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RESEARCH ARTICLE

Omics and AI in precision medicine: Maintaining socio-technical imaginaries by transforming technological assemblages

Robert Meunier*, [1], Christian Herzog[2]

Abstract • We understand precision medicine as a socio-technical imaginary according to Jasanoff. After briefly outlining how the imaginary of precision medicine emerged from the Human Genome Project and spread across national contexts, we raise the question of why the imaginary and the expectations and promises associated with it persist despite regular disappointment among practitioners about the failure of personalized healthcare. We argue that short-term technological hypes enable stakeholders to renew and maintain the promises of precision medicine. In our view, these hypes are around transformations of a technological assemblage. We discuss this in detail for omics and AI technologies and evaluate the recent transformations in light of the long-term imaginary of precision medicine.

***Omics und KI in der Präzisionsmedizin**: Die Aufrechterhaltung soziotechnologischer Imaginarien durch die Transformation technologischer Assemblagen*

Zusammenfassung • *Wir verstehen Präzisionsmedizin als soziotechnologisches Imaginarium nach Jasanoff. Nach einer kurzen Darstellung, wie die Imagination der Präzisionsmedizin im Zuge des Humangenomprojekts entstanden ist und sich über nationale Kontexte hinweg verbreitet hat, werfen wir die Frage auf, warum die Imagination und die damit verbundenen Erwartungen und Versprechen trotz regelmäßiger Enttäuschung über das Scheitern der personalisierten Gesundheitsversorgung in der Praxis fortbestehen. Wir argumentieren, dass kurzfristige technologische Hypes es den Interessenvertretern ermöglichen, die Versprechen der Präzisionsmedizin immer wieder zu erneuern und damit die Imagination aufrechtzuerhalten. Bei diesen Hypes geht es nach unserer Auffassung um Transformationen einer technologischen Assemblage. Wir erörtern dies ausführlich für Omics- und KI-Technologien und bewerten die jüngsten Transformationen im Lichte der langfristigen Imagination der Präzisionsmedizin.*

Keywords • *precision medicine, big data, artificial intelligence, imaginary, assemblage*

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Introduction

The term 'precision medicine' appeared around 2010 and gained prominence since 2015, when the Obama administration launched the Precision Medicine Initiative (Blasimme and Vayena 2016). Here is the PMI's mission statement:

"The future of precision medicine will enable health care providers to tailor treatment and prevention strategies to people's unique characteristics, including their genome sequence, microbiome composition, health history, lifestyle, and diet. To get there, we need to incorporate many different types of data, from metabolomics […], the microbiome […], and data about the patient collected by health care providers and the patients themselves." (White House, quoted from Etchings 2017, p. 17)

As the quote indicates, the goals of personalized treatment and prevention are to be achieved by collecting and integrating large amounts of heterogeneous data. Such data and the means for acquisition, management, and analysis are often referred to as '-omics' in reference to the term 'genomics', used for the information technology-aided study of DNA sequence developed in the course of the Human Genome Project (HGP). The

HGP resulted in much of the theoretical, regulatory, and material frameworks on which today's biomedical big data research is built (Hilgartner 2017). Currently, not only molecular, but also data on environmental exposure, lifestyle, or individual medical history is seen as part of the omics endeavor, despite being more heterogenous than, e.g., genomic or proteomic data. Information infrastructures, from databases to clouds, are necessary for data-intensive approaches, but in particular data science methods from statistical algorithms to artificial intelligence (AI) applications, esp. machine learning (ML), are seen as crucial in generating knowledge from data of such diverse origins (Etchings 2017; Heil et al. 2021; MacEachern and Forkert 2021).

