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High-throughput genomic data: Challenges for the physician-patient relationship and patient role. Qualitative studies on the handling of genomic information from the perspective of experts, affected persons and lay people.

(Cumulative dissertation)

References of the published articles:

Urban, A., & Schweda, M. (2018). Clinical and personal utility of genomic high-throughput technologies: perspectives of medical professionals and affected persons. *New genetics and society*, 37(2), 153-173. doi:10.1080/14636778.2018.1469976

Urban, A., & Schweda, M. (2019). Zwischen Revisionismus und Normalisierung: Eine Analyse des bioethischen Diskurses um genomische Hochdurchsatztechnologien. In G. Duttge, U. Sax, M. Schweda, & N. Umbach (Eds.), *Next-Generation Medicine* (pp. 77 - 98): Mohr Siebeck.

Urban, A. (2020). "...This Has to Do With My Identity. And I Don't Want to Make it Totally Transparent." Identity Relevance in the Attitudes of Affected People and Laypersons to the Handling of High-Throughput Genomic Data. *Frontiers in Sociology*, 5(532357). doi:10.3389/fsoc.2020.532357

Outline:

1. Introduction and main question
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 - 2.1 Genomic high throughput sequencing - concept and background
 - 2.2 Analysis of the bioethical debate on the handling of genomic high throughput technologies
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Summary

The clinical implementation of high-throughput genomic technologies for sequencing the human genome is progressing. Decreasing costs and increasing performance have greatly increased the attractiveness of these technologies. However, questions about ethical, legal and social challenges remain undiminished with respect to clinical use. There is a lack of empirical insight into subjective perspectives on the use of genomic information.

This cumulative dissertation therefore examines the academic-ethical discourse on the handling of high-throughput genomic technologies in the context of the first publication and, for this purpose, outlines three lines of argumentation that are crucial in the debate on the handling of the technology and its ethical and practical implications (Urban and Schweda 2019). Furthermore, the development of the discourse as well as its structural conditions and characteristics are presented.

Within the second publication, the perspectives on utility in the views of professionally and personally affected persons are elaborated on the basis of the results of partially standardized interviews (Urban and Schweda 2018). They show that clinical utility is used for assessment primarily by professionally affected individuals. In contrast, personal utility, without direct impact on outcome of medical treatment, is mainly mentioned by those personally affected to evaluate the technology and genomic information. In addition, other concepts of utility that go beyond the individual scope of a clinical or personal utility are identified. The results of the second publication illustrate, that those different concepts of utility point to potential discrepancies in the physician-patient relationship, which are important for the implementation of high-throughput genomic technology.

In the third publication, the identity-relevance of genomic information in the perspectives of personally affected persons and medical laypersons is explored based on the results of semi-standardized interviews and focus group discussions (Urban 2020). Based on an interpretative approach, five topics are identified to which identity-relevance can be attributed. Upon this, two basic identity concepts found in the attitudes of the interviewees are presented: First, a closed identity concept that is based on deterministic views and an exceptionalist status, regarding the informative value of genomic information. Second, an open concept of identity emphasizing relativity and a normalizing view of genomic information. In the result of the third publication, it is explained that genomic information can be attributed a temporally and contextually dependent identity-relevance. Due to exceptionalist and deterministic attributions regarding the informative value of genomic information, challenges arise for the patient role and the self-image of personally affected individuals.

Overall, the results show that the complexity of genomic information, which is emphasized in the academic-ethical and socio-scientific discourse, has no or only a minor relevance in the perspectives of those personally affected and medical laypersons, and that exceptionalist and deterministic views are used to a major extent.