Improving the identification and management of people with genetic conditions that increase the risk of bowel cancer
Tuesday 15 March 2016
Webb Johnson Hall, Royal College of Surgeons

Executive Summary
Around five per cent of all bowel cancer cases are caused by genetic conditions, such as Lynch syndrome or familial polyposis, conditions, which increases the lifetime risk of developing bowel cancer and some other cancers by as much as 80 per cent. However evidence indicates that the identification and surveillance of individuals with genetic conditions varies considerably across the UK. To seek to address this variability a new collaborative venture that links the ACPGBI, BSG, RCS, NCRI CSG, BDRF and Bowel Cancer UK was set up. Our ambition is to develop a quality assured surveillance programme for people at high risk of bowel cancer through the development of a national database, which would also facilitate participation in valuable bowel cancer research.

A meeting that brought together 35 of the leading clinicians and researchers in the field, as well as patients and gene carriers was held to discuss how individuals can be better identified as having a genetic condition, including which tests and when, and how individuals identified with a genetic conditions can be better managed to prevent and detect bowel cancer early. This document is a summary of the discussion on the day.

The following recommendations have been agreed by the group to improve the identification and management of those with genetic conditions:

Recommendations
- Develop clear guidance to simplify and streamline referral pathways for patients. The guidance should focus on who to test, when they should be tested, and the appropriate assessments which should be carried out to identify patients at high risk, and to establish nation-wide practice in this regard.
- Establish a database of people who have been identified with genetic conditions. This must be a systematic process with patient consent obtained by a clinician at the point of diagnosis.
- Establish a national surveillance and/or screening programme for people with genetic conditions to improve patient outcomes.
- Each hospital must have a dedicated single lead for genetics to oversee service delivery and ensure pathways for patients are instituted.
- Quality assurance standards must be developed. This would ensure guidance and pathways for the identification and management of those with genetic conditions are followed.
- Improve education for healthcare professionals on the importance and value of genetic testing.
- Develop high quality information for patients with genetic conditions or suspected genetic conditions that is tailored to a diverse audience and made available on a number of platforms.
Introduction

The meeting brought together 35 of the leading clinicians, scientists, researchers, patients and gene carriers with an expert interest in hereditary bowel cancer and screening. The aim was to reach a consensus on what the current issues and gaps are in the identification and management of people with genetic conditions and to agree what next steps can be taken to improve the identification and management of people with genetic conditions.

Specifically the meeting aimed to:

- consider the best methods of identifying and testing people who may have a genetic condition that increases their risk of bowel cancer;
- develop practical solutions to ensure optimal management of people who are known to have a genetic condition that increases their risk of bowel cancer;
- consider the creation of a national genetics registry as a method of developing a national surveillance screening programme for people at high risk of bowel cancer; and
- identify possible research questions

Around 5-10 per cent of all bowel cancer cases are caused by known genetic syndromes, which increase the risk of bowel cancer – these include familial adenomatous polyposis (FAP) and Lynch syndrome. The risk of someone with either of these conditions developing bowel cancer in their lifetime is very high – nearly 100 per cent and 80 per cent respectively. Yet, research from Bowel Cancer UK shows that Royal College of Pathologists’ recommendation - that everyone diagnosed with bowel cancer under the age of 50 years be tested for the Lynch syndrome - is inconsistently implemented across the UK.

Individuals with these genetic conditions should be managed through a surveillance programme that ensures they have regular colonoscopies to help reduce their risk of developing bowel cancer. Appropriate regular surveillance reduces mortality by 81 per cent in “moderate risk groups” and by 72 per cent in patients with Lynch syndrome. However, there is currently an inconsistent approach the management of people with genetic conditions linked to an increased risk of bowel cancer. This is a result of poor clinical and public awareness, patients not being seen quickly enough, and a failure to provide adequate follow up. An inconsistent approach to managing people at higher risk of bowel cancer undermines efforts to save lives.
Breakout session: How to better identify people with genetic conditions and meet needs for information?

