



Aalborg Universitet

AALBORG UNIVERSITY  
DENMARK

## A Lens for Evaluating Genetic Information Governance Models

Skorve, Espen; Vassilakopoulou, Polyxeni; Aanestad, Margunn; Grünfeld, Thomas

*Published in:*  
Informatics for Health

*DOI (link to publication from Publisher):*  
[10.3233/978-1-61499-753-5-298](https://doi.org/10.3233/978-1-61499-753-5-298)

*Creative Commons License*  
CC BY-NC 4.0

*Publication date:*  
2017

*Document Version*  
Publisher's PDF, also known as Version of record

[Link to publication from Aalborg University](#)

*Citation for published version (APA):*  
Skorve, E., Vassilakopoulou, P., Aanestad, M., & Grünfeld, T. (2017). A Lens for Evaluating Genetic Information Governance Models: Balancing Equity, Efficiency and Sustainability. In . R. Randell, R. Cornet, C. McCowan, N. Peek, & P. J. Scott (Eds.), *Informatics for Health: Connected Citizen-Led Wellness and Population Health* (pp. 298-302). IOS Press. *Studies in Health Technology and Informatics*, Vol.. 235, DOI: 10.3233/978-1-61499-753-5-298

### General rights

Copyright and moral rights for the publications made accessible in the public portal are retained by the authors and/or other copyright owners and it is a condition of accessing publications that users recognise and abide by the legal requirements associated with these rights.

- ? Users may download and print one copy of any publication from the public portal for the purpose of private study or research.
- ? You may not further distribute the material or use it for any profit-making activity or commercial gain
- ? You may freely distribute the URL identifying the publication in the public portal ?

### Take down policy

If you believe that this document breaches copyright please contact us at [vbn@aub.aau.dk](mailto:vbn@aub.aau.dk) providing details, and we will remove access to the work immediately and investigate your claim.

# A Lens for Evaluating Genetic Information Governance Models: Balancing Equity, Efficiency and Sustainability

Espen SKORVE<sup>a,1</sup>, Polyxeni VASSILAKOPOULOU<sup>b</sup>, Margunn AANESTAD<sup>c</sup>,  
Thomas GRÜNFELD<sup>d</sup>

<sup>a</sup>Aalborg University, Denmark

<sup>b</sup>University of Agder, Norway

<sup>c</sup>University of Oslo, Norway

<sup>d</sup>Oslo University Hospital, Norway

**Abstract.** This paper draws from the literature on collective action and the governance of the commons to address the governance of genetic data on variants of specific genes. Specifically, the data arrangements under study relate to the *BRCA* genes (*BRCA1* and *BRCA2*) which are linked to breast and ovarian cancer. These data are stored in global genetic data repositories and accessed by researchers and clinicians, from both public and private institutions. The current *BRCA* data arrangements are fragmented and politicized as there are multiple tensions around data ownership and sharing. Three key principles are proposed for forming and evaluating data governance arrangements in the field. These principles are: equity, efficiency and sustainability.

**Keywords.** Genetic information, governance, the commons.

## 1. Introduction

The significance of genetic testing for clinical purposes is increasing, and the rapid advances in sequencing technologies are currently amplifying this development. One outcome of this is the exponential growth in genetic data generation. New data are continuously produced as output of genetic analyses performed all over the world. The new data are subsequently used as an essential input for further analyses for research and clinical purposes alike. In this paper, we focus on data related to the (potential) pathogenicity of variants of specific genes. These data relate to single genes, they are completely anonymous, and do not raise any privacy issues. They are valuable for taking clinical decisions related to diagnostics, treatments and prevention [1][2][3]. Their importance for decision taking is making it urgent to address data governance in the domain addressing the tensions, contestations and controversies around data ownership and control. These tensions are shaping the currently fragmented and politicized landscape of gene-specific data repositories. The current situation is making it difficult to reap the benefits of the increased speed and reduced cost associated with

---

<sup>1</sup> Corresponding Author: Espen Skorve, Department of Computer Science, Aalborg University, Selma Lagerlöfs Vej 300, 9220 Aalborg East, Denmark. E-mail: [espesko@cs.aau.dk](mailto:espesko@cs.aau.dk)

new sequencing technologies [4][5]. In order to address this issue, we need a perspective that can be conducive to reaching resolutions within such a complex domain. In this paper, we draw from theoretical resources on “the commons” that can be used as a basis for resolving governance issues and for facilitating a sustainable development within the field.

We turn to the theory of the commons [6][7] aiming to draw concepts for addressing “*the age-old problem of how to induce collaborative problem solving and other forms of collective action among self-interested individuals, groups, or organizations, assuming, of course, that they share at least some common goals*” [8]. In this body of literature, a basic assumption is that different types of resources are subject to different governance considerations, and a common taxonomy for discussing this is drawn along the axis of subtractability and exclusion [9] [10]. Based on a claim that knowledge about the human genome belongs to mankind as a whole, current attempts at privatizing data that can contribute to this knowledge have been characterized as a second enclosure movement [11]<sup>2</sup>. We argue that biomedical knowledge in general, and genetic knowledge in particular, is of such a character that it constitutes what we call a public good. Hence, we adopt three fundamental concepts for evaluating any regime aiming to govern data that can contribute this knowledge resource: equity, efficiency and sustainability [10].

