Introducing Kuhn et al.’s Paper “Informatics and Medicine: From Molecules to Populations” and Invited Papers on this Special Topic

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Summary

Objective: To introduce the paper by Kuhn et al. “Informatics and Medicine: From Molecules to Populations” and the papers that follow on this special topic in this issue of Methods of Information in Medicine, which opens a debate on the Kuhn et al. paper’s assertions by an international panel of invited researchers in biomedical informatics.

Method: An introductory summary and comparative review of the Kuhn et al. paper and the debate papers, with some personal observations.

Results: The Kuhn et al. paper makes a strong case for interdisciplinary education in biomedical informatics across institutions at the graduate level, which could be strengthened by analysis of previous relevant interdisciplinary experiences elsewhere, and the challenges they have faced, which point to more pervasive and earlier-stage needs for both education and practice bridging the research and healthcare communities.

Conclusions: The experts debating the Kuhn et al. paper strongly and broadly support the key recommendation of developing graduate education in biomedical informatics in a more comprehensive way, yet at the same time make some incisive comments about the limitations of the “positivistic” and excessively technological orientation of the paper, which could benefit from greater attention to the narrative and care-giving aspects of health practice, with more emphasis on its human and social aspects.

Keywords
Informatics, medicine, interdisciplinary education, molecules to populations, genotype to phenotype, clinome, narrative medicine, evidence-based medicine

Parallel rapid developments in both the science and technologies of biology and informatics are already strongly affecting the practice of medicine. They hold the promise of eventually revolutionizing our knowledge of how genetics, development, disease, and environment interact in affecting human health, and strongly impacting medical and healthcare practices. Recently, Professor Klaus Kuhn and colleagues at Munich Technical University, the Ludwig-Maximilians University and the Munich Helmholtz Center proposed the creation of “a research-oriented interdisciplinary Graduate School” [1] which has now been established. This issue of Methods publishes a paper by Kuhn et al. [1] laying out the rationale for such a Graduate School, arguing how informatics and medicine interact uniquely in the study of health and disease across a range of biological scales from molecules to populations, in a way that requires a much stronger interdisciplinary focus than current individual programs to prepare future leaders in the study of biomedical systems affecting all aspects of biomedical research, healthcare practice, and education. Because the importance of this initiative far transcends its immediate local and national contexts in Munich and Germany, the editor of Methods solicited comments on the paper from an international group of experts in biomedical and health informatics and related disciplines, with the goal of stimulating a lively debate on the challenges facing our field. I believe that our readers will agree they have succeeded in this. The paper with these commentaries [2] and one by the past editor of Methods and founder of the IMIA Yearbook [3] which follow that of Kuhn et al. include a wide range of reasoned arguments and original position statements which, while
strongly endorsing the educational needs identified by Kuhn and his colleagues, also point out fundamental challenges that are very specific to the unusual combination of scientific, technological, personal and social problems characterizing biomedical informatics. Most importantly, these point to the ultimate objectives of managing difficult human health problems which are unlikely to yield to technological solutions alone, however effective these may prove for solving some of the better understood and constrained ones. The psychological, societal, and environmental components of health and disease are emphasized by several of the commentators, setting the stage for further debate and constructive suggestions. I appreciate the opportunity to contribute this Guest Editorial emphasizing some of the highlights of Kuhn et al.’s paper with those of the expert commentators in the light of some of my own observations and experiences [4].

