Report to UK National Screening Committee, Sciencewise and UKRI

Evaluation of a public dialogue on wider genomic sequencing for cystic fibrosis newborn screening and how it may be approached

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URSUS CONSULTING LTD

www.ursusconsulting.co.uk

57 Balfour Road

London N5 2HD

United Kingdom

Tel. 07989 554504

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GLOSSARY OF ACRONYMS

CF	Cystic Fibrosis
CF SPID	CF Screen Positive Inconclusive Diagnoses
FMCH	Fetal Maternal and Child Health reference group
GEL	Genomics England
HVM	Hopkins Van Mil
NBS	Newborn Screening
NHS	National Health Service
PHE	Public Health England
SG	Steering Group
UK NSC	UK National Screening Committee
WGS	Whole Genome Sequencing

1. Introduction

This short evaluation report has been prepared by URSUS Consulting Ltd for the UK National Screening Committee (UKNSC), Sciencewise and UK Research and Innovation (UKRI) for a mini public dialogue on wider genomic sequencing for cystic fibrosis (CF) newborn screening and how it may be approached. The work is an extension to a larger dialogue on whole genome sequencing (WGS) for newborn screening (NBS) undertaken for UK NSC and Genomics England (GEL). Both dialogues have been delivered by Hopkins Van Mils (HVM), specialists in public dialogue.

1.1 Context, framing of the dialogue and the participants' sphere of influence

The UK NSC is a Scientific Advisory Committee that provides independent advice to Ministers and the NHS on all aspects of national population screening. This mini dialogue was intended to help the Fetal, Maternal and Child Health (FMCH) reference group of the UK NSC to decide whether and how to introduce more extensive genome sequencing as a test for Cystic Fibrosis. The FMCH has already undertaken a one-year test programme carried out in Sheffield, which applied more extensive testing to some 70,000 live births a year. The test screened for a wider panel of mutations or 'gene glitches' from the current 50 to about 400. The test programme findings, reported to FMCH in January 2020, showed that by looking at a wider range of mutations it was technically possible to carry out this more detailed screening within 2-3 days, and at reasonable cost. The tests could help to identify more babies with 'true CF' and could also reduce (but not totally eliminate) the need for repeat tests. The wider panel of mutations tested for would also better reflect the range of mutations relevant to the ethnic diversity of the UK population. At a national level this might mean reducing the number of repeat tests from 300 currently to about 50 a year, with associated savings which would largely offset the additional costs of the tests. It could also avoid reporting probable carrier status of babies.

However, the test programme also threw up an issue which UK NSC considered needed further consideration: in identifying as many 'true CF' cases as possible the test could also identify many more CF Screen Positive Inconclusive Diagnoses (CF SPID) cases (increasing from the current 20-30 a year to 80) depending on where the threshold for detection was set (the so-called sensitivity or specificity of the test). CFSPID is a 'designation' rather than a diagnosis: based on current knowledge most individuals receiving the CF SPID label do not go on to develop CF symptoms and therefore currently there is no treatment pathway other than periodic check-ups in a CF or respiratory clinic. The choice of a sensitive or specific test involves a trade-off in the number of 'true CF' versus CF SPID cases detected.
FMCH were therefore keen to understand the view of three different groups of stakeholders as follows:

1. People with a personal experience of CF, CFSPID or carrier status and their families.
2. Healthcare and other professionals involved in CF screening and follow-up care and support; and
3. People drawn from a broad demographic across society.

This dialogue focuses on the third category and was expected to seek the public's views on:

- How the proposal to use expanded genetic testing in the newborn screening programme for CF is viewed by different people and groups.
- What the anticipated impacts on families and the NHS would be of avoiding or including the reporting of carriers and CF SPID cases with CF newborn screening.
- How participants make trade-offs between ensuring that no 'true CF' cases are missed versus potentially causing uncertainty or false alarms (i.e., sensitivity vs specificity) and how this is viewed by different people and groups.

There was an opportunity to extend the ongoing Sciencewise/UKSNC/GEL public dialogue on WGS for NBS to undertake this mini public dialogue with a much smaller group of public participants who already had been through the previous process and therefore had both some understanding of screening, genome sequencing and cystic fibrosis and a clear understanding of the deliberative nature of public engagement. This allowed the dialogue to be undertake more rapidly and at much lower cost than if it had been commissioned independently. Engagement with other stakeholders (groups 1 and 2 above) has been tendered and commissioned separately: it will be delivered by the appointed researchers between October 2021 and September 2022, building on the findings of this process.

1.2 Objectives of the mini dialogue

The agreed objectives, shared with participants throughout the process were the following:
How should wider genomic testing be used when screening for cystic fibrosis at birth?
How sensitive or specific should the test be in the context of newborn screening?
In contrast to the previous WGS for NBS dialogue the objectives for this process were tightly framed around a single condition and two distinct approaches to extended testing from a panel of 50 to 400 mutations. A small Steering Group (SG) set up for the dialogue agreed that the choices about how this might be done should be presented as a binary choice between a more sensitive specific test – where more cases of both 'true CF' and CFSPID would be identified - or a more specific test - where fewer cases of both 'true CF' and CFSPID would be picked up. While acknowledging that this did not capture the full complexity of the situation, it was agreed that simplifying the choice would help participants on their journey towards indicating a preferred approach and their underlying reasoning for that choice. Participants were made aware that their sphere of influence would be to feed in their views alongside those of patients and their families and those that care for them to inform the UK NSC about the issues it should pay attention to as it decides in future if, and how, an extended test should be rolled out.

1.3 Layout of this report

The rest of this report is according to the following layout:

- Chapter 2 describes the methodology for the public dialogue design and delivery and for the evaluation.
- Chapter 3 describes how far the dialogue met its objectives, the likely policy impacts of the process and how participants felt about taking part.
- Chapter 4 describes how well the design and delivery met emerging best practice standards for delivery of online dialogues; and
- Chapter 4 summarises the lessons learnt and recommendations.

2. Methodology

The dialogue was designed and delivered by Hopkins Van Mils (HVM) and overseen by a small steering group (SG) bringing together seven individuals drawn from UKNSC, Public Health England (PHE), and academics working on CF. All steering group members had been involved in the WGS for NBS dialogue and so were very familiar with public dialogue and in working with Sciencewise, HVM and the independent evaluator. The work was commissioned in late April, with design during May, workshops held online during June and July and the final report shared with UKNSC in late September.

