



## INTERVENE

Report on the qualitative assessment of the public’s perception of AI-based genetic scores

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## List of Abbreviations

**AI:** Artificial Intelligence

**BBMRI:** Biobanking and Biomolecular Resources Research Infrastructure

**EAB:** Ethics Advisory Board

**ELSI:** Ethical, Legal, and, Societal Issues / Implications

**PRS:** Polygenic Risk Score

**TUM:** Technical University - Munich

**WP:** Work package

## Introduction

Integration of artificial intelligence (AI) and polygenic risk scores (PRS) into healthcare is an important development that has the potential to revolutionize diagnosis, prevention and personalized treatment of health conditions. Considering that both AI and PRS separately and combined necessitate a thorough understanding of emergent risks and challenges, it is essential to critically examine their impact, not only from a technical, medical/epidemiological perspective, but also from a societal lens that takes into account the public’s perception around their use. Thus, this report presents an assessment of public perception informed through the relevant tasks of WP6 and the interactive exchanges with other work packages of the INTERVENE project.

The previous deliverable (D6.3) on PRS and AI from an ELSI perspective, published as an article, laid the grounds regarding the survey of literature and identification of key strands in a forward-looking mode (Fritzsche et al, 2023). The article highlighted that along with opportunities, new complexities and challenges emerge as AI and PRS are brought together. Considering that these relate to issues such as fairness, bias, representation, but also explainability, trustworthiness and use as evidence, the findings inform both the interpretation of the literature on public perception of AI-based genetic scores as well as the ongoing qualitative analysis that forms the core of this deliverable.

The scholarly literature on perception of genetic risk is diverse and fragmented and comes from many different domains of the social sciences and humanities, from psychology to sociology. Neither PRS nor its consideration together with AI has received enough attention due to their novelty and the lack of widespread implementation. Therefore, the scholarly literature on perception of genetic risk has to be interpreted in a new light and the ongoing qualitative work on public perception, once published as an empirical study, will contribute to this literature as a significant piece.

Considering these and the potential impact of AI, along with the qualitative study conducted on the public perception of AI-based genetic scores, we have followed closely the developments in INTERVENE in relevant WPs through an embedded ethics approach that informs and highlights key challenges and risks identified along with a thorough review of the legal landscape regarding genetic reporting tools, polygenic risk scores and return of results in Europe. Our work encompasses an overview of the state of the art regarding public perception of genetic testing and relevant aspects; an examination of challenges identified through our embedded ethics approach and analysis of the legal landscape; and insights from our qualitative study.

## Methods

In line with the aims of WP6, this report on the qualitative assessment of public perception of genetic scores and genetic reporting tool relies on a two-pronged approach: embedded ethics activities to keep track and provide input for ongoing research in INTERVENE including literature reviews and legal analyses, and qualitative interview study to get insights into the public perception of polygenic risk scores and AI use.

## Ongoing Embedded Ethics Activities

Embedded ethics refers to “the ongoing practice of integrating ethics into the entire development process” (McLennan et al., 2020: 3), where ethics becomes a truly collaborative, interdisciplinary endeavor. The main goal of an embedded ethics approach is to develop AI technologies that are ethically and socially responsible, benefiting rather than harming individuals and society. This involves integrating ethical considerations into every stage of development—from planning to implementation—to proactively address potential ethical issues. Additionally, involving ethicists throughout the process can foster innovative scholarship that anticipates social and ethical challenges associated with medical AI technologies. Embedded ethics involves collaboration between ethicists and the development team to continuously address these issues through an iterative process (McLennan et al., 2020; McLennan et al., 2022; Tigard et al., 2023; Tigard et al., 2024; Breuer et al., 2024).

In INTERVENE, the embedded ethics approach was implemented following the gold standard for integration, which includes having scholars of ethics and social sciences as dedicated team members. The efforts of WP6 focused on achieving this. Additionally, researchers from WP6 worked in parallel to theoretically advance the methodology of embedded ethics with other researchers. In this context, a publication with a toolbox was written for further endeavours to embed ethics (Willem et al., 2025). Another manuscript on embedded ethics is currently in preparation and will be submitted soon for peer review.