What are the major points made by Kuhn et al. to support their thesis that interdisciplinarity is more needed than ever to make informatics effective in medicine? And, does the case for biomedical informatics go beyond what is needed for other similar interdisciplinary programs? The analytical foundation of the paper rests on a four-way breakdown of how informatics has an impact on: 1) basic (systems) biology; 2) biomedical engineering; 3) eHealth; and 4) public health, which is illustrated in figure 1 of [1]. This breakdown gives structure to the arguments of how informatics and its mathematical and computational underpinnings will increasingly help support the development of molecular medicine, clinical translational research, clinical medicine, seamless healthcare and public health (figure 2 in [1]). In defining “the overall picture” early in the paper, the authors identify eight major technical trends in computing and software technology which are likely to increase the ability of systems to handle the massive amounts of complex, structured information involved. These are then divided into groups in terms of their likely impact on advancing: 1) bioinformatics methods for research and increasing the reliability of remote telemedicine applications of eHealth, 2) clinical decision support for integrating the interpretation of complex and heterogeneous data; 3) improvements in the opportunities for personalized prevention strategies; 4) effectiveness of the design of new medical devices; and 5) interface to the wealth of structured knowledge in the literature through the semantic web. The authors then argue for “structured interdisciplinary education” in four areas: bioinformatics and systems biology; informatics for biomedical engineering; health informatics and eHealth, and public health informatics and public health. The core of this paper details those aspects of each that the authors consider most valuable for such education, concluding with an observation that “traditional university systems tend to strengthen a ‘cultural gap’ between classical natural science (e.g. chemistry, biology, physics) and engineering and informatics” ([1], p 11). These in turn are considered to lead to discontinuities between “discovery, invention and innovation” and “widespread technology adoption”, which could be overcome by “changing the patterns of educational activity at the MS and PhD levels where teaching and research meet”. Implementation suggestions are to: 1) coordinate and combine courses across the disciplines; 2) encourage joint research projects and workshops to share experience; 3) foster links at the institutional and personal levels; and 4) encourage project-oriented teamwork spanning the differing research cultures at a high scientific level. The paper concludes by saying that doing this at early graduate school levels with “experts of one scientific discipline who have a profound understanding of the other disciplines’ terminology and scientific culture” is a major need for healthcare systems. The case for breadth and heterogeneity of informatics problems and solutions across the spectrum from “molecules to populations” is certainly convincing. However, what is not addressed are whether there might be lessons to be learned from experiences in other interdisciplinary fields where informatics is critical – in ecological management, earth and environmental sciences where satellite images, GPS and advanced sensor arrays are affecting all sorts of natural, civil and military problems, or the political and economic sciences, where the web of online information has likewise changed the practices of political, legal, commercial and financial competition, for instance.

The commentaries in [2] touch on many difficult problems faced by biomedical informatics as it attempts to span the incredibly wide range of differing needs from basic biological research, through individual healthcare and consumer perspectives in a networked society, to understanding public health implications for various populations at risk. There is a pattern, however, to both the praise and criticisms made by the experts. No one disagrees that more interdisciplinary education is needed and that the graduate level is the realistic one at which to do this, once students have acquired enough background in two or more contributing disciplines. The commentators on the whole take a positive view of the Kuhn et al. paper, suggesting ways of amplifying its content or strengthening its arguments. What proves more controversial is the emphasis on technological solutions to research and healthcare problems argued in [1]. This tends to downplay the personal and social aspects of biomedical informatics which often are major barriers to improving healthcare, and several commentators emphasize this as a limitation. I will next highlight some of the major supporting and critical points made by each commentator, and conclude with a summary. Russ Altman of Stanford University adds to Kuhn et al.’s analysis an emphasis on the “empowered, independent and thoroughly networked (health) consumer” [2], which has transformed the world of communication and social interaction so completely in the past decade. He argues that in the future this trend will have a strong effect on people increasingly making decisions on handling their own healthcare – which should dramatically change the concept and practice of medical and health education for practitioners and health consumers alike – and provide ample research opportunities for biomedical informatics and allied fields. Rudi Balling from the Braunschweig Helmholtz Center for Infection Control raises the vexing questions of the contrast between the “hard” vs. “soft” sciences in the cultures that make up biomedical informatics, and the difficulties of trying to overcome cur-
dent disciplinary prejudices at the graduate level – he argues for the need to do this much earlier in the educational process, while recognizing the political and societal trends that work against it. His plea to “merge the coffee rooms” for students and faculty suggests that increasing understanding and respect among colleagues might be as necessary for finding constructive solutions within competitive environments as its more typical opposite.

James Brinkley from the University of Washington compares and contrasts the Munich proposal to some of the experiences at his university, noting that differences in terminology and practice in informatics between the US and Europe gives the latter a broader scope which could help unify practices across a broad range of biomedical applications. In addition, he points out that figure 2 should more generally show a full set of interconnections between core informatics competencies and the different levels of biomedical application, rather than the strictly horizontal ones, which are too restrictive.