This break out session involved four topics of discussion that explored issues relating to current practice in identifying people with a genetic condition. These were:

1. Which test should be used;
2. Which the most appropriate referral pathway
3. What are the support and information requirements of patients
4. What are the research questions that could help to improve the way people with genetic conditions are identified

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Breakout Group 1

**Question:** What pathway should be followed to identify people with possible genetic conditions linked to a high risk of bowel cancer?

**Facilitator:** Dr Kevin Monahan

**Members:** Mr James Hill, Professor Huw Thomas, Lindy Berkman, Anne Mackie, Dr Roland Valori, Tim Elliott, Dr Bianca de Souza, Dr Farhat Din

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**Introduction**

This breakout session examined the process of identifying those who should be referred to testing; including asking if the current guidance effectively accomplishes this. Existing best practices were identified and consideration was given to how these could be incorporated nationally and locally.

**Discussion**

Current pathways to identifying gene carriers (once suspicion arises) are variable and ad hoc, and there are normally multiple entities involved. For example, at Central Manchester University Hospitals, patients are referred straight to the genetics team, at St. Mark’s Hospital and at West Middlesex University Hospitals patients are asked to complete a questionnaire to determine whether individuals require assessment by the genetics team. The Royal Marsden has a system of mainstreaming breast and ovarian patients with a genetic condition i.e. asking non-genetics clinicians such as oncologists to undertake assessment and testing, which may be useful to consider when developing a pathway for genetic conditions linked to bowel cancer.

Three key steps were noted in the pathway for identifying and caring for people with genetic conditions linked to a high risk of bowel cancer:

**Step one:** Getting the right people through the system is crucial. The primary routes to identification of gene carriers are:

- Multi-Disciplinary Teams (MDTs)
• General Practitioners (GPs)
• Gastroenterology & endoscopy (e.g. those with a high rate of polyps)
• People diagnosed with cancers at a young age
• People in the system known to be at risk
• Identifying people through cascade testing of relatives
• Public entry through websites & social media, having completed a risk assessment tool that covers all cancers.

**Step two:** A formal risk assessment should take place. Achieving this would require agreeing the questions clinicians should ask patients to assess risk.

**Step three:** A quality assured surveillance programme is required to manage the patients identified with a genetic condition that places them at high risk of bowel cancer.

The following questions were posed, which may, if answered, help to improve our understanding of the referral pathway for identifying people with a genetic condition:

- Could one online accredited risk assessment tool for all cancers be developed?
- Should we be more proactive in cascading testing to relatives of known individuals with germline mutations?
- Should we extend somatic tumour testing for Lynch syndrome to older patients; and if so how can it be ensured that testing is then actually done for this expanded group?
- Should we supply information packs for GPs? Do we need to incentivise GPs (e.g. with QOF points)?
- Should public awareness be raised?
- Are family history questionnaires reliable?

**Recommendations to be considered**

1. Referral pathways, national and locally, should have clear, accredited guidance and pathways to reduce variation. (Where possible steps and processes should be simplified. Reviews should take place to ensure that unnecessary steps are removed.)
2. Where local pathways are developed, we must ensure that patient identification, testing and follow-up conforms to nationally agreed minimum standards.
3. The implementation of a surveillance program should begin with patients known to have a genetic condition linked to an increased risk of bowel cancer and then be expanded these principles to include other high risk groups, e.g. IBD, post-colorectal cancer surveillance.
4. A registry/surveillance program should meet public expectations. This could include having online tools for public engagement.
5. It must be clearly determined who needs to see a genetic counsellor i.e. before genetic testing, or just after a test which is not normal?
6. A single lead within each colorectal MDT should have oversight of service coordination and delivery within their institution, with support from regional genetic services.
7. Steps should be taken to look at best practices of different groups.
**Breakout Group 2**

**Question:** Which test should be used to identify people at higher risk of bowel cancer and when? Is further research required to establish this?

**Facilitator:** Mr Doug Speake

**Members:** Dr Ian Frayling, Professor Phil Quirke, Professor Ian Tomlinson, Dr James East, Professor John Burn, Dr Andrew Latchford, Dr Sunil Dolwani, Dr Fiona Laloo

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**Introduction**
This breakout session examined the strengths and weaknesses of existing tests to identify people with genetic conditions linked to an increased risk of bowel cancer, best practices for testing, and the group discussed how to identify people for testing.