2.1 Dialogue design

Recruiting participants

HVM recontacted all 130 public participants from the WGS for NBS dialogue and invited anyone who was interested in taking part in this follow-on process to express an interest. From the 88 who expressed an interest, 21 were selected and, following two dropouts, 19 took part.

The participants were selected to give a sample that reflected UK demographics (gender, socio economic groups and disabilities) but weighted towards younger people (under 40s) who were closer to having families and facing real life decisions about screening of newborns. The selection was also skewed to include a larger number of individuals from Black and minority ethnic backgrounds, since a more extensive screening test is expected to give more accurate CF diagnosis for individuals from some ethnic backgrounds which are not served well by the current test. The participants were also selected to give a range of attitudes – both positive and negative – about wider WGS for NBS. Participants received a thank you payment of £100 for their participation in four hours of workshop discussions and completing two homework tasks.

Design and stimulus materials

HVM prepared a welcome pack and stimulus materials which were reviewed and signed-off by the Steering group. Having taken part in the previous dialogue, all participants came to the deliberations with a prior knowledge of newborn screening, cystic fibrosis, and genetic testing. The welcome pack described the dialogue objectives – stressing how it differed from the previous one - ways of working, agendas for the two sessions and a glossary of terms. It also included links to the previous dialogue videos on screening and CF so that participants could recap as a pre-workshop homework task. HVM, the SG and specialist contributors worked to co-develop PowerPoint presentations which would reinforce and add to participants' understanding of CF and CFSPID. The focus of each session, stimulus provided to participants and topics covered in discussions is shown in *Table 2.1*.

Involvement of Specialists

Three specialists, including two SG members, participated in both workshops. They were carefully briefed before the sessions by HVM. They presented different aspects of CF screening and answered participants questions in plenary and in small groups.

- Professor Jim Bonham provided more detail on how CF is screened for now, why wider genomic sequencing is being considered and the two choices being considered – sensitive or specific testing.
- Professor Iolo Doull presented the clinical implications of wider genomic sequencing and the implications of receiving or not receiving a CF diagnosis in terms of treatment and health outcomes.
- Professor Felicity Boardman shared insights from her research on the family implications of wider genomic sequencing, and of receiving a CF diagnosis or dealing with uncertainty of no diagnosis or a CF SPID label.

Before the second workshop the specialists also assisted HVM to provide answers raised by participants during and after workshop one (in their evaluation feedback).

Table 2.1: Design and stimulus materials

No. and length of sessions	Focus	Issues explored and deliberated	Specialist speakers
Pre-workshop homework: (18th-22nd June)	**Recap on CF and newborn screening**	Read participant pack and background information on CF and Newborn screening drawn from WGS for NBS dialogue	n/a
Workshop 1 (Tuesday June 22nd 6-8.15pm) **19 participants**	**Understanding the proposed new genomic testing and its implications**	Presentations and Q+A sessions on: • Newborn screening; Cystic Fibrosis; Current Cystic Fibrosis newborn screening & testing process. • clinician considerations: how the potential new tests may affect diagnosis, treatment, care & research – what does it mean to miss a case or give a CF SPID diagnosis. • family considerations: how the potential new tests and what they find may affect the newborn, their parents and wider family.	Jim Bonham Felicity Boardman Iolo Doull
Homework (June 23- July 5)	**Socialising the deliberations by discussing with friends and family**	Speak to 2-3 friends/family members/colleagues about the implications raised by these new potential tests: • A more specific test that reduces the number of both CFSPID and true CF cases identified • A more sensitive test that picks up a higher number of both CFSPID and true CF cases.	n/a
Workshop 2 (Tuesday 6th July 6-8 pm) **18 participants**	**Developing & finalising views on preferences & considerations between specific and sensitive tests**	• Informal presentation sharing responses to questions posed & follow up questions • Small Group Discussion Part 1: Considerations • Small Group Discussion Part 2: prioritising considerations in small groups; presentations in plenary • Response from UKNSC & what will happen next	Jim Bonham Felicity Boardman Iolo Doull

Capturing participants inputs, analysis, and reporting

Participants' thoughts, questions and recommendations were fully captured using a combination of notetaking by facilitators, recorded transcriptions, eVoting polls (Mentimeter) and homework tasks, such as talking to friends and family, between sessions. Participants were asked to email or record a short film of what they talked about for sharing with the team. All of this data was coded using NVivo software. The HVM team analysed this data in preparing a short dialogue report from which the findings emerged. The findings presented are qualitative and use the convention of 'a few,' 'some,' 'many' to give a sense of how widely views were shared. Anonymised participant quotes from the transcripts are used to illustrate the findings. The final outputs of the dialogue process were:

- **A short dialogue report** aimed at the commissioners and stakeholders involved with the UK NSC's screening committees.
- **An executive summary** suitable for a wider audience.
- **A PowerPoint slide deck** which can be used to present the process and findings to other audiences; and
- **A set of stimulus materials** which may also help inform the engagement with other stakeholders.

2.2 Evaluation Methodology

The evaluation was light touch but included both formative and summative elements. The evaluator worked closely with the SG, HVM and Sciencewise to provide formative feedback during the design and planning stages via project management meetings and materials review. The evaluator participated in online workshops with participants and stakeholders and evaluated the design, delivery, and outcomes of the process through observation against an agreed evaluation protocol and feedback collated from two short online surveys (SurveyMonkey) completed by participants after events. The full feedback is shown in *Annex A*, and data and quotes from participants, specialists and wider stakeholders are included *(in blue italics)* to illustrate evaluation points.

3. Outcomes and impacts

The draft final report was shared with the Steering Group and specialist contributors in early September. The report was structured to provide a clear description of the overall findings and the participant journey, including anonymised quotes from participants to illustrate the reasoning underlying their choices. SG members and the member of the Cystic Fibrosis NGS Active Advisory Board found the findings a useful input to their decision-making process. They also reported that it highlighted lessons about how engagement due to take place in 2021-22 with two other groups – patients and their families and those that treat and support them – should be framed. The findings from all three groups will be presented to the UK NSC and will help to inform their decision and recommendations to the NHS about how CF screening is rolled out. The findings from looking in depth at this single condition will also be shared with Genomics England to help inform its proposed roll out of whole genome sequencing for newborns.