Enrico Coeira of the University of New South Wales points to antecedents for the multilevel approach to informatics and medicine and then focuses on the barriers to adoption of IT in healthcare, which he posits are mainly of a social and not technological nature. He argues for the need to develop an understanding of the unexpected interactions between humans and technological artefacts which can help bridge the different social systems of bioscience, clinical medicine and the citizen. Fabrizio Consorti of La Sapienza contrasts the technological and evidence-based medicine (EMB) perspective of the Kuhn et al. paper to the need for a narrative-based medicine (NBM) approach which is more typical of the practicing clinician’s way of describing a patient’s condition within the clinical context. He argues that considering EMB and NBM as “two different paired dimensions of medical knowledge”[2] will reduce the boundaries between knowledge management and e-learning.

Ali Dhansay from South Africa emphasizes the challenges for biomedical informatics in developing and “hybrid” countries like his own where interracial differences and endemic HIV/AIDS, malaria and tuberculosis have a major daily impact on the lives of people. He brings up a number of economic, ethical and practical issues that arise when one tries to make the scientific achievements in genomics and biomedicine more effective and accessible through informatics, while, however, creating new challenges in terms of anonymity and confidentiality. Antoine Geissbühler of Geneva University and Hospitals argues that the “info-bio convergence” implicit in Kuhn et al.’s proposal is challenged by the need for semantic integration across very different domains, which will change the current emphasis on hypothesis-driven research towards more data-driven research in the future. This certainly will require more education and new models to encourage cross-disciplinary skills for the healthcare workforce under exceptionally difficult and complex social conditions. William Hersh of Oregon Health and Science University suggests that informatics has to go beyond computer science and embrace the human dimensions of information processing, and the need to identify the core competencies of a field which will continue to lead to a wide variety of very distinct career pathways. Yunkap Kwankam of WHO and the University of Yaounde raises the important issue of assessment of health informatics, which is underplayed in the Kuhn et al. paper. After pointing out the known financial benefits from programs that improve drinking water and sanitation worldwide, he argues that health informatics must demonstrate comparable or even greater benefits in order to obtain more adequate funding. He emphasizes the need to identify “communities of practice” that are effective in developing a “collective wisdom” that can help WHO and other organizations to include biomedical informatics in its “value chain”[2], Nancy Lorenzi, from Vanderbilt University, and immediate Past President of IMIA, and developer of IMIA’s Strategic Plan Towards IMIA 2015, comments that Kuhn at al.’s article supports the view that informatics is “the most important driver and mediator for innovation”[2] and that collaboration is essential to achieve this. Knowledge is seen as the core of innovation, which feeds into the major components of IMIA’s Strategic Plan that are designed to integrate and connect the science with the practice of informatics in biomedicine and healthcare. Publications and electronic dissemination of information are identified as the major disseminators of knowledge, and the international interdisciplinary collaborations fostered by IMIA are seen as central to achieving the future goals for informatics and medicine as envisioned by Kuhn et al.’s paper. Fernando Martin-Sanchez from the Ministry of Science and Innovation in Madrid makes the point that nanomedicine and regenerative medicine present both opportunities and challenges for biomedical informatics. With a rapidly ageing population, many European (and other, mostly developed) countries are faced with urgent needs for management and prevention of chronic conditions in disproportionately large percentages of their populations. This will require imaginative and novel approaches to prevention and rehabilitation, including the incorporation of the latest developments in nanomaterials and artifacts for effective deployment. He lists EU initiatives, such as the ACTION-Grid which is to develop healthcare information systems based on the Grid capabilities and new technologies that support these novel directions. George Mihalas of Victor Babes University in Timisoara points to the difficulties that were faced by neuroinformatics in having an impact, and the need to connect to systems biology with projects like the Virtual Physiological Human and the Physiome Project – all of which will require educational components like AMIA’s 10×10 to educate the workforce in informatics. Yuval Shahar of Ben Gurion University in Beer Sheva brings out a very essential point in Kuhn et al’s paper: “only through experts in one discipline who profoundly understand the conceptual world of another discipline can true multi-disciplinary research develop” – he argues for “boundary breaking agents”[2] as role models for students to meet during their training in order to foster true and productive interdisciplinarity. Using the specific example of developing a “smart medical home” Shahar says that only by combining the right bioengineering and medical informatics will this be achieved. He argues for tackling a number of grand challenges,
including what he terms the “Human Clini-
nome Project”[2] as a continuously chang-
ing library of declarative and procedural
clinical knowledge that can be “represented
and accessed by computational means”. He
points to preliminary successes in clinical
guideline structuring and representation as
precursors to this work. Two other chal-
lenge(s Shahar identifies are the manage-
ment of patients, especially those with
chronic disease, and with emphasis on the
temporal aspects of their conditions and the
need to define “clinarrays” of laboratory
data (in analogy to microarrays) to suggest
new ways for subtyping diseases. In addi-
tion, the need for decision support including
genetic components is seen as a fertile area
for engaging patients to take more responsi-
ibility for their own decisions while tying
this to clinical workflows. Katsuhiko Tak-
abayashi, from Chiba University emphasizes
the role of clinical studies and appropriate
security and confidentiality needed to safe-
guard the individual while yielding the
knowledge that underlies how medical deci-
sions are complemented by genetic data.
But he warns that law and ethics are also im-
portant areas of study for biomedical in-
formatics given their strong impact on
people and how they handle health informa-
tion. Finally, Gio Wiederhold from Stanford
University gives a more detailed description
of the interrelated roles of biomedical in-
formatics, engineering, and software engin-
eering (SWE). He recognizes the bureau-
cratic barriers to change, but indicates that
practical problems of security, confidentiality,
and limitations of the state-of-the-art in
SWE, and economic considerations con-
strain the development of large and effective
eHealth systems. He points out that the ratio
of 25:7:1 between research projects, their
practical implementation, and integration
into practice has remained unchanged de-
spite new initiatives such as Service-
Oriented Architectures. His commentary
touches on the social expectations which
limit collaboration between the formal
mathematically based disciplines and those
of the biological world, where variability
and ongoing experimentation is the norm,
and concludes with an interesting comment
on the nature of randomness in limiting
what we can know about our own reasoning
processes—suggesting the need for more re-
search in this direction to help improve
clinician-patient interactions.