**Discussion**
There are a number of points that should be taken into consideration when deciding what test should be used to identify people at higher risk of bowel cancer. Currently the Royal College of Pathologists guidelines recommend reflex testing for patients under the age of 50 with NICE guidelines on molecular testing for Lynch syndrome currently in development. However there are many different perspectives across the pathway—oncological, surgical and pathological that should also be taken into account.

Additionally testing for genetic conditions will impact, benefit and influence decisions along the patient pathway. The advantages of both pre-operative testing and post-operative testing need to be considered fully, although all of the above are subject to NHS funding which is, at best, limited.

There are a number of tests currently available for genetic conditions:

- MSI
- IHC
- BRAF
- Next Generation Sequencing

However there is not consensus on which of these tests is the best test to use to identify people with genetic conditions. To reach a consensus the following research questions would need to be addressed in order to provide clarification on which test to use to identify those with genetic conditions and when in the pathway to test:

- **Pre-operative testing vs post-operative testing:** There is no reliable evidence or uniform agreement on testing a biopsy vs whole tumour testing and whether testing preoperatively would change surgical decision making? Testing preoperatively (on a biopsy) is ideal as knowing if a patient has Lynch syndrome or another genetic condition may impact surgical decisions, including in some cases whether to carry out a full colectomy. This may lead to discussions for extaintestinal prophylaxis (i.e. Gynae). However surgical decisions cannot be based solely on MSI/IHC testing. It
is known that 2-3 per cent of patients do not present with microsatellite instability. Additional testing would be needed to capture these individuals.

- **Cost-effectiveness:** The health economic assessment report is now outdated because the cost of genetic testing is falling. This now needs updating to take this into consideration.
- **Sequencing:** What is the role of sequencing both pre and post-operatively, and how should/could data be interpreted? (This will include cascade screening relatives once predisposition identified in patient with CRC).

Testing for genetic conditions will involve: MSI-IHC +BRAF testing. However the ideal would be for genetic sequencing pre or post op.

**Universal testing vs age restrictions**

With regard to age and testing – essentially there are two options - universal testing and age restricted testing (in line with current Royal College of Pathology guidance).

Testing all patients would be easier and simpler to implement rather than age restricted testing. There is also a Health Technology Assessment (HTA) which has found that testing up to age 70 is cost-effective. However there may be funding and resource issues in implementing this. Universal testing will require additional money in the short term, but can save money over the long term. However, the NHS is keen that any new initiatives are in-year cost neutral. With this in mind, ensuring all under 50s are being tested first, while developing a strategy around universal testing, may be the most sensible and simple approach. It was noted that the recommendation to limit immunohistochemistry testing to the under 50s was made due to limited budgets and resources. It was suggested that this decision may not be the best for patients.

**An automated pathway**

To ensure a smoother pathway for patients both testing (including how and when) as well as sharing of results with clinicians at all points in the pathway should be automated throughout.

**Recommendations for next steps**

1. **Involvement of Oncologists.** Oncologists arguably have a more powerful voice than pathologists in influencing policy and establishing NHS-wide best practices. If oncologists take the position that genetic testing is needed to effectively determine treatment, this will carry much weight in establishing nation-wide practice.
2. **Education of healthcare professionals** - including pathologist, gastroenterologist and surgeons - in genetic testing is essential. It needs to be determined what training should happen and timelines established.
3. **More must be done to ensure MSI/IHC testing is conducted for bowel cancer patients under the age of 50 by ensuring clear reflex pathways for clinicians to follow.**
4. **Gather more evidence to support the benefits of panel screening for all bowel cancer patients in the future, taking cost implications into account.**
**Breakout Group 3:**

**Question:** What information and support do people at risk of having a genetic condition need, when do they need it and from whom?