3.1 Meeting the dialogue objectives

The objectives for this dialogue were narrowly focused on understanding how wider genomic testing should be used, and understanding participants' preferences between two options for applying the test as follows:

- A more sensitive test which would avoid missing 'true CF' cases but at the risk of identifying more CFSPID cases (60 to 80 a year compared to the current 25): and
- A more specific test which might miss some 'true CF' cases (less than 10 a year) but also identify many less CFSPID cases.

Participants fully understood the objectives and their sphere of influence

The objectives were explained to participants at workshop one and restated at workshop two. All of the 17 participants who responded to the evaluation survey after workshop one agreed that they understood the objectives. Participants' expressions of the objectives in their own words suggest that they were clear that the dialogue was focused on CF, and on widening the genomic testing already used for CF, rather than whole genome sequencing and this was clear in their own wording of the objectives. The majority seemed to have fully grasped the conundrum of sensitivity versus specificity and the trade-off between picking up more 'true CF' cases versus more CFSPID designations.

Should wider genomic testing be used for CF?

During workshop one participants explored their aspirations and concerns about wider genetic testing for CF and, by the end of the session, participants seemed generally in favour of more extensive testing for CF, appreciating that it would be valuable to cover more mutations causing CF and allowing greater equality of access to accurate diagnosis of 'true

CF' to families from wider ethnic backgrounds. Wider use of genomic testing was seen as a positive direction of travel for society, but participants were clear that this needed to be in the context of meaningful informed consent processes for parents. Some participants raised wider concerns about whether greater use of genomic testing could gradually lead to elimination of genetic conditions such as CF as families receiving a diagnosis choose not to have more children and societal pressures build for pre-natal screening. They questioned whether this would be a good thing for society.

Which testing approach should be applied?

Before and during workshop two, participants had a chance to explore the implications of the two different test approaches through a mix of: reviewing materials on their own; through discussions with family and friends; discussion in two rounds of facilitated small group deliberations; and by questioning specialists. The dialogue report describes participants' journeys and how their opinions evolved over the process. A graphic in the report annex illustrates the results of online polling (Mentimeter) which gives a useful snapshot (albeit based on very small numbers) of where individuals arrived at in their deliberations. This helped frame the in-depth qualitative findings which explored the underlying reasoning and values that informed participants' choices. The Homework task allowed the opinions of at least another 20 individuals (friends and families) to be hear in the deliberations. Steering Group members and other stakeholders reported during the dissemination event that the findings were of great interest and would be a useful input to their decision-making process.

Part of the value of the findings was in giving decision makers a greater understanding of the value that participants place on certainty and how they took this into account in weighing up which approach would minimise overall harms to society. While some participants felt that more knowledge is always better (and therefore favoured a more sensitive test), the majority felt that not knowing would be preferable to a designation of CF SPID that might lead to great uncertainty and stress for families with negative impacts for their children labelled with CF SPID. This was clearly unfamiliar territory for participants more used to an NHS screening process that leads to a certain diagnosis and a clear treatment pathway. As they grappled with this uncertainty, by the end of the dialogue the majority of participants favoured the more specific test, which would generate less CF SPID designations, but at the risk of missing a few 'true CF' cases.

The dialogue also highlighted the importance of a few key pieces of information that participants drew on in weighing up benefits and harms. In this case two pieces of information proved pivotal for those preferring the more specific test: the reassurance provided by a specialist that the handful of children who missed out on a 'true CF' diagnosis at birth would almost all be diagnosed before they were two and that new therapies becoming available would mean that their long term outcomes would probably not be affected; and the comparative numbers of individuals and their families affected (80 avoiding a CF SPID designation versus less half a dozen who might miss a 'true CF' diagnosis at birth). Some participants reflected that further information affecting these underlying assumptions might have caused them to revisit their choices.

The dialogue also provided interesting insights from the individuals preferring the more sensitive test, about how people might view screening in the NHS. They saw the primary role of any screening programme as diagnosing as many cases as possible and suggested that any test which failed to do so might risk a loss of trust in NHS screening. Some participants felt that despite the uncertainties, a CF SPID designation could actually be helpful for families and the health service in getting patients who did develop symptoms into a treatment pathway more quickly and creating a driver for more research and support to be focused on CFSPID in the future.

Stakeholder views on whether the objectives were met

- *A really valuable piece of work and a key foundation to consider alongside the other two workstreams when deciding policy.* I Steering group member
- *"Taken a really complex subject (a bit less binary than presented) and making it binary has enabled participants to go on a journey and confidently arrive at a preferred option."* I Clinician
- *"The presentation of findings was very clear – the findings blew me away, not quite what I was anticipating."* I Clinician
- *"Was exciting work to engage the public on such as tricky issue."* I Steering Group Member

3.2 Potential impacts on UKNSC decision-making on CF screening

HVM presented the report findings to the CF NGS Active Advisory Board CF stakeholder group on 21st September. The online meeting was attended by 16 members including CF specialists, academics, and representatives of patient support groups, including European representatives (CF Centre Genoa, Italy, and the European Society for Human Genetics). A Q+A session highlighted the group's appreciation of the credibility and robustness of the work. Those who shared their views were impressed by the way the HVM team had approached the complex subject, taken public participants on a journey at the end of which they had been able to arrive at a nuanced expression of which test they preferred, and why.

Several stakeholders pointed out the value of this longer form deliberative approach compared to other social research and opinion polls. Some attendees were surprised that many in the group had arrived at a preference for limiting the damage associated with uncertainty rather than pursuing diagnoses of 'true CF' at all costs. Several appreciated that participants had been able to come to a wider societal view of minimising harm for the greatest numbers, pointing out that this would not have been possible through opinion polls or other types of social research. They also took lessons about the potential importance of a few key pieces of information in shaping views.