The separate paper by Jan van Bemmels
[3] provides an excellent summary of Kuhn
et al.’s paper and draws parallels and con-
trasts to the experience at Erasmus Univer-
sity Medical Center in Rotterdam, and its
pioneering development of a framework for
interdisciplinary research in medical in-
formatics. He quotes from Gilles Holst’s
“Ten Commandments” for encouraging
productive research in a long-standing,
highly successful industrial research lab
(Philips Physics Research Lab in Eind-
hoven). These include the need for engaging
competent scientists when young, without
too much concern about their prior experi-
ence, giving them a lot of freedom, but ex-
pecting them to work hard and share their
work through publications and discussions.
Solving problems through multidisciplinary
teams and encouraging the free movement
from research to development, while being
guided by academic insights as much as by
market opportunities are also essential for
success. Van Bemmels identifies a number of
grand challenges which he ties to the adher-
ence to these “commandments” and illus-
trates how they might be solved if there was
more attention to aspects of human behavior
that define needs not easily or necessarily
satisfied by technological solutions alone.
The cultural aspects of “overstretched ex-
pectations” is a concern raised to caution the
readers about the need to realistically assess
proposals such as the one in Kuhn et al.’s
paper.

In summary, the paper “Informatics and
Medicine: From Molecules to Populations”
[1] has opened a timely debate through the
Commentary papers [2, 3] on the nature of
biomedical informatics and the challenges it
faces, especially in education, in this era of
translational and personalized medicine.
They highlight the urgent need to improve
our understanding of how structuring and
managing information is central to under-
standing the roles of genomics and other
-omics methods in contributing to the pro-
motion of individual and group health as we
attempt to map the complex pathways from
genotype to phenotype [5]. Further con-
siderations extending the scope of these

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