NIHR | Health Protection Research Unit in Gastrointestinal Infections at University of Liverpool

Thoughts and perspectives of metagenome sequencing as a diagnostic tool for infectious disease: An interpretive phenomenological study

Hannah Trivett[1], Oyinlola Oyebode[2], Alistair Darby[1]

1. University of Liverpool, 2.Queen Mary University of London

Twitter:
@HannahTriv

Email:
hlhtrive@liverpool.ac.uk

INTRODUCTION

- There is a gap in the scientific literature which contextualizes the current clinical advancements in infectious disease diagnostics and the barriers that hinder the implementation of new diagnostic frameworks.
- This study explores stakeholders' experiences within infectious disease diagnostics settings and researchers at the forefront of microbial genomics to unpack the factors driving the development and implementation of metagenome sequencing.

MATERIAL & METHODS

Participant recruitment	Purposefully selecting 10 participants, with varying degrees of knowledge and experience in infectious disease diagnostics and genomics, in England (Figure 1, Table 1).
Data collection	Semi-structured interviews recorded via Teams, to gain insight into the participants experience and knowledge of the topic. A question sheet facilitated open ended discussions between the researcher and participant.
Qualitative analysis	Thematic analysis was conducted, looking for convergence and divergence between participant experiences to form the project themes. The analysis was audited by a supervisor of the project to ensure reliability Interpretive phenomenological guidelines were followed.

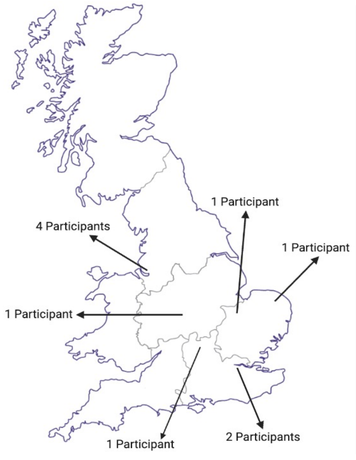


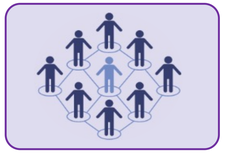
Figure 1- UK map outlining geographical locations of participants

Participant number	Sector	Knowledge of genome sequencing
1	Clinical based	Expert knowledge
2	Academia based	Expert knowledge
3	Clinical based	Some knowledge
4	Clinical based	Good Knowledge
5	Academia based	Expert knowledge
6	Clinical based	Good Knowledge
7	Clinical based	No Knowledge
8	Clinical based	No knowledge
9	Academia based	Expert knowledge
10	Academia based	Expert knowledge

Table 1- Participant characteristics, surrounding genome sequencing as an infectious disease diagnostic.

RESULTS

10 individuals were interviewed between July 2021 and October 2021, including Clinical scientists, consultants, and professors in academia, with 60% identifying as male and 40% as female. 5 themes emerged from the transcripts and were identified as: 'Barriers to implementation, 'Communication', 'COVID-19', 'Diagnostic choice', and 'Open access and data sharing'.



Communication

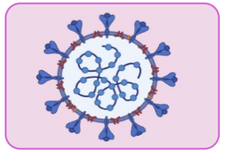
"I suppose you need to align the funding bodies. We need to bring in all the stakeholders as equal partners…think it needs an insightful, joined up, sort of mutually supporting and engaging partnership framework so that everyone can benefit from it."

Diagnostic Choice

There is a steady stream of the introduction of new things to do in response to clinical needs. However, there is a set of technologies that are finding it harder to get into the clinical place for infectious diseases of which genome sequencing is one."

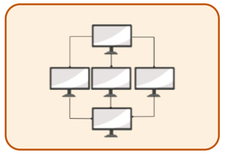
Barriers to implementation

"If we can't progress next Gen sequencing now into a routine diagnostic service, then something is wrong… it's still so specialized and should it really be?... We just don't seem to be able to get across that hurdle."



COVID-19

"SARS-CoV-2 is an exemplary example of where sequencing can bring impact to public health and individuals so it's really gone beyond proof, as it is being used every day"



Open access and data sharing

"There's a culture clash there between whether are genomes really a public good, which should be deposited in the public database, as soon as you get them versus people's individual rights to privacy for their diagnosis."

CONCLUSION

- The study filled a gap in the literature around what is known about the development and implementation of metagenome sequencing for routine clinical use.
- The study provided evidence of the enthusiasm for new diagnostics in infectious diseases laboratories and offered the opportunity for participants to express ways in which metagenomics could overcome current laboratory diagnostic problems.
- Participants highlighted that it is fundamental to introduce metagenome sequencing into clinical laboratories to move diagnostics into the Next-generation sequencing era.

IMPLICATIONS

Research	The study provided clear guidance for user requirements from new diagnostic technologies, to incorporate into the current diagnostic landscape.
Policy	Communication between stakeholders must improve for the development and distribution of genomic technologies, to meet the needs for clinical users for a positive public health impact.
Practice	Participants experiences of current diagnostic workflows and the bottleneck within them, provided case evidence of the utility genomics to supplement current diagnostic workflows.

REFERENCES

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