As noted in section 2, the Cystic Fibrosis NGS Active Advisory Board is now in the process of commissioning research with two other groups of stakeholders and this is expected to start in October 2021 and finish in September 2022 (allowing six months to secure ethics committee approvals for research with patients and their families). The results from this mini public dialogue will then be considered in parallel with the findings from the other two

groups to inform a single recommendation on introducing wider genomic sequencing to CF newborn screening. The timing of the implementation of any change to the newborn screening programme for CF is likely to be between 2024 and 2026.

The findings from the public dialogue are expected to provide important lessons for the contractors carrying out research with the patients and families. These are likely to include the time needed for participants (even with a starting knowledge of CF and testing) to fully think through the options, the benefits of having access to specialists, and the importance of how information they receive is framed. The stimulus materials and presentation prepared by specialists are also likely to be useful inputs to the research with patients and families.

Feedback from stakeholders on the dialogue findings

- *"A great piece of work offering useful insights, not just on the public view of the use of wider genomic testing as part of screening for CF but adding to the discussion around first line genomic testing as part of newborn screening."* | Commissioner
- *"Demonstrates that we really need to take time to frame the topic – we can't do cold call or snap opinion surveys – it has to be this slower form of engagement."* | Academic
- *"Really fascinating, highlights the importance of how we frame the question and how one or two pieces of information can really sway opinions and how careful we need to be in framing work with other stakeholders."* | Academic

3.3 Wider impacts on policy making

The Steering Group also expects to feedback the findings from this mini-public dialogue to wider policy decisions about the roll-out of whole genome sequencing for newborn screening. The insights gained will be shared with Genomics England and should prove helpful to the team tasked with designing and planning the pilot for WGS for NBS.

One key learning that the commissioners anticipate sharing is the length of time required for even a pre-informed audience with some prior knowledge of whole genome sequencing and its opportunities, to come to a decision about how to apply wider genetic testing to a single condition within tightly defined parameters. This has implications for how the Genomics England team think about the informed consent process; this will require parents to think about many more conditions including some conditions with inherent uncertainty which may not develop until later life, if at all. The insights from this mini-dialogue suggest that parents will need to start with a basic understanding of screening, WGS and genetic conditions – and that they will need time to be able to talk to others, including specialists but also friends and family, in coming to their decisions. The findings appear to lend weight to a view (reported to be emerging within the UK NSC) that WGS for NBS cannot be rolled out to an uninformed public: rather it should be tested with a carefully prepared cohort of families who are supported through the process.

The dialogue also highlighted the importance of families having access to consistent accurate information from those that are supporting them in their choices. This will likely have implications for healthcare workers and genetic counsellors who will need training for

the roll out of WGS for NBS. This may reinforce UK NSC's thinking that WGS for NBS would be better piloted in a research environment rather than in a service delivery setting.

These findings are also expected to be of interest to international audiences considering extending CF and wider genetic sequencing. Professor Bonham plans to submit an academic paper for the International Journal of Neonatal Screening reflecting the findings from this public dialogue.

3.4 Participants satisfaction with taking part

Almost all the participants fed back to the evaluator how pleased they had been to take part, with many saying they had felt privileged and fortunate to have been invited and describing themselves as feeling positive, more informed and educated as a result of the experience. As shown in *Annex A*, many participants remarked that it had been very interesting and a great experience. *"Again, I have absolutely loved being part of this public dialogue. I have thoroughly enjoyed learning new information and having the opportunity to have my views and opinions listened to and considered in making these important decisions for future public health/testing. Thank you so much for letting me participate."*

Almost all found it really interesting to be able to dive more deeply into exploring a single condition, bringing their prior understanding of broad WGS and NBS issues. Through focusing on a single condition participants recognised the complexity of the decisions facing UK NSC: "*Really lucky to input my voice, but also understand how some of these decisions are made. It is not as simple as I thought it was."* And how such decisions might impact the future of medicine: "*I found it very interesting, even if it made me more concerned about fear-based, medical culture evolving."*

When asked whether they felt confident that the UK NSC would take their views into account to help ensure that potential new tests are as beneficial as possible to newborns, their parents and society, almost all 15 respondents agreed – only one was unsure. This confidence appeared to reflect two factors: they felt valued, knowing they had been selected from a larger number of willing respondents; the amount of time invested and openness of the team of specialists who participated. They particularly appreciated the specialist's openness about the uncertainties about what CF SPID might mean for patients and families. As one participant put it: "*I think the genuine spirit and information given really expressed genuine concern that the "right" way forward was found*" while another remarked that *"if anything, it has raised my confidence in the internal dialogues within the official bodies.*"

Although this was already a second round of dialogue for the participants, several expressed an interest in being kept updated on the outcomes and taking part in any future research. "*I will be interested to see the various reports and hope to see any journal articles too*." And "*It has been a brilliant experience. I wish you all the best. If I can be part of any future groups, please let me know."*

This cohort could be a useful resource for UK NSC for future screening research.

4. Meeting best practice principles for online dialogue delivery

This section covers the design and delivery of the dialogue in relation to Sciencewise best practice principles for online dialogues. The dialogue was able to bring together a relatively small number of participants (19) who were able to reflect key demographics (slightly skewed to younger age groups) and attitudes towards genomic testing, and provide them with sufficient, accessible information and access to specialists to allow them to have in depth discussions in small groups about whether they saw a role for wider genetic testing for CF. The carefully designed four and a quarter hours of online deliberation, split across two workshops two weeks apart allowed them time and space to make their views heard. All participants enjoyed the experience and appreciated being able to put their knowledge gained from the WGS for NBS online dialogue to further use.

4.1 A diverse and inclusive mix of participants

The number of participants was slightly below the target of 21 due to two late dropouts at a point when it was too late to replace them. One further drop-out before workshop 2 meant that 18 participants attended both sessions and completed all the homework tasks. Despite the small numbers the group gave a really good mix of gender and socio-economic backgrounds and attitudes towards WGS, but with slightly few individuals from Black and minority ethnic backgrounds than had been hoped for. All of the participants had prior experience of using online tools (Zoom and Mentimeter) and the HVM team provided technical back-up before and during meetings and support for participants between sessions if they needed it. There were no signs of digital exclusion.