**Facilitator:** Nicola Fearnhead

**Members:** Miss Asha Senapati, Hayley Heard, Joe Higgins, Professor John Northover, Richard Gardner, Dr Pauline Skarrott, Andy Sutton, Azmina Verjee

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**Introduction**

This breakout session explored what information is available for patients and clinicians on genetic conditions and identified what gaps exist. Consideration was then given to discussing the quality of this information, and best practices in information sharing were identified, along with recommended next steps.

**Discussion**

The primary sources of information on genetic conditions that increase the risk of bowel cancer include: nurse specialists; doctors; other healthcare professionals; family members; NHS websites; charity websites; other health websites; and social media. There is little printed material in circulation and there are concerns there is a lack of information provided by the NHS on Lynch Syndrome. This highlights the need for more, and better, quality information and resources for healthcare professionals, health organisations, those who have been newly diagnosed and those who are high-risk due to family history of having a genetic condition.

Informing family members of someone who tests positive for a genetic condition is dependent on patients. Privacy laws do not allow healthcare professionals to convey the information without patient consent. It is therefore crucial that effective information and support is provided to patients with genetic conditions to assist them in talking to family members who should be tested. Having more information and materials in circulation that are science- and evidence-based will help counter much of the misinformation that is found online. For example, chat rooms can be an excellent way for patients to get support and information but there is also incorrect information and advice being passed through social media and chat rooms.

**Best practices in the provision of information**

- Health communication and social marketing materials need to be tailored to target a wide-range of groups, taking into account variables such as age, ability to comprehend, culture and language skills.
- Specific attention should be focussed on teenagers in known Lynch syndrome families where parents may be involved in influencing decisions prior to age of consent.
- Informational materials and websites must be of a high quality and there should be effective processes put in place to ensure updates are made to get the latest information and advice out to the public.
• Most informational materials should focus on providing the key facts to avoid people becoming overwhelmed and confused but there is also demand for in-depth literature for those seeking a more comprehensive understanding of the genetic conditions linked to bowel cancer.

• Support through the testing process is important and should be provided by nurses and other healthcare professionals with specialised training.

• Specialist nurses are best placed to provide information to patients and families but too few have an in-depth knowledge of genetic conditions linked to bowel cancer. More should be done to improve training but it was acknowledged that nurse specialists do not come across cases of genetic conditions linked to a high risk of bowel cancer on a regular basis. It may not be feasible to fully train and educate all nurses.

• Systems need to be put in place for healthcare professionals that support clear and simple signposting of high risk patients to specialist nurses who have training on genetic conditions.

• It was noted that at present, testing is most often being done after the tumour is removed but there is a strong argument for testing to take place before surgery. Testing should not delay treatment but is important early on as it can inform treatment options.

Recommendations to be considered:

1. Conduct research to gain an understanding of existing support and information available to patients/the public.

2. Conduct research to determine the availability of information for healthcare providers.

3. Determine the extent and level of training available to healthcare providers to support patients with a genetic condition and individuals at risk of having a genetic condition.

4. Consult with patients and healthcare providers to determine what information and additional training is needed.

5. Establish a working group to develop a project plan aimed at filling the information and training gaps.

6. Gain an understanding of good practices already in place across the UK with regard to providing care and support, including best practices being implemented by genetics counsellors.
Breakout Group 4

Question: Is there further research that, if carried out, could help us better identify people with genetic conditions or new genetic conditions?

Facilitator: Dion Morton

Members: Professor Richard Houlston, Professor Mark Hull, Dr James Buchanan, Professor Robert J C Steele, Professor Wendy Atkin, Professor Peter Sasieni,

Introduction
This breakout group examined the existing gaps in knowledge relating to identifying people with known genetic conditions and the discovery of new genetic conditions. The group then identified research opportunities that would address gaps and improve identification.

Discussion
There is a need to shift the funding emphasis to encourage greater research at the front end of the cancer pathway, on secondary and tertiary prevention (rather than advanced disease). At present, there is a significant diversity of surveillance practice because of the lack of good quality research. Moreover, the national ‘100,000 Genomes Project’ is likely to impact significantly on the types of research we will be able to do. The National Screening Committee is currently recruiting for a new chair and we will want to make sure that the new chair will be supportive of moving the bowel cancer genetic research agenda forward.