Precision medicine can be regarded as part of a continued effort by various stakeholders from science, industry, healthcare, and politics to maintain a vision of data-intensive, molecular biology-centered biomedicine that emerged in the wake of the HGP. The more commonly used term for this vision is 'personalized medicine'. An early opinion piece on this new 'post-genomic' vision from 2001 stated:

"The post-genome era has begun, and with it the promise of tailoring the practice of medicine to the individual. This emerging field of personalized medicine encompasses the use of risk algorithms, molecular diagnostics, targeted therapies and pharmacogenomics to improve health care. Personalized medicine will provide the link between an individual's molecular and clinical profiles, allowing physicians to make the right patient-care decisions and allowing patients the opportunity to make informed and directed lifestyle decisions for their future well-being. Molecular diagnostics, the use of DNA-, protein- or mRNA-based biological markers to predict the risk of developing disease or the molecular phenotype of an existing one, will change the way we currently define disease" (Ginsburg and McCarthy 2001, p. 491).

From the metaphor of 'tailoring' to the notion of individual characteristics, the emphasis on both prevention and treatment, and the idea that data from various molecular levels will be integrated with clinical records, there is much continuity between this quotation and the above statement on precision medicine. Indeed, the US National Research Council report (NRC 2011) that introduced 'precision medicine' as a science policy category, suggested it as a replacement for 'personalized medicine' and many treat the terms as synonymous. We will use 'PM' to refer to both.

The question is: How is this post-genomic biomedical vision – that we will analyze as a socio-technical imaginary – maintained, even though its promises remained unfulfilled (Section 1)? Our answer is that this is achieved by stakeholders through connecting the imaginary to ever-new technologies – a process we address as transforming a technological assemblage – and by depicting these transformations as revolutionary, i.e., through technological hype (Section 2). By reviewing the biomedical literature and drawing on relevant STS work, we show how short-term hypes (applying to transformed assemblages) enable a renewal of a long-term promise (pertaining to an imaginary). For the case at hand, this means that the current omics and AI technologies need to be assessed as elements of an assemblage and in the context of the assemblage's role in the long-term imaginary (Conclusion).

How is the post-genomic biomedical vision – that we will analyze as a socio-technical imaginary – maintained, even though its promises remained unfullfilled?

Maintaining the post-genomic biomedical imaginary

While practitioners often speak of a paradigm (shift), commentators have characterized PM as a scientific/intellectual social movement (Juengst et al. 2016; Au 2021), umbrella term (Pokorska-Bocci et al. 2014), or socio-technical imaginary (Erikainen and Chan 2019; Strand 2022), among others. All these denominations bear value, depending on the question asked. We adopt the notion of a socio-technical imaginary. This is appropriate, because of the longitudinal outlook, where imaginaries are described as relatively stable or durable, albeit not as static, but as undergoing dynamic changes. This serves our interest in long-term trends. Even though PM has itself been described as a hype (Maughan 2017), we reserve 'hype' for the more short-term dynamics pertaining to technological novelty.

Socio-technical imaginaries are defined as "collectively held, institutionally stabilized, and publicly performed visions of desirable futures, animated by shared understandings of forms of social life and social order attainable through, and supportive of, advances in science and technology" (Jasanoff 2015, p. 4). The concept thus usefully ties together politics, meaning, morality, and technology in the context of promises and expectations that envision socio-technical futures, but are shaped by current interests and in turn have effects in current socio-economic systems.

Regarding politics, socio-technical imaginaries "are not the same as policy agendas" and yet they involve "active exercises of state power, such as the selection of development priorities, the allocation of funds, the investment in material infrastructures, and the acceptance or suppression of political dissent" (Jasanoff and Kim 2009, p. 123). We cannot discuss this in detail but refer to Blasimme and Vayena (2016) for the case of the U.S. Importantly, while imaginaries were originally seen as playing out on the national level, Jasanoff's 2015 account suggests that imaginaries can be effective across nations when propagated by economic or professional networks.