4.2 Fulfilling a duty of care to participants

The HVM team had put considerable thought into fulfilling their duty of care to participants. The small team were already aware of the selected individuals' lived experiences of genetic conditions and were able to create a warm and safe environment. A ratio of one facilitator to 6-7 participants allowed everyone to contribute actively and we observed that they were fully engaged and felt confident to share their opinions from the outset. Several participants praised the style of individual facilitators and how comfortable they felt sharing their views. As one SG observer noted, "*HVM dealt with tricky subjects in a very sensitive way*."

Participants were invited to contact the lead facilitator directly if they felt the need to talk anything through privately. If they felt upset by anything they heard during the sessions they were encouraged to step out of the small group and re-join as and when they felt ready. In the event no one needed to do so. Links to support organisations1 were also provided in the participant pack.

All 17 respondents after workshop one strongly agreed that the facilitator team respected their input and after workshop two all agreed that facilitators were able to make sure that

[1] Breaking down Barriers, Genetic Alliance, and NHS Mental Health services

everyone's views were heard and respected even if they were views that were not widely shared: "*including a religious [view] - not mine - but I was pleased to see it was respected*". Almost all participants agreed after workshop two that they had felt confident to share their views in small group sessions and in plenary.

4.2 A mix of deliberation opportunities

Participants had the chance to deliberate in several ways: synchronously in their small groups; and asynchronously through their homework discussions with friends and family. The online sessions were focused on structured discussions within small groups of mixed ages and backgrounds.

The facilitation team were already well versed in WGS for NBS issues and provided continuity with the previous dialogue and between the two rounds of discussions in small groups. The evaluator observed that the participants' familiarity with public dialogue approaches and what was expected of them and the safe and supportive environment created by the facilitation team enabled participants to quickly get fully engaged in discussions. Both of the small groups observed by the evaluator gelled well and quickly got to the key issues. They listened closely to each other and built on different viewpoints to inform their thinking.

A number of participants provided evaluation feedback that discussions with people from different backgrounds had been the most valuable part of the dialogue for them in helping work through their own views. Several participants reported that they really enjoyed meeting people who had taken part in the WGS for NBS dialogue from different locations or specialist groups. One participant suggested he would have gone further and mixed the groups again for workshop two: "*Mixing of the small groups to hear a varied range of viewpoints*."

Participants also welcomed the opportunity to engage with friends and family through the required homework task. Almost all participants appeared to have explored the test options with at least one other person and several had canvassed more widely, seeking out opinions that might be different from their own. The majority of participants felt that the 'homework' task had helped them think through the implications raised by the new potential genomic test for CF. Most gave examples of how the exercise had helped them either in clarifying their understanding, challenging their views, or raising new questions that they needed answers to in workshop two. Given the relatively small numbers involved in this mini dialogue, the homework task proved an effective way of amplifying the number of voices heard in the process.

4.3 Sufficient, balanced information to enable informed discussion

The information provided to participants was balanced, accessible and varied and included a participant pack shared before the first event, PowerPoints, and a homework task. The information pack was shared well in advance of the first workshop and included a link to pre-workshop materials accessed via the HVM website. This included short videos familiar to participants (and previously reviewed for accuracy and balance) from the previous WGS for NBS dialogue. Many appreciated the chance to recap before the first workshop: and all

participants agreed by the end of the process that having had some prior understanding of WGS and NBS had helped them to understand the issues for this dialogue.

By the end of workshop one all respondents (17 of 19 participants) agreed that presentations on how the potential new genomics tests would work and could affect diagnosis, treatment, care, and research were clear and easy to understand: they also agreed that they did not feel there had been anything that had been difficult to understand.

4.4 Access to a mix of specialists to answer participants' discussions

The group of three specialists worked well together to provide participants with helpful insights into the complexities around how the extended genomic tests might be used and the issues that might arise for patients, families, and wider society. The participants appreciated the mix, expertise, and balance of views they heard: *"It's nice to have a mix of experts coming from different angles."* Participants unanimously agreed that specialists had explained the issues and answered their questions in an accessible way. "*The specialists spoke well and helped to explain the difficulties that come with making this decision."* And *"They were amazing, they clarified questions and gave very informative answers.*

Participants also appreciated the effort made to address unanswered questions between workshops: almost all (12 out of 14 respondents) found the written answers shared before workshop two helpful in addressing their questions and all agreed that the Q+A session with specialists in workshop two helped to answer any outstanding questions. Indeed, participants reported that this session had really clarified some grey areas including the implications of a late diagnosis of true CF, the relative trade-off in numbers for CF or CF SPID results that would be missed/picked up by the different tests and the likely support available to families receiving a CF SPID designation.

Several participants reported being favourably surprised by how open specialists were in talking about the areas where there is not yet sufficient research to give clear answers. One noted *"I appreciated the honesty and confusion around how to deal with CF SPID."* and another that *"It was good to have their honesty too, instead of just being told what they think we want to hear."* As noted in *Section 3*, these answers were a decisive input for some participants in feeling confident to make a choice about which test they preferred. This openness also appears to have been a strong contributing factor in participants feeling a trust in the decision-making process they were contributing to.

4.5 Sufficient time and space for discussion

All sessions ran to time and started and finished promptly. Overall, the two sessions allowed ample time to cover the key topics. Timings were slightly tighter for workshop one where the presentations slightly over-ran but the programme had sufficient in-built flexibility to ensure that all topics were covered and specialists were able to answer at least two questions from each of the three small groups in plenary. Participants' familiarity with working on zoom and the specialist/small group format helped to make the most of the time available and all participants (17 respondents) felt after workshop one that the pace of the session felt about

right and appropriate to the complexity of the topic. One participant commented that *"It seems like the appropriate amount of time to talk about such a complex issue*." and another that it felt like the "*Right pace, right focus*." Workshop two had ample time for small groups to cover all the topics in the required depth.

4.7 Facilitation was professional and independent

The design, discussion guide and expertise of the facilitators enabled them to both create groups where everyone felt confident to share their views and build on each other's points and allowed them to probe participants' responses to understand what underlay their views and to encourage them to think about wider societal viewpoints. After workshop two all participants agreed that facilitators were able to make sure that everyone's views were heard and respected.

