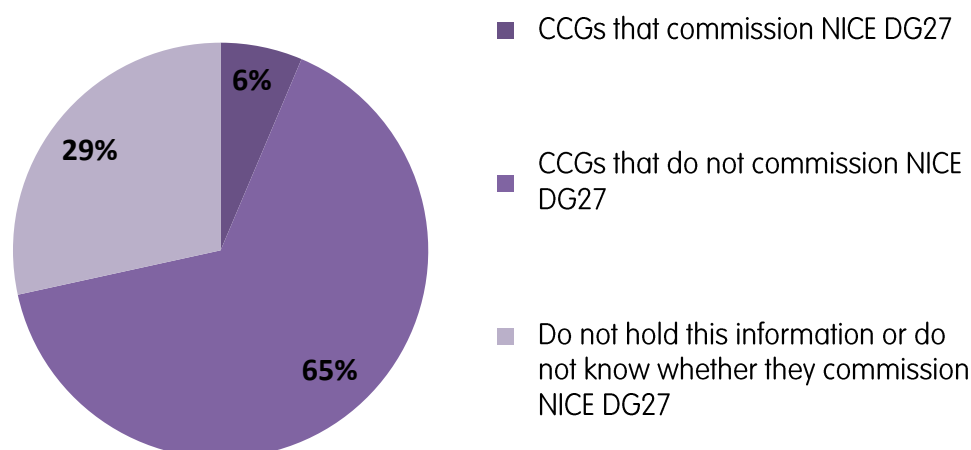
*Two specialist hospitals in England were excluded from this table as the FOI request was not applicable. Powys Health Board in Wales was also excluded as patients are referred to other health boards or hospitals in England.

In England a 92% response rate was achieved, which is an increase of 9% from 2016. An 83% response rate was achieved in Wales, which is lower than the 100% response rate received in 2016, as one Health Board did not respond. In Scotland and Northern Ireland a 100% response rate was achieved.

England

Clinical Commissioning Groups

Figure 1: Proportion of CCGs commissioning hospitals to implement universal testing, in line with NICE DG27



In England, 13 (6%) out of 204 CCGs commission their local hospital(s) to test all bowel cancer patients in line with NICE guidance. Most CCGs (65%) stated they do not commission Lynch syndrome testing in all newly diagnosed bowel cancer patients, while the remaining 29% responded saying they either do not know what services are provided by their local hospitals or do not hold this level of information.

The main reasons CCGs provided for not commissioning or not knowing whether they commission NICE DG27 are:

- **Directing responsibility elsewhere:** They do not consider it their responsibility and comes under the remit of another commissioning body or programme (e.g. NHS England or the Bowel Cancer Screening Programme); or are awaiting information on whether molecular testing will be nationally funded (29%).

"Specialist clinical genomic services, including cancer genetics, are not commissioned locally but instead form part of the Prescribed Specialised Services commissioned by NHS England."

"No, we do not intend to commission DG27 – A new Bowel Scope Screening programme is soon to be rolled out, where everyone aged 55 years and over will be offered a flexi sigmoidoscopy."

- **They commission the whole cancer pathway:** They do not hold information on specific tests, but expect their local hospitals to be implementing NICE guidance (29%).

"We do not specify the service provision to this level of detail for cancer services though we do have a general understanding with the trust that they apply relevant NICE guidance."

- **Developing business cases and planning implementation:** They are still in the planning phases and/or are assessing the implications for commissioning DG27 (10%).

"No, we do not commission this test. The number of patients that will require testing will increase from 170/year to 1,700. A business case will be developed during 2018. We await information regarding national funding for the increase in molecular testing."

"The proposed guidelines will have a significant impact on the current service. The recommendations, as they are, will considerably increase the workloads to all Histopathology departments in England... This highlights that increased funding would be required and the extra workload that would need to be managed for Pathology, Clinical Genetics, Cellular Pathology, Genetics Labs, Endoscopy and other members of the multidisciplinary team (MDT)."

