Deliverable

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Explanation according to GA Annex I:

Publication of the EBCD method used within the initial two ERNs in order to provide the opportunity for the other 22 ERNs to adopt the findings or follow a similar process to co-design their own intervention or services.

The following report is based on a peer-reviewed publication in the *Journal of Community Genetics*. The full citation for the paper is:

Costa A, Franková V, Robert G, Macek M, Patch C, Alexander E, Arellanesova A, Clayton-Smith J, Hunter A, Havlovicová M, Pourová R, Pritchard M, Roberts L, Zoubková V, Metcalfe A. Co-designing models for the communication of genomic results for rare diseases: a comparative study in the Czech Republic and the United Kingdom. *J Community Genet*. 2022 Jun;**13**(3):313-327. doi: 10.1007/s12687-022-00589-w. Epub 2022 May 6. Erratum in: J Community Genet. 2022 Jul 1

Abstract:

The communication of genomic results to patients and families with rare diseases raise distinctive challenges. However, there is little evidence about optimal methods to communicate results to this group of service users. To address this gap, we worked with rare disease families and health professionals from two genetic/genomic services, one in the United Kingdom and one in the Czech Republic, to co-design that best meet their needs. Using the participatory methodology of Experience-Based Co-Design (EBCD), we conducted observations of clinical appointments (n=49) and interviews with family participants (n=23) and health professionals (n=22) to gather their experience of sharing/receiving results. The findings informed a facilitated co-design process, comprising 3 feedback events at each site and a series of meetings and remote consultations. Participants identified a total of four areas of current service models in need of improvement, and co-designed six prototypes of quality improvement interventions. The main finding was the identification of post-test care as the shared priority for improvement for both health professionals and families at the two sites. Our findings indicate the need to strengthen the link between diagnostics (whether or not a pathogenic variant is found) and post-test care, including psychosocial and community support. This raises implications for the reconfigurations of genomic service models, the redefinition of professional roles and responsibilities and the involvement of rare disease patients and families in health care research.

Introduction:

The communication of genomic results to rare disease patients and their families presents distinctive challenges due to both the mode of testing and the needs of this particular group of service users. As opposed to testing targeting single genes or gene ‘panels’ of limited size, genomic testing involves examination of a much larger amount of an individual’s DNA, meaning there is wider room for uncertainty about possible findings and their implications for the individual family. This means that established challenges related to the disclosure of genetic information not only remain relevant in the context of genomics, but are likely to be exacerbated (Eisler et al. 2016; Dheensa et al. 2017; Metcalfe et al. 2011; Hallowell et al. 2006). Moreover, genomic testing also raises novel and unique issues for genetic counselling. The communication of inconclusive and uncertain results has been identified as an area of concern (Clift et al. 2020; Mighton et al. 2021; Skinner et al. 2018; Fenton et al. 2020; Bartley et al. 2020, 2021; Han et al. 2017). The limitations for informed consent and pre-test counselling have also been highlighted, as the uncertainty surrounding genomic results means it is increasingly difficult to ensure that families can be prepared for the testing possible outcomes (Horton and Lucassen 2019; Samuel et al. 2017; Newson et al. 2016).

Compounding these general issues are the unique challenges associated with rare disease diagnostics. The rarity and heterogeneity of rare diseases, and their chronic and progressive nature can have a significant psychosocial impact on families. Genomic testing is crucial to ensure families can receive a timely and accurate diagnosis, but can only alleviate the burden of living with a rare disease (Rosell et al. 2016). With or without a diagnosis, families often face significant financial, social and emotional challenges, affecting their quality of life and their capacity to access high quality care (Pelentsov et al. 2015; von der Lippe et al. 2017). However, there is scarce evidence on what model of genomic services might best meet patients' and families' needs. Understanding the implications of these issues will be crucial to maximise the benefits of genomic sequencing as this rapidly moves from research into routine care (Stark et al. 2019; 100000 Genomes Project Pilot Investigators 2021),

To help collect cross-national responses to these issues, we worked with families of patients with rare diseases and health professionals to co-design communication models based on their experience of sharing/receiving genomic results. The study was conducted as part of the EU Horizon 2020 research project Solve RD (http://solve-rd.eu) and was set in two genetic/genomic services in the United Kingdom (UK) and in the Czech Republic. We used the participatory methodology of Experience-Based Co-Design (EBCD), which draws on design thinking to enable service users to become integral to the process of service design and quality improvement (Bate and Robert 2007; Robert et al. 2021). The approach was initially developed and piloted in a Head & Neck Cancer service in an English acute hospital in 2004–2005, and has subsequently been implemented in over a hundred different services in approximately ten countries, as well as being used to help develop complex interventions (Donetto et al. 2014; Robert et al. 2021). Using EBCD, we worked with families and health professionals at the two participating services to explore their experience, identify areas of current models in need of improvement and co-design quality improvement interventions.

In this article, we present and compare the EBCD process and outcomes at the two sites. We focus in particular on the issue of post-test care, as this was the key priority for improvement that was identified by both health professionals and families at both sites. The findings can be adapted to improve the communication of genomic results in other countries and health care settings.

Report:

Please see: Costa, A., Franková, V., Robert, G. et al. Co-designing models for the communication of genomic results for rare diseases: a comparative study in the Czech Republic and the United Kingdom. J Community Genet 13, 313–327 (2022). https://doi.org/10.1007/s12687-022-00589-w

Conclusion:

To the best of our knowledge, this was the first study to use the EBCD methodology with families of rare disease patients in the context of genomic health services (Donetto et al. 2014). Our findings highlighted the need of improving post-test care as an integral part of future genomic health service, and the importance of involving rare disease families and health professionals as active participants in the process of service design and quality improvement. The immediate contributions of genomics to health care are likely to be the greatest in the field of rare diseases. Improving diagnostics remains essential, but will not, in and of itself, suffice to maximise the benefits of genomic testing in medical care. Importantly, involving affected families together with health professionals will be crucial to the design of future genomic services that best meet their evolving needs.