4.8 Participants' thoughts were fully captured

All sessions were audio recorded on zoom. Small group facilitators also took simultaneous notes in PowerPoint to mimic a flipchart which they could share with participants. This allowed participants to see that all their substantive points had been captured. We observed that they found it helpful in formulating their own thoughts and correcting the meaning of what was being recorded. It also helped the group to prioritise questions to ask to specialists and the pros and cons of different testing approaches.

Participants seemed very confident to disagree with each other and explain their own thinking in small groups. One participant voiced a slight concern that the framing of the probe questions was slightly repetitive and in looking for something new to add may have introduced views that were not fully representative: "*My only concern is that sometimes I feel pressured not to repeat myself and come up with unique perspectives others may not have thought about - maybe this does not represent our views all the time."* **I** public participant. In the view of the evaluator the dialogue report has been able to make it very clear where views were widely shared and where they were not.

EVoting, using the Mentimeter app, was used to good effect to capture participants' top of mind reactions to presentations and their preferences between sensitive vs specific tests at the beginning and end of workshop two. This worked well to give participants an idea of the sentiment of all three small groups and how this changed from the start of workshop two (favouring the more sensitive test) to the end of the dialogue (favouring the more specific test.) The results are very much mirrored in those of an evaluation question[2] completed on their own after the workshops which appears to confirm that people were giving their own considered opinions rather than being influenced by what they thought the majority view was.

[2] SurveyMonkey after workshop two showed that by the end of the dialogue most participants were significantly less concerned about missing some cases of 'true CF' - an average score of 73 where 1 = very concerned and 100= not at all concerned - compared to significantly greater concern (an average score of 29) about identifying an increased number of designations of CF SPID.

5. Conclusions and Lessons learnt

This mini dialogue has been designed and delivered in a short time frame (five months from start to final reporting) and very cost effectively (less than £25,000 for the dialogue and evaluation). The process was overseen by a small steering group with previous experience, and realistic expectations, gained through their involvement in the previous online public dialogue on WGS for NBS. The process has demonstrated that, as a follow on to an existing project, it is possible to deliver a small online dialogue, with closely defined objectives and produce useful findings of wider applicability. This was made possible because the commissioners were familiar with public dialogue, the participants and delivery contractors were warmed up to the subject, and some materials were already in place.

Alongside inputs from future engagement with other stakeholders, these findings provide evidence to UK NSC to help inform its decision-making about the future extension of CF testing. The stimulus materials and lessons learnt from the mini dialogue will also help to inform the engagement with families and practitioners. The insights gained from a deeper dive into choices about testing for a single genetic condition will also be fed by UK NSC into work being carried out by Genomics England to develop the Whole Genome Sequencing for newborn screening pilot for a wider set of conditions.

The evaluation has highlighted a number of lessons for future online delivery of public dialogues by UK NSC and Sciencewise:

- Carrying out the work as an extension to a previous project allowed it to be effectively steered and delivered with very light touch commissioning, governance, and project management arrangements.
- Tight framing of the dialogue questions allowed the commissioners to get useful insights from relatively short sessions over a two-week period (four and a quarter hours of synchronous group deliberation and an hour of asynchronous 'homework'). The homework task helped to amplify the number of voices heard in the dialogue.
- A very experienced public dialogue contractor was able to apply best practice lessons on how to structure and deliver an effective online process. The team's prior knowledge of WGS and genetic conditions, including Cystic Fibrosis, allowed them to pull together accurate and balanced materials and brief a small pool of specialists to provide participants with the information they needed in an accessible way.
- In an emerging field such as genomics/genetic interventions where there is limited evidence about long term impacts it is really important to be clear when sharing information what is known and backed by robust evidence and what is still uncertain, or where medical opinions vary.
- The success of the previous process contributed to the team being able to recruit from a large pool of informed and willing individuals. This contributed to a very streamlined recruitment process and choice of a sample that, despite being small (19 individuals)

gave a good mix across all the required demographic, socio economic and attitude characteristics.

- Holding the sessions online made it possible to recruit participants from across the UK, which would not have been financially or logistically feasible face-to-face.
- A prior understanding of the likely sensitivities and participants' lived experiences helped the facilitation team meet their duty of care to participants. Excellent facilitation meant that participants felt respected, heard and that their opinions would be considered by decision makers. The time invested and the openness of specialists contribute to a strong trust in the process.
- The experience that participants brought – including an understanding of WGS for NBS opportunities and concerns, how public dialogue works and how to use online tools such as Zoom and Mentimeter – allowed the dialogue to cover a lot of ground in a short period.

Annex A: Participants responses to evaluation questionnaires

Participant feedback from the first session 22/6/2021. Based on 17 participants (of 19)

1. I understand the objectives of this dialogue and how it differs from the previous workshops on Whole Genome Sequencing for Newborn Screening

Strongly agree	Tend to agree	Neither	Tend to disagree	Strongly disagree
11	6			

Participants expressed the objectives as:

- *To discuss the issues surrounding changing the current genomic test within the newborn heel prick test to a wider screening programme.*
- *Opinions on new screening options for CF*
- *Screening specifically for CF via genome to refine testing though it is partly carried out this way anyway via the spot test*
- *This is to decide what is best to do with regards to the potential for using wider genome testing specifically for Cystic Fibrosis. It is about making a decision on the testing method - whether to test broadly and generate more CFSPID diagnoses as a by-product, or to test more specifically which could reduce CFSPID cases but potentially miss true CF cases.*
- *To gather opinions on the advantages and disadvantages of changing how we carry out newborn testing to include genomic testing for CF.*
- *To evaluate the pros and cons between genomic sequencing for CF that is either more specific or more sensitive.*
- *Consider the impacts of sensitivity and specificity on CF screening and how it will lead to more CFSPID and number of CF suffers recognised OR miss some CF sufferers but reduce the stress of those left in the grey area of CFSPID.*
- *To concentrate on the CF screening with pros and cons*
- *To ensure that the most number of children are diagnosed at birth*
- *To find out what type of test to do going forward and the benefits and drawbacks of including/mixing CFSPID "diagnosis"*
- *To decide if specific CF testing should be performed in newborn screening?*
- *Balancing the decision between correctly diagnosing True CF against the number of CFSPID that may be incorrectly diagnosed, and the stresses involved.*
- *Support in weighting the best decision on whether to identify more CFSPID vs true CF diagnosis*

2. The presentation on how the potential new genomics tests would work and could affect diagnosis, treatment, care & research was clear and easy to understand.