Recommendations for next steps
1. Further investigation should be taken into whether having a dedicated nurse/named individual to identify high risk groups has a positive impact on identification (RFPB funding?).
2. Efforts should be made to build a stratified approach to screening by identifying ‘high risk’ groups in two ways:
   o Focus on the ‘polyp formers’ – e.g. detailed investigation on patients identified with recurrent adenomas (family history and molecular studies)
   o Focus on identifying the lower-risk groups and seek to remove them from the population receiving invasive surveillance (endoscopy) follow up.
3. There is a need to set up a complex, multi-modal, interventionist study involving multiple cancer centres exploring the benefits/disadvantages of different pathways. The study could be iterative and include looking at the benefits of pre-op testing of colorectal cancers.
4. Efforts should be directed to developing new non-invasive tests – a big issue is how to reduce the number of people who drop out of regular surveillance because of the invasive nature of regular colonoscopies.
5. A longitudinal study should be undertaken involving creating surveillance data base linked to medical records.
Breakout session: How can we better improve the management of people with genetic conditions?

The afternoon session’s presentations and breakout group discussions focused on the management of genetic conditions (including current UK practice), the development of a national genetics registry, the establishment of a national surveillance screening program, and research opportunities linked to data collected from a national genetics registry.

Breakout Group 1

Question: Local processes: MDTs and genetics clinics – how could this work better?

Facilitator: Kevin Monahan

Members: Professor Gary Middleton, Dr Pauline Skarrott, Andy Sutton, Azmina Verjee, Dr Andrew Latchford, Dr Sunil Dolwani, Dr Bianca de Souza, Dr Farhat Din and Pauline Skarrott

Introduction

This breakout group explored current practices and explored their advantages and disadvantages. The discussion then focused on identifying best practices and considering how these could be implemented nationally and locally.

Discussion

Contact with the CNS is of great importance to patients who have been diagnosed with bowel cancer. However there is currently a lack of good patient information to give to Lynch syndrome patients and the management of surveillance of those with Lynch syndrome is more challenging than for those with IBD. This is because there is lack of clarity around how these patients should be followed-up. Additionally, most patients will not remember follow-up guidance that is given immediately after colonoscopy due to sedation.

Every hospital should have a lead as well as a named contact or department for the patient and consideration should be given to a specific CNS/genetic counsellor/other role to liaise with people who have genetic conditions. Lynch syndrome patients are being bumped down the waiting list for surveillance scopes and they should not be on the 16 week pathway. The 2010 BSG/ACPGBI guidance for follow-up is not implemented in a joined up way. There is a significant need for a co-ordinated follow up across the UK. Further consideration should be given to defined lists for people with genetic conditions. Managing patients together in a registry setting – 2/3 per region, or in each local colorectal MDT site could be considered to rectify this.
Recommendations for next steps

1. Establish a multi-modal approach to addressing issues to improve outcomes.
2. Make it a requirement of each hospital to have a lead clinician for the management and coordination of the hereditary cancer patient service.
3. Develop a link role (CNS, genetic counsellor or other member of the colorectal MDT) as a point of contact for patients, and embed this standard as part of KPI’s.
4. Evidence-based care bundle/passport for patient-centred care which would help patients in their care pathway, e.g. negotiation with their GPs who may have little experience of their condition.
5. Develop quality assurance standards for primary & secondary care and co-ordinated screening pathways.
6. Develop a database of people who have been identified with genetic conditions that increase risk of bowel cancer.
Breakout Group 2

Question: National Processes: should we develop a national genetics registry for conditions leading to a high risk of bowel cancer and if so how can this be achieved and funded.

Facilitator: Miss Nicola Fearnhead

Members: Professor Wendy Atkin, Professor Peter Sasieni, Dr Fiona Laloo, Professor John Burn, Tim Elliott, Dr Jem Rashbass

Introduction
This breakout group explored which genetic conditions should be included in a registry or a national database, what the first steps should be to develop one and what funding opportunities are available.