Indeed, the HGP as well as the development of the imaginary of PM are strongly connected to U.S. science and healthcare policies. However, PM has been adopted by research institutions and policies in developed countries, e.g., in Europe and Japan (Bando 2017). While facing similar challenges for healthcare systems, such as an aging society and the rise of chronic diseases, each of these contexts is characterized by specific political and regulatory environments, as well as varying healthcare systems. Also developing countries such as Brazil and China joined the 'bandwagon' (Au and Da Silva 2021).

Analyzing PM as scientific/intellectual movement (SIM) through collaborations as documented in articles published between the 1990s and 2019, Larry Au generates network graphs, which are centered on the U.S. and U.S. institutions, but indicate a global distribution: "These network diagrams can be interpreted as the uptake of a SIM's language by researchers across countries or institutions, and crucially, a working agreement as to what the SIM is – at least enough to facilitate collaboration." (Au 2021, p. 6) The propagation of the imaginary through these networks is then accompanied by continued negotiation of the meanings of the respective labels (e.g., 'personalized' vs. 'precision'), the locus of the novelty compared to previous medical practice or health care models, and normative consequences (e.g., regarding the position of the patient) (Juengst et al. 2016; Erikainen and Chan (2019). We focus here on the technological dynamics accompanying the development of the imaginary.

Before we address the hype around omics and AI (Section 2), we need to highlight a historical conundrum to which it speaks. So far, we have made clear how the socio-technical imaginary of PM emerged from the HGP in the late 1990s and was established first in the U.S., then in the EU and in many other places. This, however, does not explain why the imaginary has persisted for such a long time. One might think that this is because it brought about or at least partly realized the desired social progress it envisions. And indeed, proponents constantly repeat success stories. However, these success stories are few compared to the candidate targeted drugs and molecular markers pursued in research. Proponents, as well as critical commentators, have thus repeatedly observed that PM "falls a long way short of the predictive and preventative healthcare paradigm it once promised", as an editorial in *Nature Biotechnology* put it about ten years after the conclusion of the HGP (Anonymous 2012). Tim Maughan, a research active physician, after discussing some of the success stories for cancer treatment, cites a study reporting "that the mean improvement in overall survival from 71 targeted cancer therapies approved by the FDA between 2002 and 2014 was only 2.1 months" (Maughan 2017, p. 14). He then diagnoses that the "main scientific reason that has emerged for this relative failure of targeted therapies for cancer is the presence of profound tumour heterogeneity and clonal evolution that can be identified in most cancers" (Maughan 2017, p. 14). Twenty years after the HGP, researchers still observe that "[d]espite personalized medicine […] being featured prominently in industry and academia, its promise has largely not been realized" (Lamb et al. 2021, p. 20). However, referring to the blood serum proteome as a source for biomarkers, they maintain that "[t]echnological advances for high-throughput measurement of proteins in biological samples have facilitated this work" (Lamb et al. 2021, p. 21). This is a common pattern. Most practitioners see no obstacle to PM's success, in principle. They point out the failure, only to suggest a technological feat that has been missing to reach the goals. Hence, technology plays a crucial role in maintaining the imaginary despite disappointments. We substantiate this view with respect to omics and AI in the context of PM.

Transforming the assemblage: the omics and AI hype

It is a truism for technology assessment as well as for social studies of technology that technologies are not isolated. To understand how omics technologies and AI are subject to hype and to contextualize this hype in the long-term development of the post-genomic biomedical imaginary discussed above, we need to think of them as integrated in an interdependent manner into a socio-technological nexus, which we conceptualize as an assemblage.

Technology plays a crucial role in maintaining the precision medicine imaginary despite disappointments.

The word *assemblage* is an unfortunate translation of the French term *agencement*, originally used by Deleuze and Guattari (1980). Rather than referring to something larger that is assembled to produce a new and emerging unit, an assemblage is to be conceived of as an "arrangement or layout of heterogeneous elements" (Nail 2017, p. 22). Assemblages are not systems in the narrow sense of constellations whose characteristics emerge from the properties of their components. The description as an assemblage serves to maintain diversity – instead of essence – and emphasizes relations and events between its constituents. Hence, an assemblage allows for the recombination

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or removal of its "self-subsisting fragments" (Nail 2017, p. 23) without its (immediate or even eventual) dissolution. Hence the notion is inherently about dynamics and contingencies, rather than stability and unification.