Strongly agree	Tend to agree	Neither	Tend to disagree	Strongly disagree
10	7			

3. Was there anything that was unclear or where more information next time would be helpful

- *More info on why not still doing sweat test and if CF is actually treatable would have been useful*
- *No... other than it is weighing the pros and cons to either decision.*

4. The specialists explained the issues and answered our questions in an accessible way.

Strongly agree	Tend to agree	Neither	Tend to disagree	Strongly disagree
13	4			

- *All speakers provided the information in a clear and easy way for non- medical personnel to understand.*
- *Really good explanations to questions*
- *They were amazing, they clarified questions and gave very informative answers.*
- *The specialists spoke well and helped to explain the difficulties that come with making this decision. Appreciated the honesty and confusion around how to deal with CFSPID.*
- *It's nice to have a mix of experts coming from different angles.*

5. The facilitator team captured my questions and respected my input.

Strongly agree	Tend to agree	Neither	Tend to disagree	Strongly disagree
17	0			

- *Very engaging*
- *I feel comfortable and able to share my views*
- *Excellent facilitator in [name]*
- *[name] was a delight and ran the group well.*

6. The pace of the session felt about right.

Strongly agree	Tend to agree	Neither	Tend to disagree	Strongly disagree
12	5			

- *It seems like the appropriate amount of time to talk about such a complex issue.*

7. Anything that you would like to talk more about next time?

Participants suggested more discussion on areas that were already coming up in the Q+A sessions and these were shared with the delivery team.

Clarifying issues around the current test

- *Funding medication and how it is different in parts of the country*
- *The false negative rate as it currently stands from the heel prick test vs positive false negative rates from both wider genomic testing options*

What are the implications of not picking up true CF cases?

- *We talked a lot about the possibility that true CF cases may be missed if we do not go with a very specific testing option but we did not discuss that those cases may be picked up by parents noticing the symptoms therefore there is a potential that there could be no missed cases.*
- *Is a later diagnosis more problematic?*
- *Maybe the worst-case scenario for a child that goes undiagnosed with CF as a baby.*

Support for families and patients

- *Support for diagnosis was mentioned as being both poor and good. Which is it?*
- *How the stress of diagnosis may lead to more illness?*

How will public dialogue results fit into the decision-making process

- *How they will use our research and findings to shape what is done going forward.*

Timing if wider genetic testing is rolled out

- *I would like to know how soon these testing methods would be put in place (i.e., when will they be used on newborns going forwards)? Is this something that will be rolled out in the immediate future or in 3-5 years for example?*
- *When and how soon could it be put in place?*

8. Anything we can do to improve the experience next time?	All but one said they did not see the need for any changes. One suggestion: • *Is it still appropriate to share ongoing questions live within the chat (rather than raise hands or butt-in) or is that distracting? There was more use of this feature in the previous consultation.*

Workshop 2: 6th July, 16 participants of whom 14 filled the evaluation survey

	Strongly agree	Tend to agree	Neither	Tend to disagree	Strongly disagree
The homework task (talking to family and friends) helped me think through the implications raised by the new potential genomic test for CF.	9	5	1		

Please give examples of how:

Clarifying or highlighting key issues:

- *Clarifying the issues by explaining it to others.*
- *It gave me a better understanding of CFSPID from the family's point of view.*
- *The mental health implications of a CFSPID diagnosis to new parents.*
- *Highlighted the fact we are entering into a fear-based culture where medical doubts are more and more being put into people's minds, inducing fear and stress which in turn cause more illnesses.*
- *[highlighted] The difference in opinions depending on stage of life.*
- *Highlighted the need for investment in better technology for screening.*

Challenging my own views

- *It helped to give me the chance to consider and explore different perspectives from my own. I thought it was a useful way of exploring the topic and gathering views.*
- *My family and friends were leaning more to a sensitive test while I mainly supported the specific so it was really interesting to have my view challenged.*
- *Just led to a very lively debate on a rooftop and helped me understand other aspects and opinions that hadn't crossed my mind.*

Raised more questions

- *Are there any implications if treatment was given to an incorrect diagnosis? The security of the information and does it open the door to further Genome testing without further consultation. What are the implications for a child if True CF diagnosis is missed?*

	Strongly agree	Tend to agree	Neither	Tend to disagree	Strongly disagree
The written answers shared after workshop 1 helped address some of my unanswered questions.	7	5	3		

	Strongly agree	Tend to agree	Neither	Tend to disagree	Strongly disagree
The Q+A session with specialists helped answer some of my priority questions.	14	1			

- *Clarified lots of grey areas in a very complex topic.*
- *Really helped focus our next discussion as I felt more confident speaking.*
- *Numbers were key.*
- *Clarification on late or non-diagnosis of True CF.*
- *Steered my view based on the comment that no support is likely to be offered to CFSPID families.*
- *It was good to have their honesty too instead of just being told what they think we want to hear.*

	Strongly agree	Tend to agree	Neither	Tend to disagree	Strongly disagree
I felt confident to share my views in small group sessions and in plenary.	10	4	1		

- *I really like the small group sessions. Some questions were very similar though and I was worried about repeating myself frequently.*
- *I was a little more frazzled this time than last, but I think that's just because it's such a complex issue.*

	Strongly agree	Tend to agree	Neither	Tend to disagree	Strongly disagree
Facilitators were able to make sure that everyone's views were heard and respected.	12	3			

- *I felt very comfortable talking to them.*
- *Yes, including a religious [view] - not mine - but I was pleased to see it was respected*
- *Asking everyone in turn was good so you didn't feel as if you were dominating the discussion.*
- *[name] is a brilliant facilitator. She always makes sure everyone is included in the discussions and encourages people to further develop their answers to better explain what they are meaning.*

This dialogue has allowed me to contribute an informed opinion about how wider genomic testing should be used for Newborn screening for CF in the future.

