Policy Ethics And Life Sciences

Rare Disease, Big Data:
the views of rare disease patients on systems for sharing data and biospecimens

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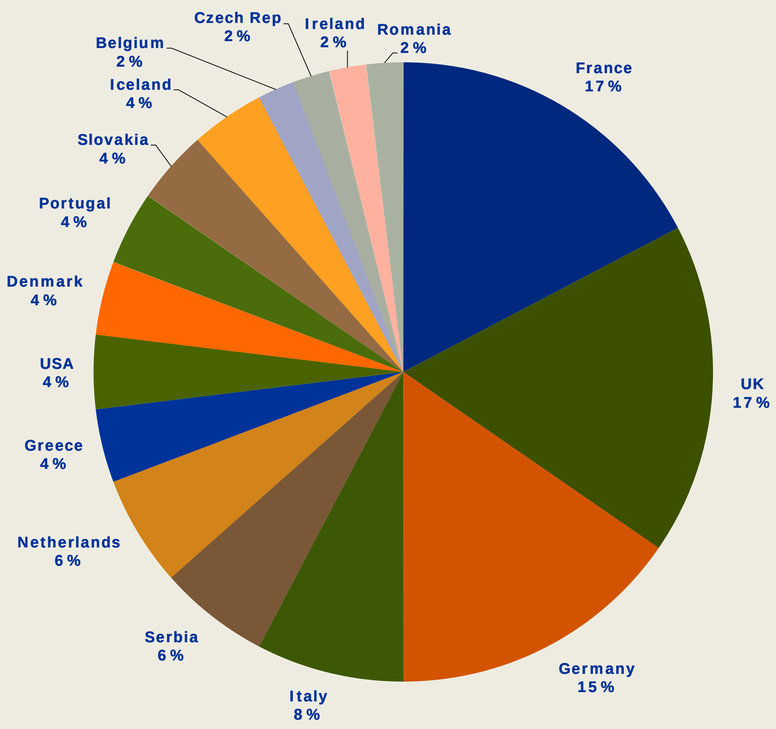
Background

Within the myriad articles about participants' opinions of genomics research, the views of a distinct group – people with a rare disease (RD) – are unknown. It is important to understand if their opinions differ from the general public by dint of having a rare disease and vulnerabilities inherent in this. Here, we document RD patients' attitudes to participation in genomics research, particularly around large-scale, international data and biosample sharing. This work is unique in exploring the views of people with a range of rare disorders from many different countries[1].

Methodology

As part of RD Connect – an integrated platform connecting databases, registries, biobanks and clinical bioinformatics for rare disease research – we convened five focus groups made up of 52 rare disease patients and advocates from 16 different countries. We used scenarios to encourage free-form discussion, and the raising of issues and opinions that were of importance to the participants, rather than topics being chosen by the researchers. Participants explored what consent and information provision is expected when researchers are using historic samples and the preferences of patients when health and genomic data are widely shared beyond a local research team or care provider.



Focus group participants by country

The need to share

Members of the focus groups were positively disposed towards participating in research and allowing their own data and biosamples, as well as those of any affected children, to be shared internationally. All the participants understood the incentive for RD in sharing data and samples, in fact there were several pleas for research systems to be standardised across the EU in order to make data sharing easier. Participants would give broad consent to their/their child's data and samples being shared internationally.

Sharing versus risk

There was a strong notion among the discussants of personal choice, in that participants take decisions which they deem correct for themselves or their child and which they feel comfortable with. Risk versus benefit decisions can be informed by the characteristics of the disease, for example one mother whose child has a life limiting, progressive disorder was prepared to be more permissive regarding the use of data than a mother whose child had an intellectual disability and a normal life span. The former wanted to use all avenues possibly to move quickly towards a cure, whilst the latter was concerned about discrimination and used the example of past cultural norms of societal exclusion of people with intellectual disability as a reason to proceed with caution in sharing data which identifies such difference.

Most participants had some concerns about risk of stigma and discrimination against themselves or their children which they perceive could have a negative impact on access to healthcare and other opportunities in life. They would therefore look to a global, data sharing platform to safeguard against this and to mitigate, as far as possible, against uncertainties which might lead to their data "getting into the wrong hands".

Participants emphasised the following characteristics for a global platform

Consent: participants are protective of their autonomy and see consent as a social, reciprocal agreement where they expect to be re-consented when use falls outside of the original consent. Also expect children to be re-contacted when they come of age.

Governance: favour systems which are managed by public or not-for-profit institutions. Patient inclusion in governance was seen as crucial.

Access: should be limited to people working in health care and health research. Individual's activity should be tracked.

Management and security: information provided should be specific, easily available and transparent about procedures.

Conclusions

Rare disease patients and parents see their dealings with researchers as motivated by common interests and participants are keen to make their samples and data available to researchers as long as this action is treated with respect and reciprocity.

As their data and samples are shared and used by researchers around the world, participants could perceive that the locus of influence has shifted and their control over information about their and their family's health is diminished. Hence the strongly expressed desire to protect their and their children's autonomy, and to protect against lapses in privacy and the potential for subsequent discrimination.

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1. McCormack P, Kole A, Gainotti S, Mascalzoni D, Molster C, Lochmüller H, Woods S. 'You should at least ask'. The expectations, hopes and fears of rare disease patients on large-scale data and biomaterial sharing for genomics research. *European Journal of Human Genetics* 2016, **30**.