Assemblages, like imaginaries, are dynamic but persistent. There is no cause-and-effect relation between the two. The imaginary involves broader socio-political processes which support the formation of a scientific-technological assemblage that is seen to enable an envisioned social order. But the assemblage and its anticipated capabilities in turn shape this vision. In line with the notion of an imaginary, "desire" gives things the properties they have in an assemblage (Buchanan 2020, p. 56). But this also connects it to short-term technological hypes as organizing collective expectations, which can be understood as "real time representations of future technological situations and capabilities" (Borup et al. 2006, p. 286). We locate hype in the events of transformation of an assemblage that are perceived or presented as disruptive. Unlike systems-of-things, assemblages are not defined by their components, but rather by their products (Buchanan 2020, p. 47). Even with material elements removed, an assemblage can still result in *actual* effects, e.g., in the form of stories, expectations and hopes that matter to people. Transformations of assemblages through removal or addition of elements, can, hence, perpetuate the promises of an imaginary, even if the specific promises associated with specific technologies fail to be actualized.

The PM imaginary supports an assemblage that involves heterogeneous sets of apparatuses, infrastructure, institutions, and social actors associated with academic, clinical, and industry research. This assemblage underwent several transformations. Many can be described as alterations of existing technologies, integration of technologies from different fields, or the introduction of novel technologies. Some of these events are described by stakeholders as revolutionary.

‚Omics' already refers to a very heterogeneous set of technologies. In the course of the HGP, DNA sequencing technology evolved quickly and with it bioinformatics tools and infrastructures. These formed the core assemblage of the PM imaginary, which was initially conceived primarily as 'genomic medicine'. Around 2005, next-generation sequencing technologies (NGS) became integrated into the assemblage. This was seen as revolutionary in many fields, as it made access to sequence faster and cheaper (Bösl and Samida 2021). It not only made it possible to collect more genomic data across populations or for individual tumors, but also to study more extensively RNA transcripts (transcriptomics) or the human microbiome (metagenomics). At the same time, various technologies to study proteins and metabolites were further developed for high-throughput data generation (proteomics, metabolomics). 'Systems medicine' was suggested as way to deliver PM by providing a framework for integrating molecular omics data. It promised to be more holistic and dynamic than the gene-centered reductionism and determinism of the HGP. The new data generation technologies and computational tools expanded the assemblage and, again, this was described as an 'omics revolution' (MacEachern and Forkert 2021).

The shift to the 'precision medicine' label is sometimes associated with the inclusion of yet more data sources made available through digital devices (e.g., wearable sensors) and digitalization of many processes, including patient records and insurance claims (Etchings 2017; Fröhlich et al. 2018). This 'extended omics', includes new data types, e.g., on exposure, lifestyle, or medical history. Extended omics come with a 'data deluge' that is seen as threatening the success of these technologies, similar to the earlier 'bioinformatics bottleneck' diagnosed in the context of NGS. In this situation, AI technologies such as ML are suggested as solutions as they are expected to meaningfully integrate these heterogeneous data sets (Fröhlich et al. 2018; Heil et al. 2021; MacEachern and Forkert 2021). This merging of omics and AI represents a recent transformative event. Indeed, hype has been diagnosed for biomedical big data and AI (Fröhlich et al. 2018). At the same time, this event aligns the PM imaginary with other big data imaginaries. In fact, none of these technologies are exclusive to biomedicine. They are used in other fields in biomedicine, biology, and elsewhere. But each lead to new infrastructures, institutions, social roles, and research designs in the context of the PM imaginary.

The big data approach fostered by extended omics and AI technologies is seen to overcome the limitations of an earlier genome-centric personalized medicine.

