

IBD BioResource genetic feedback

a conversation between
participants and researchers



Executive summary

December 2023

Introduction

Research into the genetics of common, complex conditions generates findings that are both pertinent to the subject of the study and those related to a range of other genetic conditions.

On November 20th 2023, 15 NIHR IBD BioResource participants, 6 gastroenterologists and 3 genetics researchers joined members of the Wellcome Connecting Science Engagement and Society team at the Wellcome Genome Campus in Cambridgeshire. The purpose of the workshop was to inform a helpful and reassuring process for how the NIHR IBD BioResource shares genetic feedback related to additional findings with the study participants who have opted in to receive it, and to provide learnings for the wider field of genomic research into common, complex conditions.

Additional findings are not those directly related to IBD, but to a range of other specific, rare, and treatable genetic conditions that could be identified in the course of genome sequencing. These conditions are drawn from the Genomics England 100,000 Genomes project list of additional findings and include bowel, breast and ovarian cancer pre-disposition as a result of single gene mutations and familial hypercholesterolaemia.

The one day workshop involved a mix of presentations, Q&As and facilitated small group discussions and activities. **Wellcome Connecting Science** worked with the **Sanger Institute Human Genetics team** and the **NIHR IBD BioResource** to commission **Hopkins Van Mil** to design facilitate and report on the workshop.



Participants' perspectives

Participants hopes for receiving additional feedback as part of the IBD BioResource programme were that:

- The process starts with re-consent for receiving additional findings because participants may well have forgotten that they had signed up to receive them;
- It increases personal health knowledge, in a tailored way;
- The feedback sharing process is managed by a central resource;
- There is a clear timeline for the feedback process;
- Additional genetic conditions could be included in future feedback from the IBD BioResource and that more broadly, genomic health screening becomes mainstream in the NHS.



I'd like to see a future where everyone gets access to this screening information, not just those who are signed up to a BioResource.

BioResource participant



Participants concerns about receiving additional feedback included:

- Health system capacity and connections: can NHS services cope with the numbers of people receiving information about a genetic risk? Is the IBD BioResource planning to share feedback in partnership with the NHS? Will support be available when results are shared?
- How feedback information will be shared: will risk of a genetic condition be communicated clearly and without causing alarm, how will false positives be dealt with and what if someone has already been diagnosed with the condition on which they have just received feedback?
- Consequences of receiving feedback: how receiving genetic information would affect health insurance for them or family members, potential impact on people's mortgages or employment, would feedback go on medical records?
- Data: how secure is it, who has access?



Key findings for the future

Discussions at the workshop generated the following important considerations for the design of the additional findings feedback process:

1. Updates on the condition of concern: IBD

Returning additional genetic feedback to IBD BioResource participants should go hand in hand with providing an update on the status of the IBD BioResource programme and its findings. Helping to prevent, treat and cure inflammatory bowel disease are the main motivations for joining the study.

2. Managing expectations for what genetic feedback is available

Some workshop participants had the attitude of ‘tell me all, even if not treatable, so I can keep any eye on things’. There needs to be clarity on the limited number of genes being screened for and why these genes were chosen.

3. Where can the process be tailored to individual circumstances?

Participants are hopeful that choice will be designed into the feedback process. For example, choosing to receive feedback, but at a later date; choosing to only receive feedback on some genes/conditions and not others.

4. Be mindful of the interest in bowel cancer as part of the 13 genes

The additional genetic findings will include conditions that are of particular interest to people with IBD, especially bowel cancer. Particular care will be needed on how this is communicated to a patient population some of whom may have had parts of their colon removed or have been told there is a link between their specific type of IBD and bowel cancer.



IBD surgery may mean people have had their colon removed. It's important to personalise information. Does that mean they're not at any risk? People may disregard an 'increased risk' finding if it isn't explained properly.



BioResource participant

5. Use a range of tools to make the information accessible

Feedback that promotes comprehension through providing contextual information on the conditions, visual aids to explain risk and supporting materials to educate people on uncertainty. Include risk ‘benchmarks’ so that results are comparable with the general and IBD populations.

6. A process that doesn't leave participants waiting anxiously for results and follow up

Participants expect each stage to be conducted within weeks of each other rather than stretched over several anxiety-inducing months, with support in between provided through website FAQs and a helpline.

7. Information received from the most appropriate sources

The IBD BioResource is seen to be the most appropriate information provider for re-consent and no additional findings. For contact on the need for a confirmatory blood sample the NHS – acknowledging the link to the IBD BioResource – is most appropriate as this carries trust and provides the bridge between research and clinical findings. For the return of genetic results for a variant, the NHS speciality should be the lead communicator.

8. A clear appetite for more involvement with the IBD BioResource in the future

During the workshop conversations IBD BioResource participants demonstrated an appetite for more involvement in its work and also in deciding what is researched in the future.



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The most inspiring aspect of the event was that non-clinical guests (like myself) felt comfortable to share experiences and thoughts without feeling looked down upon, this made the day much more engaging and enjoyable.

BioResource participant

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Each aspect of the workshop drilled down on what was most important to our patients, and how all our hopes, fears and aspirations for IBD research outcomes fit in the 'real' world of the NHS.

Clinician

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I felt privileged to have the opportunity to have an open discussion with both patients and gastroenterologists, the former is something I do not have the chance to experience often enough.

Scientist

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Full report at wellcomeconnectingscience.org/project/ibd-bioresource-and-genetic-feedback



Watch the film about the event at vimeo.com/wcsethics/IBDBioResourceConvo

The workshop was commissioned by Wellcome Connecting Science, in partnership with the NIHR Inflammatory Bowel Disease (IBD) BioResource and the Human Genetics team at the Wellcome Sanger Institute and was designed and facilitated by Hopkins van Mil.

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