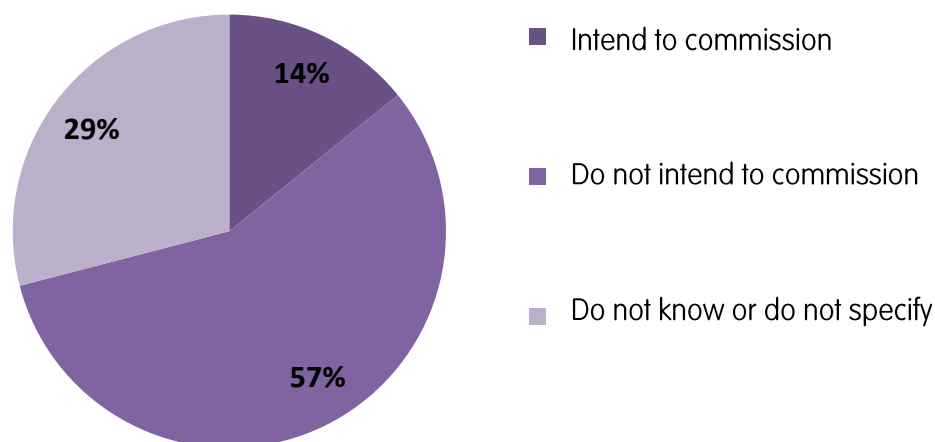
- **Assessing the value and cost of testing for their local areas:** They are duplicating NICE processes and assessing cost-effectiveness of the guidance themselves (7%)

"... It was felt that in a situation of limited staffing and resources, testing all patients would be expensive and of limited benefit in a population."

"... Risk has been assessed as low if the guidance is not in place as it does not alter the treatment and/or procedures for this cohort of patients."

The remaining 25% do not specify their reasons for not commissioning NICE DG27, or do not hold the information.

Figure 2: Proportion of CCGs that intend to commission universal testing, in line with NICE DG27



Of the CCGs that do not commission testing in all newly diagnosed bowel cancer patients, over half (57%) have no intentions to commission this, 29% have intentions to commission, and the remaining 14% do not know or have not specified.

NHS England

To gain further clarity on commissioning responsibility, we submitted an FOI to NHS England. They responded to our request with:

“There is no national commissioning policy for the tests outlined above. NHS England does not provide additional financial support specifically for the implementation of NICE guidance DG27. This testing should be commissioned through local budgets by CCGs given that cancer testing is based on local tariff arrangements.”

Hospitals across the UK

Table 1: Proportion of hospitals, within each nation, that carry out universal testing

| Country | Yes, all bowel cancer patients | Testing under 50s (under 60s in Scotland) | Testing based on family history only | Do not test |
|------------------|--------------------------------|---|--------------------------------------|-------------|
| England | 35% (45) | 48% (63) | 11% (14) | 6% (8) |
| Scotland | 43% (6) | 50% (7) | 7% (1) | 0 |
| Wales | 0 | 40% (2) | 60% (3) | 0 |
| Northern Ireland | 20% (1) | 80% (4) | 0 | 0 |
| Total | 34% (52) | 49% (76) | 12% (18) | 5% (8) |

Of the 130 hospitals that responded to this request, 45 (35%) test all bowel cancer patients for molecular features of Lynch syndrome. Almost half of these hospitals (48%) are still testing bowel cancer patients under the age of 50, with another 11% testing based on family history of the disease only (i.e. case by case basis). In addition, 30 (35%) hospitals have no intentions to introduce testing in accordance with NICE guidance.

A range of responses were provided from hospitals in England to explain why they had not implemented universal testing. This includes the additional cost and increase in resource required to implementing universal testing, a lack of clarity on funding and commissioning arrangements, in process of submitting or awaiting the outcome of a business case or that they have a good system in place already to detect Lynch syndrome in bowel cancer patients:

"Although a full business case was not submitted, extensive enquiries were made with the CCG and with NHS England about funding for the test. As all answers came back in the negative, an agreement was made for limited and not full testing at this centre."

"We are aware of the recent NICE recommendation. A consultant in genomic medicine from the regional genetics service has been attempting to secure central funding. We have not submitted a local business case in light of this ongoing discussion."

"Awaiting guidance from the network as currently we have been advised there is not enough capacity to process all tumours for Lynch syndrome testing."

