Ethical and governance considerations for genomic data sharing in the development of medical technologies for melanoma - The iToBoS Project.

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Short Abstract

Balancing the risks and benefits of using genomics data in health service provision is a complex task. Social, ethical, and legal considerations are nuanced, often complicated by the fact that regulations lag behind rapid pace of technological development. Ethical considerations such as autonomy, beneficence, and non-maleficence are weighed against (and within) complex concepts such as privacy, security, safety, and proportionality. This paper will discuss European H2020 funded project IToBoS[1], in which an AI diagnostic platform for the early detection of melanoma is being developed. Assuring the project's solutions are produced in an ethically and socially responsible manner, with regulatory compliance at their core, is one of the project's primary goals. This paper will communicate the existing tensions within the health sector, including between the European Commission's desire for open-data – governed through its proposed Digital Strategy and practically achieved through the creation of a European Health Data Space[2] – and the risks inherent with the generalised sharing of genomics (and other health related) data.

Extended Abstract

Introduction

Balancing the risks and benefits of using genomics data in health service provision is a complex task. Social, ethical, and legal considerations are nuanced, often complicated by the fact that regulations lag behind the rapid pace of technological development. Ethical considerations such as autonomy, beneficence, and non-maleficence are weighed against (and within) complex concepts such as privacy, security, safety, and proportionality. With the potential for privacy violations, discrimination, financial exploitation and algorithmic bias, information revealed by genomic methods is sensitive in most (if not all) situations.[3] However, excessively rigid regulations governing the creation, sharing, and use of genomic information limit potential advantages.[4] Recent studies on the use of genetic data have revealed that the most often mentioned concerns by patients and caregivers were "lack of security," "control over information access," and "extraction of information beyond the

[1] This project has received funding from the European Union's Horizon 2020 research and innovation programme under grant agreement No. 965221. More information may be found at: https://itobos.eu/
[2] https://health.ec.europa.eu/ehealth-digital-health-and-care/european-health-data-space_en
[3] Clayton, E. W., Evans, B. J., Hazel, J. W., & Rothstein, M. A. (2019). The law of genetic privacy: applications, implications, and limitations. *Journal of Law and the Biosciences*, *6*(1), 1-36.
[4] World Health Organization. (2022). Accelerating access to genomics for global health: promotion, implementation, collaboration, and ethical, legal, and social issues: a report of the WHO Science Council.

research objectives".[5] That said, the majority of respondents reported the "discovery of curative treatment" as the most important factor (*ibid*). This paper will discuss project IToBoS[6], in which an AI diagnostic platform for the early detection of melanoma is being developed. A total body scanner forms part of the platform, along with a computer-aided diagnostics (CAD) tool that incorporates relevant patient data including genetic information. Assuring the project's solutions are produced in a responsible manner, with regulatory compliance at their core is a stated goal of the project. It seems relatively straightforward, but achieving the goal is not – especially when research tasks are considered alongside a rapidly evolving health sector. At the heart of the problem is the tension between the European Commission's desire for open-data – governed through its proposed Digital Strategy, practically achieved through the creation of a European Health Data Space[7] – and risks inherent with the generalised sharing of genomics data.[8] This paper will communicate some of the existing tensions through the contextual lens of the iToBoS project, culminating in the acknowledgement that a generally accepted Code of Conduct (CoC) for genomics data is required.

Genomics Data – Sharing, Privacy and Governance

Genomics (or genetic) data is, from a European perspective, classified as personal data. A recital within the current data protection regulation defines the type: "Genetic data should be defined as personal data relating to the inherited or acquired genetic characteristics of a natural person which result from the analysis of a biological sample from the natural person in question, in particular chromosomal, deoxyribonucleic acid (DNA) or ribonucleic acid (RNA) analysis, or from the analysis of another element enabling equivalent information to be obtained."[9] Dankar et al. outlined four broad risks associated with the use of genomics data – 1. Distinguishability, 2. Genetic conditions and predispositions, 3. Ancestral implications, and 4. Potentially hidden information.[10] Additionally, the authors state technical approaches to privacy are currently inadequate when applied to genomics data, and that privacy laws are not keeping pace with the evolution of understanding - a view echoed by privacy researchers. European Commission initiatives have focused on understanding legal barriers and governance structures[11], developing an open-sharing roadmap entitled '1+ Million Genomes

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[5] Amorim, M., et al (2022). Benefits and Risks of Sharing Genomic Data for Research: Comparing the Views of Rare Disease Patients, Informal Carers and Healthcare Professionals. *International Journal of Environmental Research and Public Health*, *19*(14), 8788

[6] This project has received funding from the European Union's Horizon 2020 research and innovation programme under grant agreement No. 965221. More information may be found at: https://itobos.eu/

[7] https://health.ec.europa.eu/ehealth-digital-health-and-care/european-health-data-space_en

[8] van Kessel, R., Wong, B. L. H., Forman, R., Gabrani, J., & Mossialos, E. (2022). The European Health Data Space fails to bridge digital divides. British Medical Journal, 378, e071913

[9] REGULATION (EU) 2016/679 OF THE EUROPEAN PARLIAMENT AND OF THE COUNCIL of 27 April 2016 on the protection of natural persons with regard to the processing of personal data and on the free movement of such data, and repealing Directive 95/46/EC (General Data Protection Regulation), Recital 34.