The term 'artificial intelligence' does not reference any single or specific mathematical method, computer hardware, or way of deployment. Rather, AI in itself constitutes an assemblage of all elements able to achieve an output behavior reminiscent of intelligent human behavior (cf. Wang 2019). AI technologies have even broader applications than omics and are currently discussed in terms of hype with respect to many fields. Both research and popular literature are rife with statements signaling "overpromised and underdelivered" results of AI in health care (Strickland 2019, p. 24) and remaining "major technical and ethical questions" (Rajpurkar et al. 2022, p. 36). Yet the belief in the potential of AI remains firm, even while proponents openly call for "a need for bold imagination" (Rajpurkar et al., 2022,

p. 36). As Schneider and Lösch (2019, p. 205) put it with reference to assemblages, such imagination "would not be conceived as something of the isolated mind or person but an emergent effect of the relations and co-operations of elements".

To be sure, in its current shape, the assemblage associated with the PM imaginary involves more than extended omics and AI. Older technological elements are still included. Omics still includes sequencing and AI technologies are built on long-term bioinformatic infrastructures. Furthermore, there are other elements, some constituting recent novelties, that we do not discuss here, such as advanced imaging, organoids, or single-cell omics, each perceived as disruptive in an ever-accelerating cycle of technological transformations. The key insight is that hyping new technologies and, hence, transforming an assemblage is related to the technologies' ability to speak to and re-new the promises that have been put forward with respect to earlier configurations of the assemblage in the context of an imaginary. Paradoxically, the fact that previous technologies integrated into the evolving assemblage did not deliver what was expected does not create distrust in the new ones but makes them stronger, as they can be presented as overcoming the shortcomings of the older ones. The big data approach fostered by extended omics and AI technologies is seen to overcome the limitations of an earlier genome-centric PM.

Conclusion

This research article clarified the relation between long-term socio-technical imaginaries, entailing promises and expectations, and short-term technological hypes, which we have described as applying to technologies as integrated into and transforming an assemblage associated with an imaginary. The PM imaginary emerged in the wake of the HGP and has persisted for a quarter of a century, carrying forward the same set of promises of improving healthcare based on individual characteristics of patients and healthy citizens. Despite some success stories, the promises have not been fulfilled to an extent that would justify the amount of support mobilized by the imaginary. What maintained the imaginary was the continued transformation of the assemblage of research technologies. Most recently, the perceived revolutionary event of introducing and merging extended omics and AI technologies enabled stakeholders to maintain the PM imaginary.

How then should we assess these technologies, resisting the hype? It is certainly not warranted to infer inductively that because previously hyped technologies did not deliver on the promises of PM, extended omics combined with AI will not deliver either. However, the long-term developments suggest a more cautious approach regarding the expectations. This is relevant regarding the normative aspects of the PM imaginary. The emphasis of omics and AI approaches on the use of more data on more levels of a citizen's life, from life-style information gathered by mobile devices to medical records, for research and clinical practice, brings with it a number of ethically relevant issues: increased medicalization of healthy individuals (Erikainen and Chan 2019), the moral duty of data donation (Lee 2021), or concerns about data security or discrimination resulting from undetected biases (Geneviève et al. 2020). These issues can be represented as 'costs' on the side of health care receivers and need to be related to expected benefits. Hence it is important to reflect on how realistic these expectations are.

Nonetheless, previous hypes maintaining the imaginary also teach us that transformations of the assemblage had many effects on how research is done, even if those were not always the expected ones. Likewise, the current big data regime will certainly result in innovations in research and industry, new infrastructures, institutions, and actors (Heil et al. 2021). Big data approaches might even eventually lead to personalized treatment and prevention. But then, one can also assess the PM imaginary as a whole and the social order it envisions. Many have criticized the way it unevenly supports economic aims and diminishes efforts to improve public health measures (most recently, Tabery 2023).

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