Newborn WGS Survey

Please read the following information and complete the survey below.

Consent Form

This survey is part of the study exploring UK medical student's opinions on employing whole genome sequencing in newborn screening and is intended for clinical medical students in their final two years of study.

The survey should take 10-20 minutes to complete.

The aim of the survey is to help us understand your views on offering whole genome sequencing to all newborns within the NHS, the potential risks and benefits and your educational experience and training needs in genomics. You do not need to have any experience in genomics to take part.

To maintain anonymity of participants, the survey findings will be grouped for analysis and reporting.

Please read the participant information sheet below, which includes detailed information on how we protect your data. If you have any queries, please get in touch with Dr Cristine Sortica da Costa at Cristine.sorticadacosta@gosh.nhs.uk

If you are happy to take part in the survey, please tick each box below to give your consent to take part in the survey.

[Attachment: "Participant information sheet.pdf"]

I confirm that I have read and understood the participant information sheet for this survey.	☐ Yes
I understand that completing the survey is my choice and that I am free to stop at any time without submitting a completed survey.	☐ Yes
I understand that the survey is anonymous and that it will not be possible to withdraw my survey answers after the survey has been submitted.	☐ Yes
I understand that certain phrases or sentences that I write in the survey may be quoted in future reports or publications but that my name will not be included and any possible identifying comments will be removed.	☐ Yes
I consent to taking part in this survey.	☐ Yes

Demographics

Which university/medical school do you currently attend?	◯ University of Aberdeen ◯ Anglia Ruskin University ◯ Aston University ◯ Barts and the London School of Medicine ◯ Brighton and Sussex Medical School ◯ University of Bristol ◯ University of Cambridge ◯ Cardiff University ◯ University of Dundee ◯ University of Edinburgh ◯ University of Exeter ◯ University of Glasgow ◯ Hull York Medical School ◯ Imperial College London ◯ Keele University ◯ King's College London ◯ Lancaster University ◯ University of Leeds ◯ University of Leicester ◯ University of Liverpool ◯ University of Manchester ◯ University of Newcastle ◯ University of East Anglia ◯ University of Oxford ◯ University of Plymouth ◯ Queen's University Belfast ◯ University of Sheffield ◯ University of Southampton ◯ St George's, University of London ◯ Swansea University ◯ University College London ◯ University of Warwick
Which of the following best describes the main form of teaching at your medical school?	◯ Integrated ◯ Problem-Based Learning ◯ Traditional
In what year do you expect to graduate?	◯ 2023 ◯ 2024
How old are you?	◯ 21 ◯ 22 ◯ 23 ◯ 24 ◯ 25 ◯ Over 25
What is your gender?	◯ Male ◯ Female ◯ Non-binary ◯ Prefer not to say

Genomics Educational Experience

Approximately how much teaching have you received during your medical degree on the basic sciences of genomics?

None Some Lots

(Place a mark on the scale above)

Approximately how much teaching have you received during your medical degree on genomic medicine (the clinical application of genomics)?

None Some Lots

(Place a mark on the scale above)

Have you had any additional genomics experience beyond your core medical school curriculum?

(Tick all that apply)

- ☐ Intercalation or other undergraduate degree
- ☐ Student selected component of your medical degree
- ☐ Research project outside of a degree (e.g. summer projects)
- ☐ Graduate degree (e.g. MSc, MPhil, PhD)
- ☐ Other
- ☐ None

Genomics Knowledge

How confident do you feel in your understanding of the following:

	Not at all confident	Somewhat confident	Neutral	Confident	Very confident
The difference between DNA, genes and chromosomes	○	○	○	○	○
Identifying inheritance patterns from family pedigrees e.g. autosomal dominant, X-linked, mitochondrial	○	○	○	○	○
The difference between copy number and sequence variants	○	○	○	○	○
The difference between loss-of-function and gain-of-function variants	○	○	○	○	○
The difference between synonymous and missense variants	○	○	○	○	○
The difference between somatic and germline variants	○	○	○	○	○
The concept of mosaicism	○	○	○	○	○
The difference between clinically used genomic tests, such as microarray, single gene test, gene panel, whole exome sequencing, whole genome sequencing	○	○	○	○	○
The concept of genetic contributions to common complex diseases such as type 2 diabetes	○	○	○	○	○
How you might approach interpretation of variants eg identifying whether a variant is more likely to be pathogenic or benign	○	○	○	○	○

Newborn Screening Programme

Were you aware of the proposed pilot for expanding newborn screening to include whole genome sequencing before this survey?

◯ Yes
◯ No

Based on your current understanding, how much do you agree with the following statements about newborn whole genome sequencing (WGS):

If you feel unable to either agree or disagree due to a lack of knowledge on the subject, please select "Not enough knowledge".