Some of the embedded ethics activities include the following: 1. Regular meeting with or attendance of regular meetings of researchers from various work packages, especially WP5 and the Flagship meetings; 2. Meetings and communication with Ethics Advisory Board (EAB): WP6 has actively participated in the meetings of the consortium with the EAB and additionally facilitated *ad hoc* meetings when a regulatory issue was identified in WP3’s work; 3. Meeting work packages particularly for T6.2.1., providing input on the WP5 reporting tools and on the IGS4EU platform; 4. Embedded ethics workshops for the whole Consortium to facilitate the integration of ethics in the project, in person as well as virtually; 5. Incorporating legal expertise: Using the existing networks, talking to experts (e.g. regarding legislations); Besides the EAB, asking for legal expertise from BBMRI and reaching out to further legal experts via the existing network of WP6; 6. Ongoing review and analyses of peer-reviewed ethical, legal and scientific literature; 7. Joint interdisciplinary peer-reviewed manuscripts and articles.

Throughout the embedded ethics activities, WP6 has provided guidance, critical perspective and insights to respective WPs/researchers, and overall, these contributed to the development of the qualitative research component of WP6 activities and the pre-analysis for the assessment of the public perception.

## Literature reviews and analyses of the legal landscape

Details of the methods for the additional legal analyses and literature reviewing will be shown in forthcoming publications. Since these are pending they can unfortunately not be provided in this report. However, they are available upon request.



Qualitative Interview Study

Our qualitative study includes in-depth interviews with three interviewee groups to provide a variety of perspectives and in-depth insights into the perception of genetic scores and AI: Patient representatives, researchers as well as clinicians. In addition to the ethics approval of INTERVENE, the researchers from WP6 acquired an ethics approval (2024-395-S-NP) for the qualitative interview study from the research ethics committee of the Technical University of Munich. The interviews were designed as semi-structured qualitative interviews.

Details of the methods will be provided in an upcoming peer-reviewed publication and are available upon request.

Results

The results of the research and activities conducted are clustered in three categories due to their character. They are available upon request.

First, challenges that are relevant for the consortium, especially from a public perception viewpoint focusing on PRS and AI and informed through the **embedded ethics activities and literature review**. These results will be detailed in forthcoming publications.

Then, our results comprise an **overview of regulations** that might be relevant for the return of genetic findings based on the literature and legal landscape with the support of a legal scholar from BBMRI. While legal frameworks do not provide insights into the public perception, they are important to understand the variety of regulations that shape practices across different countries as well as providing insights on what legislative and bureaucratic processes culminate in regarding return of findings.

Finally, we provide findings from the **interview study on** the public perception of genetic tools including PRS and AI, which will be discussed in detail in an upcoming peer-reviewed publication. The semi-structured interviews captured a broad range of insights on PRSs and AI-driven PRSs, including both lay perspectives and cutting-edge expertise perspectives. Study participants included researchers, clinicians, and patient representatives, covering views from recent scientific development of PRSs for multiple diseases and disorders, the development of AI-driven PRSs, and clinical trials with PRSs.

The development of extended dynamic scores using multi-omics data and additional health data such as electronic health records raises ethical questions that can be informed by additional, broader analyses. We have addressed these topics in our collaborative work with other scholars, contributing to research on ethical considerations such as potentials of large-scale data and AI from the perspective of biobanking (Akyüz et al., 2024), digital endpoints in clinical trials (Tackney et al., 2024), and trustworthy use of AI in medicine (Lekadir et al., 2023 preprint, Lekadir et al., accepted).

Our research on public perception of genetic scores, including PRS and the application of AI in genetic scores, revealed a number of ethical, social and legal challenges. These results will be detailed in forthcoming publications. Since these are pending they can unfortunately not be provided in this report. However, they are available upon request.