"We have operational plans regarding how we can introduce testing both in the short and longer term, however, we require clarification regarding who will be commissioning this new service."

"It is being addressed by CCG and discussed through the commissioning group to address funding issues. All of our appropriate clinical teams are aware of this testing and of the current status in relation to financial arrangements being discussed with the aim that implementation will follow once agreed."

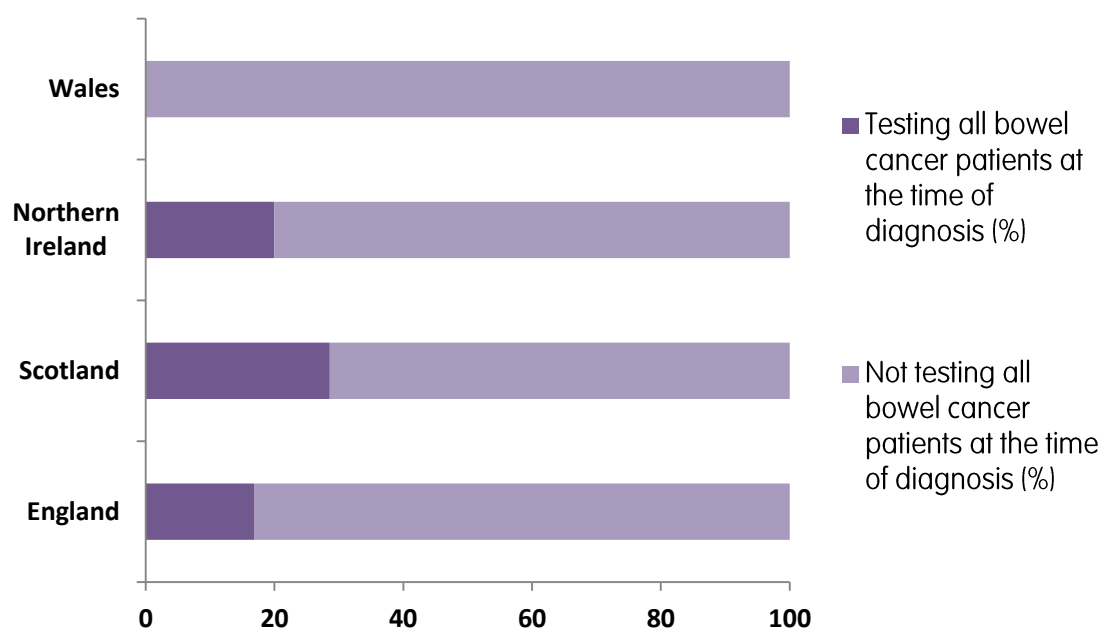
"We have had discussions and have delayed making a decision at present. We feel we have a well-functioning Lynch diagnostic service at present and do not feel an urgent clinical need to extend screening to all patients, particularly as the cost would be excessive with relatively limited benefit."

In Scotland, 6 (43%) out of the 14 health boards either test or refer all bowel cancer patients for features of Lynch syndrome. Of the remaining health boards, 7 (50%) follow Scottish guidance to test only those under age 60, and one (7%) tests based on family history only. In Northern Ireland, one Health and Social Care Trust (20%) has implemented universal testing. Wales is the only nation not testing all bowel cancer patients in any of their health

boards; however, three health boards (60%) have intentions to introduce testing in line with NICE guidance.