## 2. Case Background and Method

A typical example of the genetic data governance challenges can be found in the specialized field of breast cancer genetics, and the way this has evolved over the two decades that passed since the identification of the *BRCA* genes. The *BRCA* genes (*BRCA1* and *BRCA2*) are related to susceptibility to breast and ovarian cancer. Multiple data repositories containing information regarding the pathogenicity associated with specific variants of these two genes have emerged during that past decades. A main distinction between these repositories is how contribution and access rights are organized and governed, and their differences are most often related to the degree of openness. The Breast Cancer Information Core (BIC) was the first open access repository of *BRCA* data (it was established in 1995). This is a shared repository where information generated from research or clinical practice, within private and public laboratories all over the world can be deposited and retrieved. In the U.S.A, one private laboratory (Myriad Laboratories) was able to establish a dominant position in the local market for *BRCA* testing after having been granted several patents based on its central role in the discovery of the *BRCA* genes. This laboratory was the primary contributor to the BIC database for several years, and the ‘*BRCA* community’ was fairly dependent on Myriad’s sharing of knowledge and information. When Myriad decided to discontinue their contributions to BIC in 2004, this was therefore a major event that caused several counter actions aimed at amending the problems arising from significant information being disclosed from the community. Through initiatives like the ‘Sharing Clinical Reports’ and ‘Free the Data’ projects, new open access repositories were established, and doctors and patients were encouraged to register the detailed results from their lab tests at Myriad’s [12][13]. Furthermore, several other initiatives have emerged on both

---

<sup>2</sup> The privatization of common land in Europe during the 15th century constitutes the first enclosure movement [11].

sides of the Atlantic. The Leiden University Medical Center (Netherlands) launched 'LOVD' as early as 2005, and 'ClinVar' was released by the US National Center for Biotechnology Information (NCBI) in 2012. In 2015, the 'BRCA Mutation Database' (by the University of Utah's Department of Pathology and the ARUP Laboratories, USA), the 'BRCA Share' repository (INSERM and Quest Diagnostics, France) and the repository 'BRCA Exchange' (by the Global Alliance for Genomics and Health) were launched. The latter is particularly interesting, as it constitutes a global, networked initiative (as indicated by the name), and thus most strongly manifests the general tendency of initiatives the span the boundaries of single, local or even national actors. However, this increasing multiplicity of repositories and initiatives in the domain results in duplication of efforts and increased difficulty in data retrieval.

The research reported in this paper is designed as a case study [14] with focus on the shaping of governance arrangements for information related to the (potential) pathogenicity of *BRCA* variants. We collected empirical material over a two years' period as part of the activities of a research and development project that aimed to develop a secure IT platform to facilitate distributed collaboration and access to genetic information. We performed 12 interviews with experts in the domain and we reviewed more than 100 documents (journal papers, specialised press reports, commercial announcements) to identify key events that mark the evolution of governance arrangements in *BRCA* domain. We adopted an interpretive approach for the analysis of the data [15] going through transcripts, notes and documents in order to identify relevant themes. We structured our analysis around the key theoretical concepts of equity, efficiency and sustainability. Initial findings were refined and verified based on continuous communications with practitioners in the field of genetic analysis.

### **3. Addressing Information Governance Based on a Commons Perspective**

#### *3.1. Equity*

Any public good is subject to some sort of distribution of rights and obligations amongst its stakeholders. Who does the work related to growth and maintenance, and who gets to benefit from what the resource has to offer? Is the resource available to those who need it, where and when they need it? Whose voices are heard when decisions are made that affect larger parts of the stakeholder community? Indeed, the question of who is considered to have a legit stake is in itself an important question in the context of a public good resource. While basic democratic values are obviously relevant, these issues are also pivotal in ensuring a wide commitment when few sanctions are available to force adherence to a common governance regime. The knowledge produced in the Myriad Laboratories is built on – and thus inherently part of – the global body of scientific knowledge about human genetic variations and their clinical significance. When they refuse to share their data that can contribute to further advancing the knowledge, they breach the commons' logic. The community's response to Myriads enclosure policy is a reaction to the perceived inequity in the distribution of benefits (enclosure means that one key actor can reap disproportionately more benefits) and incurred costs (the generation of valuable data today is built upon the accumulated efforts and resources invested for many decades around the world).