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Detailed results on the theoretical work for advancing the methodology of embedded ethics with other researchers can be found in a peer reviewed publication with a toolbox designed for further endeavours to embed ethics (Willem et al., 2025).

Conclusions

With this deliverable, we aimed to contribute to the evaluation of the perception of genetic scores-based risk among experts (clinicians and researchers) and patients and the analysis of the ethical aspects surrounding the notion of genetic risk prediction across different European countries. Adopting an embedded ethics approach, WP6 has consistently provided ongoing advice to the consortium whenever possible towards the goals of the project with sensitivity to ethical, legal and societal implications and aspects. Based on our comprehensive literature review, embedded ethics work, legal analyses and qualitative study, we provide conclusions that will inform the next task of the WP, i.e., *T6.3. Framework for trustworthy AI and ethical guidance principles for genetic score-based risk prediction and personalised medicine.*

## References

- Akyüz, K., Cano Abadía, M., Goisauf, M. and Mayrhofer, M.T., 2024. Unlocking the potential of big data and AI in medicine: insights from biobanking. *Frontiers in Medicine*, 11, p.1336588. doi:10.3389/fmed.2024.1336588.
- Breuer, S., Witz, S., Skerlj, J., Braun, M., Bak, M., Naceri, A., & Müller, R. (2024). Putting Embedded Ethics and Social Science into practice: the role of peer-to-peer relationships. *Journal of Responsible Innovation*, 11(1), 1-25. doi:10.1080/23299460.2024.2426854.
- Fritzsche, M. C., Akyüz, K., Cano Abadía, M., McLennan, S., Marttinen, P., Mayrhofer, M. T., Buyx, A. (2023). Ethical layering in AI-driven polygenic risk scores – new complexities, new challenges. *Front. Genet.*14, 1098439. doi:10.3389/fgene.2023.1098439.
- Lekadir, K., et al. (2023). FUTURE-AI: International consensus guideline for trustworthy and deployable artificial intelligence in healthcare. *arXiv preprint arXiv:2309.12325*.
- Lekadir, K., et al. (accepted). FUTURE-AI: International consensus guideline for trustworthy and deployable artificial intelligence in healthcare. *British Medical Journal*.
- McLennan, S., Fiske, A., Celi, L. A., Müller, R., Harder, J., Ritt, K., Haddadin, S., & Buyx, A. (2020). An embedded ethics approach for AI development. *Nature Machine Intelligence*, 2(9), 488-490. doi:10.1038/s42256-020-0214-1.
- McLennan, S., Fiske, A., Tigard, D., Müller, R., Haddadin, S., & Buyx, A. (2022). Embedded ethics: a proposal for integrating ethics into the development of medical AI. *BMC Medical Ethics*, 23(1), 6. doi:10.1186/s12910-022-00746-3.
- Tackney, M.S., Steele, A., Newman, J. et al. Digital endpoints in clinical trials: emerging themes from a multi-stakeholder Knowledge Exchange event. *Trials* 25, 521 (2024). doi:10.1186/s13063-024-08356-7.
- Tigard, D. W., Braun, M., Breuer, A., McLennan, S., & Buyx, A. (2023). Toward best practices in embedded ethics: Suggestions for interdisciplinary technology development. *Robotics and Autonomous Systems*, 167(1), 104467. doi:10.1016/j.robot.2023.104467.
- Tigard, D. W., Braun, M., Breuer, S., Fiske, A., McLennan, S., & Buyx, A. (2024). Embedded Ethics and the "Soft Impacts" of Technology. *Bulletin of Science, Technology & Society*, 0(0). <https://doi.org/10.1177/02704676241298162>.
- Willem, T., Fritzsche, MC., Zimmermann, B.M. et al. Embedded Ethics in Practice: A Toolbox for Integrating the Analysis of Ethical and Social Issues into Healthcare AI Research. *Sci Eng Ethics* 31, 3 (2025). <https://doi.org/10.1007/s11948-024-00523-y>.