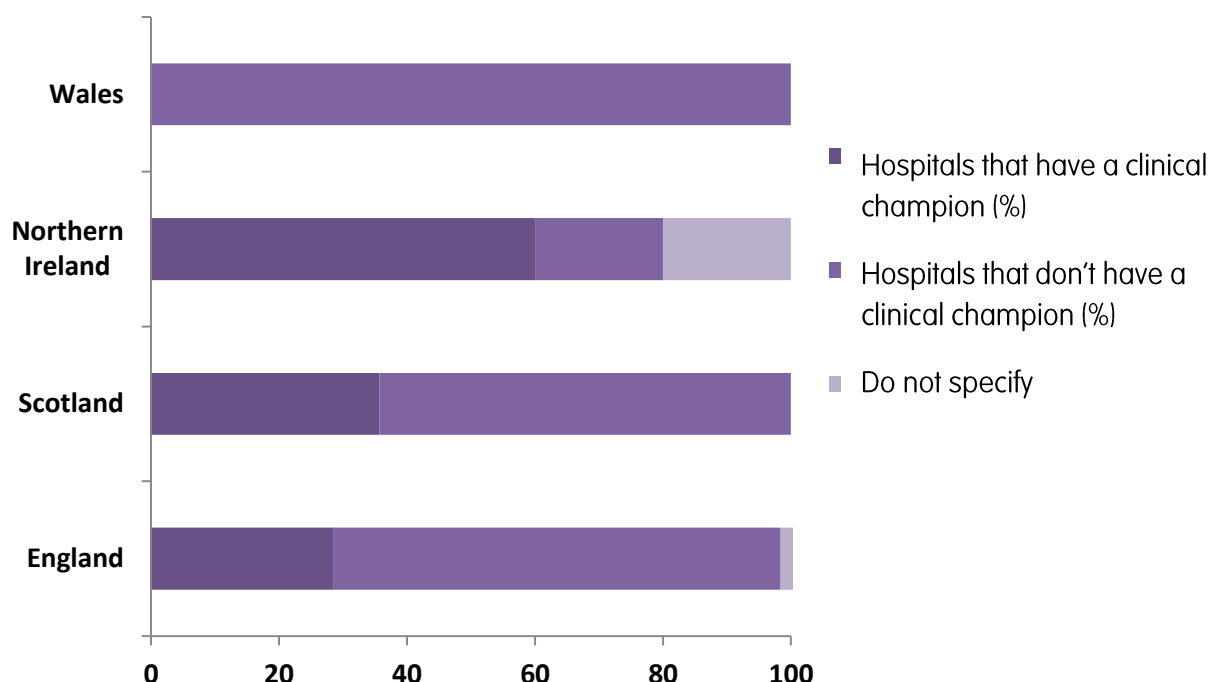
"The All Wales Genetics Service has Welsh Health Specialised Services Committee funding for MSI analysis in (a) patients and families who meet our guidelines for Lynch syndrome, and (b) colorectal cancer when chemo is being considered. The department plans to introduce testing of all patients in line with the NICE guidance as soon as possible within the next few months."

Figure 3: Proportion of hospitals implementing best practice, i.e. testing all bowel cancer patients at the time of diagnosis, within each of the four nations



Although 45 (35%) hospitals in England test all bowel cancer patients for molecular features of Lynch syndrome, only 22 (17%) test at the time of diagnosis (i.e. pre-treatment). Therefore, the remaining 108 hospitals (83%) are not implementing best practice for Lynch syndrome testing, in line with NICE guidance. Similarly in Scotland, only four health boards (29%) test at the time of diagnosis and 20% of Health and Social Care Trusts in Northern Ireland. Wales is the only nation that does not have any health boards testing in line with best practice.

Figure 4: Proportion of hospitals, within the four nations, that have a 'clinical champion'



Only 37 (28%) hospitals in England have identified a 'clinical champion' to oversee implementation of universal testing and drive forward improvements in care pathways. 36% of health boards in Scotland and 60% of HSCTs in Northern Ireland have identified a clinical champion. Wales is the only nation that has not identified any clinical champions.

Table 2: The named clinical champions

| | England | Scotland | Northern Ireland | Total |
|----------------------------------|----------|----------|------------------|----------|
| Colorectal Surgeon | 17 (46%) | 2 (33%) | - | 19 (41%) |
| Oncologist | 10 (27%) | 1 (17%) | 1 (33%) | 12 (26%) |
| Gastroenterologist | 5 (14%) | - | - | 5 (11%) |
| Histopathologist | 3 (8%) | - | - | 3 (7%) |
| Pathologist | 2 (5%) | 2 (33%) | 2 (37%) | 6 (13%) |
| Clinical Geneticist | 3 (8%) | 1 (17%) | - | 4 (9%) |
| Clinical Nurse Specialist | 2 (5%) | - | - | 2 (4%) |

***Note:** some hospitals had two clinical champions, so these were recorded twice under the respective clinical champions.