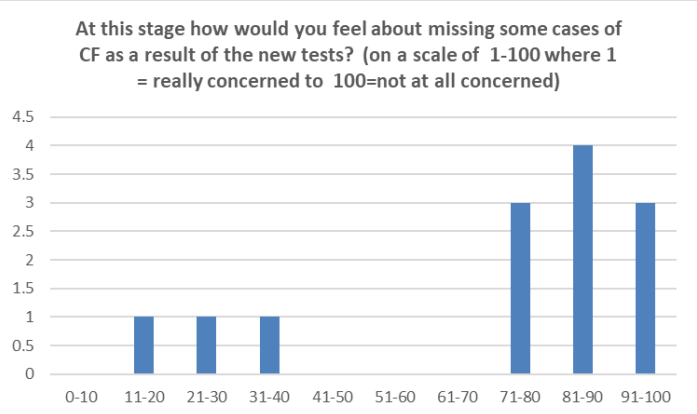
Strongly agree	Tend to agree	Neither	Tend to disagree	Strongly disagree
12	3			

Having some prior understanding of whole genome sequencing and Newborn screening helped me understand the issues.

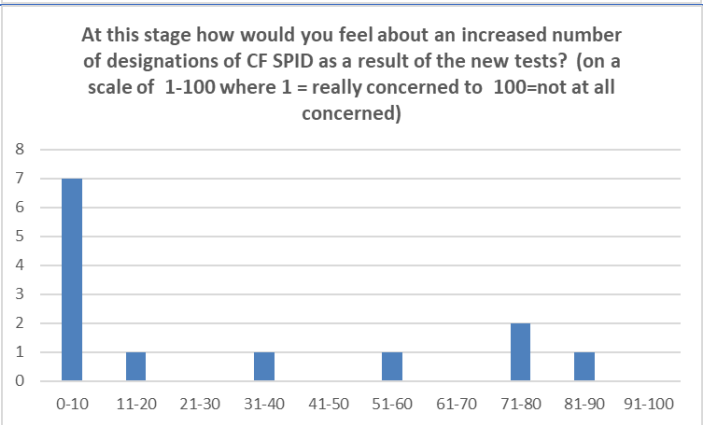
Strongly agree	Tend to agree	Neither	Tend to disagree	Strongly disagree
14	1			

- *The terms and wider consequences were already understood so the issue at hand could be focused on.*

At this stage how would you feel about missing some cases of CF as a result of the new tests? (On a scale of 1-100 where 1 = really concerned to 100=not at all concerned)
Average 73



At this stage how would you feel about an increased number of designations of CF SPID as a result of the new tests? (On a scale of 1-100 where 1 = really concerned to 100=not at all concerned)
Average 29



I am confident that UK National Screening Committee will take our views into account to help ensure that potential new tests are as beneficial as possible to newborns, their parents and society.

Strongly agree	Tend to agree	Neither	Tend to disagree	Strongly disagree
9	5	1		

- *I will be interested to see the various reports and hope to see any journal articles too.*
- *I think the genuine spirit and information given really expressed genuine concern the "right" way forward was found.*

What, if anything, was most valuable about the public dialogue workshops?

- *Right pace, right focus.*

Understanding screening plans and talking to experts:

- *Gaining an awareness of new plans for Screening in the UK.*
- *Having the opportunity to listen to the views and knowledge of the experts working in this field was really helpful to better understand the implications of the new testing methods.*
- *Speaking to our experts on the life of people with CF and CFSPID. It is so important not to forget the people who are affected by this rather than numbers.*
- *Giving me an understanding of the issues.*
- *Panellists very good, well organised as well*

Having a dialogue with other people to help work out their own views:

- *Hearing other people's views and having time to review my own.*
- *Getting into small groups and sharing opinions and thought with others from different backgrounds in order to gain different perspectives.*
- *Open dialogue.*
- *Having the different discussions was really helpful because we got a mix with professionals and with people that are 'normal' with everyday opinions.*
- *Hearing everyone's different views.*
- *Hearing everyone's views.*
- *That we were able to share our views and opinions with each other in order to come to a conclusion.*

What, if anything, might have been done differently?

Only 10 answered this question of whom 6 said that this was not applicable, nothing could have been improved, for example:

- *Nothing very, N/A - the workshops have been great, the team is great!*
- *Nothing it was great professional throughout.*

Two made suggestions:

- *Maybe mixing of the small groups to hear a varied range of viewpoints*
- *More statistics showing how small fears of missed CF cases actually are.*

Overall, how do you feel about having taken part in this public dialogue?

- *Positive. Informed. Educated.*
- *Again, I have absolutely loved being part of this public dialogue. I have thoroughly enjoyed learning new information and having the opportunity to have my views and opinions listened to and considered in making these important decisions for future public health/testing. Thank you so much for letting me participate.*
- *I am happy to have had the chance to take part.*
- *Very positive and if anything has raised my confidence in the internal dialogues with the official bodies.*
- *THANK YOU SO MUCH for this opportunity. It has been amazing and my thanks go to [name] our facilitator. The talks have been really informative and I felt my views were valued. My only concern is that sometimes I feel pressured not to repeat myself and come up with unique perspectives others may not have thought about - maybe this does not represent our views all the time Still this has been a brilliant experience. I wish you all the best. If I can be part of any future groups, please let me know.*
- *More informed and privileged to be able to take part in it.*
- *Found very interesting, even if it made me more concerned about fear-based, medical culture evolving.*
- *Really lucky to input my voice but also understand how some of these decisions are made. It is not as simple as I thought it was.*
- *Privileged.*
- *Very fortunate to have my opinion and views considered.*
- *I have enjoyed the experience and feel privileged to have participated in this dialogue.*
- *It was very well organised.*

Annex B Steering Group and specialists

Jim Bonham	PHE, Laboratory Lead for Newborn Screening
Lauren Cooper	PHE Newborn screening bloodspot programme
Christine Cavanagh	PHE Newborn screening bloodspot programme
Catherine Joynson	UKNSC and Nuffield Foundation
David Elliman	NHS Foundation Trust Great Ormond Street Hospital for Children
Iolo Doull	Consultant Respiratory Paediatrician Children's Hospital for Wales, Cardiff
Felicity Boardman	Professor in medicine ethics and society, Warwick Medical School
Suzannah Lansdell	Sciencewise
Philippa Lang	UKRI