PEER REVIEW HISTORY

BMJ Open publishes all reviews undertaken for accepted manuscripts. Reviewers are asked to complete a checklist review form and are provided with free text boxes to elaborate on their assessment. These free text comments are reproduced below.

ARTICLE DETAILS

TITLE (PROVISIONAL)	Perceptions of genomic newborn screening: A cross-sectional survey conducted with UK medical students
AUTHORS	Seed, Lydia; Scott, Anna; Pichini, Amanda; Peter, Michelle; Tadros, Shereen; Sortica da Costa, Cristine; Hill, Melissa

VERSION 1 - REVIEW

REVIEWER NAME	Chudleigh, Jane
REVIEWER AFFILIATION	City University, School of Health Sciences
REVIEWER CONFLICT OF INTEREST	None
DATE REVIEW RETURNED	05-Jul-2024

GENERAL COMMENTS	Thank you for submitting this interesting and timely paper. Please see minor suggestions on the attached. (The reviewer provided a marked copy with additional comments. Please contact the publisher for full details.)

REVIEWER NAME	Vears, Danya F
REVIEWER AFFILIATION	University of Melbourne
REVIEWER CONFLICT OF INTEREST	I have no competing interests to declare
DATE REVIEW RETURNED	10-Jul-2024

GENERAL COMMENTS	This manuscript reports on the findings of a questionnaire about genomic newborn screening distributed to medical students in the United Kingdom. The paper is well written and the findings are novel and of importance to the field. I have the following minor comments. Abstract Can you say something about what nature of the questions that were asked? Introduction The authors might also want to comment on public perspectives other than those in the UK, for example the work by Lynch et al. Results Page 13, line 3: What, if anything, were the participants told about the likelihood of uncertain or incidental findings?

VERSION 1 – AUTHOR RESPONSE

Reviewer 1 comments:

Thank you for submitting this interesting and timely paper. Please see minor suggestions on the attached. (Ed: attached the marked up PDF).

Comment: [Abstract, Methods] Was this online? What platform was used?

Response: We have added that the survey was disseminated online and used the RedCap platform.

Comment: [Abstract, Results] It would be helpful to have an explanation of what this means i.e., it sounds like it came from data collected using the Likert scale?

Response: As detailed in the Methods section of the main paper, participants expressed their overall support of genomic newborn screening using a visual analogue scale. We have added to the methods section of the abstract to say that the survey used a mix of multiple-choice questions, Likert scales, visual analogue scales and free-text questions. We felt that it would be too much detail to include in the type of scale for each quoted result in the Results section of the Abstract and full details are in the Methods of the main paper. As per a further comment from this reviewer, we have added the standard deviation and range for the mean support score in brackets to provide further information in the abstract.

Comment: [Abstract, Results] Other perceived benefits....

Response: As suggested, we have added for this for the sentence to read 'Other perceived benefits included earlier diagnoses...'

Comment: [Abstract, Results] perceived

Response: As suggested, we have changed this sentence to read 'However, several perceived challenges were highlighted...'

Comment: [Introduction] This reference might be helpful here: https://authors.elsevier.com/sd/article/S2667-3215(24)00064-7

Response: Thank you for highlighting an interesting paper on a topic within the broader field of genomic newborn screening. While this is an important paper that explores a similar topic, it is not felt to be directly relevant to our piece of work. This reference explores parents' and children's views on using genomic testing as a follow-up test to confirm a diagnosis of cystic fibrosis after an indicative bloodspot screening result. This differs to the focus of our paper in two main ways: we are exploring views around using genomic tests as the screening test itself (not as a follow-up test), and for a large number of conditions (not just focussed on one condition). We have therefore decided not to cite this paper in our manuscript.

Comment: [Methods] As per the abstract, was this online and using which platform?

Response: We have now clarified that this was an online survey in this opening sentence of the Methods section. We have now included the details of the online platform used in the abstract of the paper. We have left further details of the online platform in the relevant ‘Survey dissemination’ subsection of the Methods.

Comment: [Methods] I think this should be earlier as indicated

Response: We have now clarified that this was an online survey in this opening sentence of the Methods section and kept the specific details of the online platform in this subsection.

Comment: [Results: Overall gNBS support and relevance to future practice, referring to SD and range of mean support score] Maybe this should be included in the abstract?

Response: This has been added to the abstract, as suggested.

Comment: [References: Reference 13] This reference needs updating

Response: Reference has been updated to include full surnames of all authors and place of publication.

Reviewer 2 comments:

This manuscript reports on the findings of a questionnaire about genomic newborn screening distributed to medical students in the United Kingdom. The paper is well written and the findings are novel and of importance to the field. I have the following minor comments.

Comment: [Abstract] Can you say something about what nature of the questions that were asked?
Response: A brief outline of the topics broadly covered in the questions asked has been added to the Methods section of the Abstract.

Comment: [Introduction] The authors might also want to comment on public perspectives other than those in the UK, for example the work by Lynch et al.

Response: A sentence describing the Australian public's general support of gNBS and citing Lynch et al has been added.

Comment: [Results] Page 13, line 3: What, if anything, were the participants told about the likelihood of uncertain or incidental findings?

Response: Likelihood of uncertain or incidental findings was included as a potential drawback of screening in the survey. Participants were not told anything about the likelihood of uncertain or incidental findings because this was felt to be quite variable and dependent on decisions made for specific screening programmes. The full questionnaire with all information, such as the participant information sheet, that was disseminated to participants is available in the supplementary information for readers to access.

VERSION 2 – REVIEW

REVIEWER NAME	Vears, Danya F
REVIEWER AFFILIATION	University of Melbourne
REVIEWER CONFLICT OF INTEREST	N/A
DATE REVIEW RETURNED	02-Sep-2024

GENERAL COMMENTS	I am satisfied with the responses to my minor comments

VERSION 2 – AUTHOR RESPONSE