Colorectal surgeons were the most commonly identified 'clinical champion' within each colorectal multidisciplinary team across England (46%) and Scotland (33%). In Northern Ireland, pathologists (37%) made up the highest proportion. Oncologists (England 27%; Scotland 17%; Northern Ireland 33%) were also commonly chosen to be the 'clinical champion' to effectively implement testing for molecular features of Lynch syndrome. Wales is not represented in Table 2 above as none of their health boards have identified a named clinical champion.

Table 3: The main barriers to testing all bowel cancer patients for Lynch syndrome within each of the four nations

| Main barriers to testing | England | Northern Ireland | Wales | Total |
|--------------------------------------|----------|------------------|---------|----------|
| Financial | 69 (91%) | 3 (60%) | 2 (50%) | 74 (87%) |
| Staff resources | 46 (61%) | 1 (20%) | 1 (25%) | 48 (56%) |
| Awareness of current guidance | 13 (17%) | 0 | 0 | 13 (15%) |
| Policy | 11 (14%) | 1(20%) | 1 (25%) | 13 (15%) |
| Patient Consent | 2 (3%) | 0 | 0 | 2 (2%) |

***Note:** In Scotland, no health boards recorded any major barriers to testing due to the centralised system of testing bowel cancer patients.

Across the whole of the UK, finances were cited as the main barrier to implementing universal testing. Of the hospitals that responded to this question, 69 (91%) in England, 3 (60%) in Northern Ireland and 2 (50%) in Wales listed this as the main barrier to testing. 'Staff resources' was another common obstacle to testing, particularly in England, where 61% of hospitals listed this as a barrier to implementing NICE guidance. 'Policy', i.e. patients tested in line with local/regional policies, was also listed as a barrier.

Below are examples of hospitals' responses that highlight barriers to testing:

"Even with implementation, the main barriers have been, and remain, financial and staff resources. NICE produce guidance but the government do not provide the funding for the implementation of the guidance. It is a recurring theme and with the increasing use of genomic medicine is a major problem for pathology departments."

"Funding relies on NHS England proposing commissioning guidelines (policy and service specification) which would then drive CCGs to fund this. We have received no updates at present regarding any funding from the CCG."

"Lack of clarity regarding funding of test, lack of time and awareness of diagnosing practitioners, lack of nursing staff to support pre-test counselling, lack of policy and pathways to support routine testing."

Discussion

Identifying Lynch syndrome in patients diagnosed with bowel cancer using molecular diagnostics results in clear benefits to the patient, their family members and the NHS. Both in terms of preventing and detecting cancer early but also the cost-savings that this can deliver for a cash strapped NHSⁱⁱⁱ. These substantial benefits make it necessary that more of the estimated 175,000 Lynch syndrome gene carriers in the UK are identified. In 2016, we recommended NICE publish a positive recommendation on universal testing and that it stipulates a clear referral pathway on who to test, when to test and what test to use in

order to reduce the variation in testing approaches. But despite the known benefits of universal testing and the clarity provided by clinical guidance, our findings show that there is still variation in testing strategies and expansion of testing to all ages has been slow, with local NHS bodies and Governments still assessing the implications of universal testing to their health economies.

Accelerating progress

To date, both Scotland and Wales have agreed in principle to adopt universal testing. It is positive that almost half of the health boards in Scotland have expanded testing to all patients, despite current guidance only recommending testing those under 60. However, this must be rolled out to all health boards in Scotland. An update to the national consensus guidance would accelerate adoption. In Wales, local health boards are responsible for delivery of NICE guidance but our FOI results have consistently shown year after year health boards have struggled to implement testing, whether it was previous RCPATH guidance to test under 50s or more recent NICE guidance to test everyone. The situation in Wales has remained much the same, with little progress made. The Welsh Government committed to assess the implications of NICE guidance, with a view to implementing a national system of testing, similar in operation to Scotland. This would be a welcome move. However, how and when this will be implemented, and which body will be responsible remains unclear.

Who's responsible?