[10] Dankar et al. Human Genomics (2018) 12:19 https://doi.org/10.1186/s40246-018-0147-5

[11] Hansen, J., Wilson, P., Verhoeven, E., Kroneman, M., Kirwan, M., Verheij, R., & van Veen, E. B. (2021). Assessment of the EU Member States' rules on health data in the light of GDPR. Specific Contract No SC 2019 70 02 in the context of the Single Framework Contract Chafea/2018/Health/03

Initiative' (1+MG)[12], drafting a broad health data space Regulation[13], and the publication of a formal opinion on specific data protection implications.[14] Further back, the European Society for Human Genetics (ESHG) outlined potential risks of opportunistic genomic screening (OGS) and secondary findings (SF). Their position rested on a number of distinct recommendations.[15] Accordingly, the European 1+MG roadmap covered four distinct 'dimensions': Governance, Trust Framework, Infrastructure, Data.

Figure 1. 1+MG Roadmap Overview[16]

Alongside this, researchers have communicated the distinct requirement for a genomics specific CoC – to act as a framework to guide the field and provide dimensional interoperability across jurisdictions. The CoC would work alongside frameworks developed through the 1+MG initiative, and are proposed to act as a tool to establish best practice and support compliance with GDPR.[17]

iToBoS and a PIA+

Within the iToBoS project, specific concerns related to genomics are discussed through the conduct of a privacy impact assessment (PIA), which considers risks based on an ISO PIA standard (ISO/IEC 29134:2017).[18] The process is intended to help minimise potential risks, while signposting future (post-project) concerns. The process supports the pursuit of

[12] The '1+ Million Genomes' (1+MG) initiative aims to enable secure access to genomics and the corresponding clinical data across Europe for better research, personalised healthcare and health policy making. Mor information is available at: https://digital strategy.ec.europa.eu/en/policies/1-million-genomes
[13] Proposal for a regulation - The European Health Data Space COM(2022) 197/2, available at: https://health.ec.europa.eu/publications/proposal-regulation-european-health-data-space_en
[14] EDPB-EDPS Joint Opinion 03/2022 on the Proposal for a Regulation on the European Health Data Space, https://edpb.europa.eu/our work-tools/our-documents/edpbedps-joint-opinion/edpb-edps-joint-opinion-032022-proposal_en
[15] de Wert, G., Dondorp, W., Clarke, A., Dequeker, E., Cordier, C., Deans, Z., ... & Forzano, F. (2021). Opportunistic genomic screening. Recommendations of the European society of human genetics. European Journal of Human Genetics, 29(3), 365-377.
[16] The 1+MG Roadmap, available at: https://digital-strategy.ec.europa.eu/en/policies/1-million-genomes
[17] Matar, A., Hansson, M., Slokenberga, S., Panagiotopoulos, A., Chassang, G., Tzortzatou, O., Pormeister, K., Uhlin, E., Cardone, A., & Beauvais, M. (2022). A proposal for an international Code of Conduct for data sharing in genomics. Developing World Bioethics, 1–14. https://doi.org/10.1111/dewb.1238
[18] https://www.iso.org/standard/62289.html

compliance with pertinent international standards and regulatory frameworks, including the EU General Data Protection Regulation (GDPR).[19] For this specific project, a 'PIA+' methodology is being implemented. The PIA process is extended to include ethical and social considerations. At present, the medical field is overloaded with new "disruptive technologies" and "digital transformations", and the requirement to rethink and rephrase medical ethics is pertinent.[20] At times of significant advancements, approaches that rethink and prioritise patients' rights and the medical obligations should be at the core of technological developments.

Medical genetics raises challenging ethical questions for both patients and clinicians, specifically relating to the patient's family or social environment. Addressing these problems requires commitment to transparency as well as attention to the complexities of the findings.[21] For instance, genetic data may present incidental findings and identification of variants not of current significance. Consequently, reports are often preliminary rather than final. In project iToBoS, to support the return of results, pre-test genetic counselling will be provided to willing participants, to advise of the benefits and limitations of genetic testing, as well as the anticipated outcomes of the test for high penetrance family melanoma genes. According to the terms of 'specific' consent, participants will receive their data in an aggregated format, as opposed to raw genetic data, which is largely uninterpretable. Additionally, further consenting procedures will be required if genetic data will be used beyond the current project. It is hoped that the marriage of a responsible research and innovation practice will mitigate some of the overarching concerns expressed by patients regarding genomics use.

Conclusion

The IToBoS project is currently at the 'coal-face' of medical research, intersecting nuanced conversations regarding data privacy, AI, genomics data, and melanoma research. The consortium's commitment to responsible research and innovation requires openness, transparency and accountability. The nature of the project requires consideration of emerging regulations and evolving technological implications, including understanding complex questions of proportionality with regards to genomics data, health service provision, data sharing, as well as broad privacy, ethical and social implications.

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[19] https://gdpr-info.eu/

[20] Sharon, T. (2016). The Googlization of health research: from disruptive innovation to disruptive ethics. *Personalized medicine*, *13*(6), 563-574.

[21] Clarke, A. J., & Wallgren-Pettersson, C. (2019). Ethics in genetic counselling. *Journal of Community Genetics*, *10*(1), 3-33.