Discussion
There is strong support for the establishment of a national registry for genetic conditions leading to a high risk of bowel cancer. However the group discussion suggested that the term ‘registry’ may not be best suited to the progress of the project and perhaps “central linked database” would be a better term to use in moving forwards.

The following should be considered when establishing a registry:

- Patient consent would be mandatory before adding personal details to a database.
- A systemic process would be best for adding newly-diagnosed patients to a registry. This process may be more effective if the responsibility of requesting patient consent is added to a list of required duties for clinicians.
- A suitable point to seek patient consent would be upon diagnosis as leaving the responsibility of joining the database to individual patients will result in fewer uptakes, especially by communities with low levels of literacy, low income communities, people with learning disabilities, etc.
- Consideration should be given to each hospital assigning a lead clinician who would be responsible for ensuring pathways for data collection are being followed. Perhaps this could be incorporated into KPIs – key performance indicators.
- From the point of data entry there will be a need for an established process of clear escalation pathways.
- If there is useful data derived from the database or if there are other positive outcomes, this will naturally support the input of information into the database.
- Thought needs to be given to the consequences of the database holding this information (e.g. implication for insurance).
- The need for a database is supported by a need for different types and elements of research, including clinical delivery research. However, while the database will be used for research there should be a multipronged strategy with a clear quality improvement pathway.
- New therapies coming in the future, such as vaccines, may be a driver for the database.
Recommendations for next steps:

1. Consideration should be given to designing an app or other ways of getting information out to patients who have joined the registry and improving services to patients.

2. Engaging clinical nurse specialists will be integral to the success of the database. There will need to be an educational pathway for CNS’s.

3. It was advised not to proceed by launching a pilot but instead to incorporate a database for genetic conditions into existing databases held by NHS i.e. the existing cancer registry.

4. Funding will need to be found to cover costs associated with establishing a data system with recall capabilities. Recall is essential to ensure adequate surveillance for individuals at high risk.

5. It was suggested that the process begin with a defined group (perhaps those with a genetic mutation that puts them at the highest risk) and then expand/roll out to all those who had genetic testing.

6. There should be steps taken to learn from places that have succeeded at establishing a registry and evaluate based on outcomes.

7. Applications should be made for grants to put the 6000 known Lynch carriers into a registry. Data would be pseudonymised and later, patients can be identified and consent can be requested. It was noted that in the past there has been much success when requesting patient consent to be included in a centrally-linked database.

8. Success in obtaining grant funding is more likely if the proposal is not a stand-alone pilot but instead deliverable within existing services.

9. Consideration should be given to working in collaboration with other groups with genetic conditions linked to a high risk of cancer – e.g. ovarian cancer.
**Breakout Group 3**

**Question:** How can we develop a national surveillance screening programme for people with genetic conditions leading to a high risk of bowel cancer?

**Facilitator:** Doug Speake

**Members:** Dr James Buchanan, Professor Robert J C Steele, Hayley Heard, Joe Higgins, Professor John Northover, Dr James East, Lindy Berkman, Anne Mackie, Dr Roland Valori

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**Introduction**

This breakout group explored what main issues need to be addressed in establishing a national surveillance screening programme, and considered whether a national approach should be taken and who should lead this initiative. The barriers to developing a programme were examined and best practices were identified.

**Discussion**

Currently responsibility for surveillance screening of high risk groups falls to local providers. This has led to local inequalities and a ‘postcode lottery’- some areas do it really well, while others do not. This leads to many high-risk patients being marginalised in favour of others. A national approach to surveillance screening would need:

- The identification of all at-risk at the national level;
- A system/database/registry of individuals considered high-risk;
- A call and recall system;
- An identified team in each region to take responsibility for service;
- Quality assurance mechanism – this could be delivered by JAG

The Bowel Cancer Screening Programme (BCSP) has all of the above in place already for the asymptomatic population. The Programme is run and delivered to a very high standard, it has robust quality assurance mechanisms already in place and it has a good system for the call and recall of patients. However, for various reasons the screening programme may not be able to deliver on this.