In England one issue in particular has hindered progress, and that is commissioning responsibility. It is clear from responses from NHS England that there is no national commissioning policy for these tests and that this should be commissioned through local budgets by CCGs. The Health Minister's response to a Parliamentary question provided further clarification that the "commissioning of services for people diagnosed with Lynch syndrome, including surveillance, generally sits with clinical commissioning groups (CCGs). CCGs are best placed to commission services to meet the needs of local populations, taking into account the best available evidence."^{xiii} This includes implementation of DG27.

However, 65% CCGs reported that they do not commission the guidance and nearly a third of these cited that this is because it is not their responsibility but either the responsibility of NHS England, Public Health England, or that these individuals should be tested as part of the bowel cancer screening programme. These responses are incredibly concerning. The lack of local responsibility and national leadership has consequently affected adoption of clinical guidance. Caught in the middle are hospitals, unable to carry out universal testing and patients who are at high risk of bowel cancer.

Part of CCG's confusion may be due to the split in commissioning that currently exists in England; while genetic testing is funded nationally by NHS England, the molecular diagnostic test is funded locally. Greater clarity on funding and commissioning

arrangements in England is needed, as nearly 60% of CCGs have no intention to commission DG27. Health services would benefit from a funding and commissioning pathway at the time of publication of NICE guidance to ensure clarity of responsibility.

Mainstreaming genomics

In addition, plans from NHS England to mainstream genomic medicine in the NHS by consolidating regional genetic centres and the development of a national single directory of tests is contributing to this confusion. To date no public announcement has been made on future funding arrangements for molecular diagnostic tests and whether these tests will be eventually nationally commissioned. For some CCGs, these national plans have stalled local implementation.

The genetics procurement process due to conclude in October 2018 would present a good opportunity to provide clarification and resolution of the commissioning and funding process. If NHS England wish to mainstream genomic medicines in the NHS, ensuring that there are processes, procedures and resources in place to govern the implementation of molecular diagnostics is necessary. This is because molecular diagnostics play a crucial role in guiding referrals for genetic testing and for the selection of treatment that will have the most positive impact on outcomes.

However, until the conclusion of this programme of work CCGs, hospitals and cancer alliances must work together to ensure all bowel cancer patients are tested, at diagnosis, for defects in their MMR genes and that clinical and laboratory teams are fully engaged with the task of delivering this service and that implementation is regularly audited.

Communicating guidance

To encourage adoption of universal testing, education and training on Lynch syndrome, why testing is important, the benefits to both patients and local NHS services is crucial. But NICE and the NHS also has a role to play in ensuring wide dissemination of their guidance, with many CCGs unaware of the guidance and its importance to identifying individuals at high risk of bowel cancer. Currently NICE communications on guidance is limited to publication on their website and reliant upon media to increase awareness. More could be done to ensure wide dissemination of guidance and of commissioning responsibility.

Clinical champions have a vital role in providing local clinical leadership to oversee service delivery and ensuring pathways for patients are implemented. However, only 29% of hospitals reported having a champion in place. If implementation is to be successful, local health bodies must work together to share best practice, develop ways of working, and identify barriers and local solutions to overcoming them. This is essential to ensure that responsibility is taken at a local level.

Upfront cost, long-term savings

We recognise that there is an upfront cost and resource implication to realise testing. But there must also be recognition that this cost is offset by the longer term savings that can be made further down the care pathway. No additional funding for the implementation of NICE guidance DG27 has been provided but additional capacity is needed in laboratory services to process testing, in genetic services to provide adequate follow-up and counselling of patients, and in endoscopy units to provide colonoscopic surveillance. These are issues that we have highlighted in our first assessment in 2015 and again in 2016^{xiv}. It is clear that little progress has been made to better support and enable hospitals to implement best practice.

Conclusion

There is a clear case for universal testing and each nation must ensure adoption of this is accelerated in their health systems and that investment is made to ensure there is the capacity to deliver the pathway. Molecular diagnostics has the power to transform the way we diagnose and treat patients but new technologies such as next generation sequencing is the future of genomic medicines and will allow us to sequence genes quickly and cheaply, as well as better understand the genetic variants of cancer. However the system and structures to deliver it must enable rapid and widespread adoption, so all cancer patients can benefit.