	Completely disagree	Somewhat disagree	Somewhat agree	Completely agree	Not enough knowledge
The current newborn screening programme identifies all early-onset conditions that we need to know about in childhood.	◯	◯	◯	◯	◯
Newborn WGS could identify important childhood-onset conditions that the current programme does not	◯	◯	◯	◯	◯
Newborn WGS could cause an increase in unnecessary anxiety amongst parents	◯	◯	◯	◯	◯
Newborn WGS could be reassuring for parents	◯	◯	◯	◯	◯
Newborn WGS could speed up diagnosis of childhood illnesses	◯	◯	◯	◯	◯
Newborn WGS could improve long term outcomes of individual paediatric patients	◯	◯	◯	◯	◯
Newborn WGS could lead to over-medicalisation of children	◯	◯	◯	◯	◯
Newborn WGS could provide useful data for paediatric research e.g. clinical trials	◯	◯	◯	◯	◯
Newborn WGS will cost too much to the NHS; the money is better off being spent elsewhere	◯	◯	◯	◯	◯
Newborn WGS would cause more harm than good	◯	◯	◯	◯	◯

Do you agree or disagree that the following categories of disease should be included in newborn WGS screening:

	Completely disagree	Somewhat disagree	Neutral	Somewhat agree	Completely agree
Diseases where symptoms or treatment would start during infancy (0-1 years)	○	○	○	○	○
Diseases where symptoms or treatment would start during childhood (2-12 years)	○	○	○	○	○
Diseases where symptoms or treatment would start during adolescence (12-17 years)	○	○	○	○	○
Diseases where symptoms or treatment would start during adulthood (18 years+)	○	○	○	○	○
Diseases for which there are effective treatments available	○	○	○	○	○
Diseases for which there are currently no effective treatments available, only supportive care	○	○	○	○	○

Who do you think should have access to the raw data generated by newborn WGS?

(Tick all that apply)

- ☐ Clinicians directly involved in the patient's care
- ☐ Clinicians not directly involved in the patient's care
- ☐ Parents
- ☐ Research groups in the NHS or affiliated universities
- ☐ Research groups in the pharmaceutical industry
- ☐ Public health bodies
- ☐ Other

If you have selected "Other", please specify your answer. _____

Benefits of newborn WGS

What do you think are some of the main potential benefits of introducing routine WGS in newborn screening?

Please rate the importance of the following potential benefits of newborn WGS:

	(Not important) 1	2	(Neutral) 3	4	(Very important) 5
Earlier diagnosis	◯	◯	◯	◯	◯
Enabling a diagnosis to be reached in more patients	◯	◯	◯	◯	◯
Earlier interventions and personalised care	◯	◯	◯	◯	◯
Enabling research into new treatments	◯	◯	◯	◯	◯
Lifetime genomic record for the patient	◯	◯	◯	◯	◯
Increased awareness of genetic conditions among the public and healthcare professionals	◯	◯	◯	◯	◯
Long-term cost-effectiveness to the NHS	◯	◯	◯	◯	◯

Which potential benefit do you believe would be the most significant?

◯ Earlier diagnosis
◯ Enabling a diagnosis to be reached in more patients
◯ Earlier interventions and personalised care
◯ Enabling research into new treatments
◯ Lifetime genomic record for the patient
◯ Increased awareness of genetic conditions among the public and healthcare professionals
◯ Long-term cost-effectiveness to the NHS

Please describe any other potential benefits of newborn WGS.

_____

Drawbacks of newborn WGS

What do you think are some of the main potential drawbacks of introducing routine WGS in newborn screening?

Please rate the importance of the following potential drawbacks of newborn WGS:

	(Not important) 1	2	(Neutral) 3	4	(Very important) 5
Screening for a wider range of conditions with WGS so soon after birth could interfere with the early bonding process	○	○	○	○	○
Emotional impact on parents/carers in receiving an early genetic diagnosis who otherwise appears healthy	○	○	○	○	○
Upskilling the workforce	○	○	○	○	○
Parents consenting to obtaining information that has the potential to impact the newborn at any point throughout their lifetime	○	○	○	○	○
Risk that parents are falsely reassured by a normal report	○	○	○	○	○
Risk of incidental or uncertain findings	○	○	○	○	○
Identifying conditions for which effective treatments aren't available	○	○	○	○	○
Lack of resource in the NHS to support families (e.g. counselling)	○	○	○	○	○
Potential for future discrimination on the basis of genomic information (e.g. insurance implications)	○	○	○	○	○
Privacy and security of data	○	○	○	○	○
Cost effectiveness	○	○	○	○	○

Which potential drawback do you believe would be the most significant?

- ◯ Screening for a wider range of conditions with WGS so soon after birth could interfere with the early bonding process
- ◯ Emotional impact on parents/carers in receiving an early genetic diagnosis who otherwise appears healthy
- ◯ Upskilling the workforce
- ◯ Parents consenting to obtaining information that has the potential to impact the newborn at any point throughout their lifetime
- ◯ Risk that parents are falsely reassured by a normal report
- ◯ Risk of incidental or uncertain findings
- ◯ Identifying conditions for which effective treatments aren't available
- ◯ Lack of resource in the NHS to support families (e.g. counselling)
- ◯ Potential for future discrimination on the basis of genomic information (e.g. insurance implications)
- ◯ Privacy and security of data
- ◯ Cost-effectiveness

Please describe any other potential drawbacks of newborn WGS.

Overall, do you support the introduction of WGS to the newborn screening programme?

Definitely not — Neutral — Definitely yes

(Place a mark on the scale above)

How relevant do you see the newborn WGS programme to your future medical practice?

Not at all relevant — Neutral — Extremely relevant

(Place a mark on the scale above)

Please explain your answer to the previous two questions.

If you have any final comments or thoughts regarding the introduction of WGS to the newborn screening programme, please write them here.