An additional system, Redcap, a cloud based system could be used to ‘call and recall’ patients both directly and each identified clinical lead in each region. High quality screening colonoscopy can still deliver quality endoscopy out with screening programme. Regional genetics centres and charities should also be leading on this.

**Potential barriers to implementing a national screening programme are:**

- Funding of registry and recall
- Determining who should be the body responsible for implementation, quality assurance and standards. The BCSP is a population screening programme for ‘well’ people. It is not Public Health England’s responsibility to look after these patients but the responsibility of the NHS. Furthermore,
the surveillance of patients is a wider problem and not just one for those at high risk of bowel cancer. The BCSP needs to retain its credibility. If the BCSP takes on Lynch syndrome patients then there may be requirements to take on other genetic cases too. The Breast Screening Programme has seen problems with agreeing on the definition of high risk, as well as identifying those at high risk. There is a strong possibility that the same would happen for bowel cancer as well.

- National variation exists in the screening programmes across the UK therefore we may not be able to take a UK-wide approach to this.
- Ability to identify all patients at risk within a registry with capacity to recall and capture data.
- The funding of the colonoscopy screening for high risk patients with a genetic condition should not be a barrier to the BCSP taking on the surveillance of these patients as the programme does not currently fund colonoscopies.

**Recommendations for next steps**

1. Establish a mechanism of identification of at-risk individuals and funding of a national registry which has the capacity to recall, monitor and demonstrate quality assurance.
2. Establish who will deliver high quality screening (ideally NHS service delivery as per screening).
3. Further explore the options for a national surveillance screening programme and to also explore funding models for this.
Breakout Group 4

Question: What are the research opportunities linked to improving access to a national surveillance screening programme?

Facilitator: Mark Hull

Members: Professor Richard Houlston, Professor Dion Morton, Miss Asha Senapati, Dr Ian Frayling, Professor Phil Quirke, Professor Ian Tomlinson, Mr James Hill, Professor Huw Thomas

Introduction
This breakout group identified the key research questions that a national genetics registry could help answer and facilitate, and explored what would need to be built into the registry to allow for this. Research priorities were also identified research priorities.

Discussion
- To determine whether a national surveillance programme would lead to an earlier stage of diagnosis and improved patient outcomes.
- Explore the impact of different Quality Assurance models.
- Explore the impact of different interventions in randomised trials – e.g. surgical, chemoprevention and lifestyle changes.
- Explore colorectal cancers with microsatellite instability (MSI) and analyse germline and somatic mismatch repair gene mutations.
- Explore what impact having people with Lynch syndrome has on a national surveillance screening programme and hence the implications for the inclusion of other high risk groups.
- Better explore the connections with other cancers and health conditions.
- Better explore the impact of endoscopies on high risk groups and linkage with the proposed national endoscopy database.
- Compare/contrast with other countries which have a similar surveillance programme (e.g. Denmark).

Recommendations to be considered
1. The case is already made for having a national registry/surveillance screening programme for Lynch syndrome gene carriers but we should continue to explore whether there is additional research we should do to develop the case further and whether a better case can be made for other high risk groups.
2. How do we ensure consent for inclusion in a national surveillance screening programme research database?
Final Summary
(Professor Dion Morton)

A multi-pronged and multi-model strategy is required to improve outcomes for high risk patients. How to engage the clinical workforce has to be the link to the delivery of this. An educational pathway needs to be established and this should be embedded as part of their clinical training and KPIs. Patient groups have an important role to play in this.

Clear escalation pathways are needed. Each hospital should have a lead clinician for genetics to ensure pathways for patients are in place. This includes clear deliverables for MDTs.

A surveillance recall programme is essential and will require funding. Charities can have a role to pump prime and engineer transformation.

We need different topics of research to deal with complex problems:

1. Clinical delivery research
2. Qualitative research
   a. What happens if we have a molecular diagnosis earlier in the pathway (pre operative diagnosis)? This has implications for the acceptability to patients and family
   b. What are the barriers in clinical service that could be unblocked?
3. Stratified screening into high risk and low risk groups

In conclusions, we need three things to happen:

1. Pathway – consensus, so patients know what to expect
2. Registry – this is the key to surveillance
3. Research