Recommendations

England

NHS England must rapidly establish national protocols and funding to ensure testing can be carried out on all bowel cancer patients. In the interim CCGs, hospitals and cancer alliances must work together to implement NICE guidance.

Scotland

The Scottish Government must accelerate the development of a national consensus on universal testing and subsequent implementation.

Wales

NHS Wales must identify funding within their existing budgets to kick start universal testing and provide national clinical leadership to drive forward implementation of testing.

Northern Ireland

The Department of Health and Health and Social Care Board must accelerate adoption of NICE guidance and ensure sufficient funding and capacity is available.

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- ⁱ National Institute for Health and Care Excellence (2016), Diagnostics consultation document: Molecular testing strategies for Lynch syndrome in people with colorectal cancer
<https://www.nice.org.uk/guidance/GID-DG10001/documents/diagnostics-consultation-document>
- ⁱⁱ Snowsill T. et al (2016), Molecular testing for Lynch syndrome in people with colorectal cancer, Peninsula Technology Assessment Group (PenTAG), University of Exeter Medical School (Report for NICE).
<https://www.nice.org.uk/guidance/dg27/documents/diagnostics-assessment-report>
<https://bowelcancerorguk.s3.amazonaws.com/Final2016DataBriefingLynchsyndrome.pdf>
- ⁱⁱⁱ Gatalica, Z et al. (2016). High microsatellite instability (MSI-H) colorectal carcinoma: a brief review of predictive biomarkers in the era of personalised medicine, *Fam Cancer* 15: 405 – 412.
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4901118/>
- ^{iv} UK colorectal cancer patients are inadequately assessed for Lynch syndrome, Adelson et al, *Frontline Gastroenterology* (2013)
- ^v National Institute for Health and Care Excellence (2016), Molecular testing for Lynch syndrome in people with colorectal cancer: Final scope, <https://www.nice.org.uk/guidance/dg27/documents/final-scope>
- ^{vi} National Institute for Health and Care Excellence (2017), Molecular testing strategies for Lynch syndrome in people with colorectal cancer <https://www.nice.org.uk/guidance/dg27/resources/molecular-testing-strategies-for-lynch-syndrome-in-people-with-colorectal-cancer-pdf-1053695294917>
- ^{vii} Royal College of Pathologists (2017), Dataset for histopathological reporting of colorectal cancer
<https://www.rcpath.org/resourceLibrary/g049-dataset-for-histopathological-reporting-of-colorectal-cancer.html>
- ^{viii} Prevention of colorectal cancer by colonoscopic surveillance in individuals with a family history of colorectal cancer: 16 year, prospective, follow up study, Isis Dove Edwin et al (2005) *BMJ*
- ^{ix} Snowsill T. et al (2016), Molecular testing for Lynch syndrome in people with colorectal cancer, Peninsula Technology Assessment Group (PenTAG), University of Exeter Medical School (Report for NICE).
<https://www.nice.org.uk/guidance/dg27/documents/diagnostics-assessment-report>
- ^x Ibid
- ^{xi} Bowel Cancer UK (2016), *Data Briefing: Reflex testing for Lynch syndrome in people diagnosed with bowel cancer under the age of 50:*
<https://bowelcancerorguk.s3.amazonaws.com/Final2016DataBriefingLynchsyndrome.pdf>
- ^{xii} National Institute for Health and Care Excellence (2016), *Diagnostics consultation document: Molecular testing strategies for Lynch syndrome in people with colorectal cancer*
<https://www.nice.org.uk/guidance/GID-DG10001/documents/diagnostics-consultation-document>
- ^{xiii} Written parliamentary question, *HC Deb*, 28 February 2018
<http://www.parliament.uk/business/publications/written-questions-answers-statements/written-question/Commons/2018-02-20/128627/>
- ^{xiv} Bowel Cancer UK (2016), *Data Briefing: Reflex testing for Lynch syndrome in people diagnosed with bowel cancer under the age of 50:*
<https://bowelcancerorguk.s3.amazonaws.com/Final2016DataBriefingLynchsyndrome.pdf>