### 3.2. Efficiency

For a governance arrangement to reach widespread support, it must provide an acceptable level of resource access and quality, at an acceptable cost, to members of the community. An inefficient arrangement will inevitably create a sense of a non-working system that might undermine its legitimacy and subsequent support. A particular challenging aspect of this is aligning, or balancing potentially diverse – and sometimes contradictory – values and requirements amongst different stakeholder groups. For instance, while uncertainty related to the clinical significance of a genetic variant is problematic for clinicians, it might represent a potential research question for the researchers. An efficient governance regime will have to make sure that such issues are reflected in the knowledge resource and its representations when possible.

### 3.3. Sustainability

Though a knowledge resource cannot be depleted, its long-term trajectory is normally subject to considerable dynamics. For a relatively novel field such as that of human genetic variants, the creation and addition of new knowledge is a main part of the dynamics. The evolution of knowledge and technology mutually drive each other. How can access, quality and costs be maintained within acceptable limits in this context of rapid change? In what direction is the trajectory moving and what are the issues driving and hampering its development? As new knowledge is primarily built on existing knowledge, an enclosure policy represents a potential threat to sustainability. Single actors growing too dominant can also undermine the community's trust and commitment, which in turn can jeopardise a responsible long term development. In the field of genetic knowledge, the continuous addition of new actors and their contributions and requirements is also an aspect that must be catered for in a long term perspective.

## 4. Concluding Remarks

The commons perspective advocates governance arrangements that favour the community as a whole while still allowing single stakeholders or groups of stakeholders to pursue their own interests. The ever increasing complexity related to the formation, maintenance and propagation of genetic and other biomedical knowledge will inevitably force the emergence of such arrangements. Fulk et al. explore the role of connective and communal public goods in discretionary repositories [8]. Information is discussed there as a hybrid (neither private nor public) good, where public benefit is achieved by individuals or companies acting out of their private interests. In the case discussed here the balancing of private interests is still in flux and this creates tensions for sustaining the discretionary repositories. It is our contention that a level of abstraction, where the various stakeholders can reach a common understanding and a shared goal, is a prerequisite for establishing a working governance regime. Based on the commons perspective, we have suggested *equity*, *efficiency* and *sustainability* as such an abstraction. As key principles, they may serve as a platform for translations and operationalisations into governance arrangements that will work for the community they are supposed to serve.

## Acknowledgements

The authors would like to thank Morten Christoph Eike for fruitful discussions and for commenting on the manuscript.

## References

- [1] C. Bennett et al., A toolkit for incorporating genetics into mainstream medical services: Learning from service development pilots in England, *BMC health services research* **10.1** (2010), 125.
- [2] H. Skirton, C. Patch and J. Williams, *Applied genetics in healthcare: A handbook for specialist practitioners*, Garland Science, 2005.
- [3] R. Snyderman, Personalized health care: From theory to practice, *Biotechnology Journal* **7.8** (2012), 973-79.
- [4] D. Gomez-Cabrero et al., Data integration in the era of omics: current and future challenges, *BMC systems biology* **8.2** (2014), 11.
- [5] M. Shabani, B.M. Knoppers and P. Borry, From the principles of genomic data sharing to the practices of data access committees, *EMBO molecular medicine* **7.5** (2015), 507-509.
- [6] E. Ostrom, *Governing the Commons: The Evolution of Institutions for Collective Action*, Cambridge University Press, Cambridge UK, 1990.
- [7] E. Ostrom, J. Walker and R. Gardner, *Rules, games, and common-pool resources*, University of Michigan Press, Michigan US, 1994.
- [8] J. Fulk, et al., Connective and Communal Public Goods in Interactive Communication Systems, *Communication Theory* **6.1** (1996), 60-87.
- [9] M.D. McGinnis, An introduction to IAD and the language of the Ostrom workshop: a simple guide to a complex framework, *Policy Studies Journal* **39.1** (2011), 169-183.
- [10] C. Hess and E. Ostrom, A framework for analyzing the knowledge commons, in C. Hess and E. Ostrom (eds.), *Understanding Knowledge as a Commons: from Theory to Practice*, MIT Press, Massachusetts US, 2006.
- [11] J. Boyle, The second enclosure movement and the construction of the public domain, *Law and contemporary problems* **66.1/2** (2003), 33-74.
- [12] S. Nguyen and S.F. Terry, Free the data: the end of genetic data as trade secrets, *Genetic testing and molecular biomarkers* **17.8** (2013), 579-80.
- [13] G. Kolata, DNA Project Aims to Make Public a Company's Data on Cancer Genes, *The New York Times* (April 12 2013). <[http://www.nytimes.com/2013/04/13/health/dna-project-aimsto-make-companys-data-public.html?pagewanted=all&\\_r=0](http://www.nytimes.com/2013/04/13/health/dna-project-aimsto-make-companys-data-public.html?pagewanted=all&_r=0)>, accessed 15 September 2014.
- [14] R.K. Yin, Case study research design and methods third edition. *Applied social research methods series* **5** (2003).
- [15] G. Walsham, Doing interpretive research. *European journal of information systems* **15.3** (2006), 320-330.