The New Role of Biomedical Informatics in the Age of Digital Medicine

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Summary
Objectives: To reflect on the recent rise of Digital Medicine, as well as to analyse main research opportunities in this area. Through the use of several examples, this article aims to highlight the new role that Biomedical Informatics (BMI) can play to facilitate progress in research fields such as participatory and precision medicine. This paper also examines the potential impact and associated risks for BMI due to the development of digital medicine and other recent trends. Lastly, possible strategies to place BMI in a better position to face these challenges are suggested.

Methods: The core content of this article is based on a recent invited keynote lecture delivered by one of the authors (Martin-Sanchez) at the Medical Informatics Europe conference (MIE 2015) held in Madrid in May 2015. Both authors (Lopez-Campos and Martin-Sanchez) have collaborated during the last four years in projects such as the ones described in section 3 and have also worked in reviewing relevant articles and initiatives to prepare this talk.

Results and Conclusions: Challenges for BMI posed by the rise of technologically driven fields such as Digital Medicine are explored. New opportunities for BMI, in the context of two main avenues for biomedical and clinical research (participatory and precision medicine) are also emphasised. Several examples of current research illustrate that BMI plays a key role in the new area of Digital Medicine. Embracing these opportunities will allow academic groups in BMI to maintain their leadership, identify new research funding opportunities and design new educational programs to train the next generation of BMI scientists.

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1. Introduction

The biomedical and clinical research community has long been known for its whole-hearted commitment to advancement. Discoveries are made every day that lead us closer and closer to the eradication of disease. However, these developments, alongside the rapid progression of the technological era and medical diagnostics, have led us into interesting times. In the past, the pharmaceutical industry focused primarily on developing drugs and therapies, whilst the medical diagnostic industry concentrated on producing laboratory tests and devices, and the Information Technology industry aimed to offer software and information systems. Now, we are witnessing a convergence of these fields around a shared concept: information in health. The evidence for this is plentiful, with technological giants such as Apple pouring resources into analysing human DNA [1] and with Google developing surgical robots [2]. This company has even begun work on developing nanoparticles in an effort to detect cancer [3]. IBM is also partnering with Apple, Johnson and Johnson and Medtronic in a bid to transform healthcare [4]. Digital medicine, characterised distinctively by the use of digital information for healthcare application, has attracted the interest of some very high-profile parties.

This so-called digital revolution also has one more critical element to it: patients are no longer willing to be patient. The public is anxious and ready for the solutions they are demanding from clinical information systems to be put in place [5]. For instance, one particular patient recently tattooed a QR code onto her chest that, when scanned in case of an emergency, provides direct access to her personal health record [6]. Patients are now, more than ever, empowered and engaged and wish to be more active players in the maintenance of their health [7]. Patients, nowadays, are heavily enabled to do this by the access that they have to the diverse range of affordable devices, sensors, mobile applications, Direct to Consumer (DTC) services, and social networks available to them. Partly due to Biomedical Informatics, they are starting to engage in these technologies, using them for reasons linked directly to their health and well-being.
2. What is Digital Medicine

Digital medicine could be defined as the convergence between the digital revolution and medicine. The digital revolution itself has been prevalent in our society for a significant amount of time, impacting greatly on vital domains such as banking, insurance, leisure and government. However, in the field of medicine, digital technology has remained predominantly at hospital and research centre levels, and is yet to be fully accessible to everyday citizens. When it comes to medicine, hardly anything exists on an online platform that allows us control over our own health information. Nevertheless, the emergence of the concept of digital medicine is now beginning to change this [8].

The term digital medicine is increasingly starting to be used synonymously with ideas such as mobile health, digital health, health IT, or health 2.0. Whilst digital medicine does encompass numerous different concepts, i.e. quantified self, sensors, apps, telehealth, games, electronic health records, interoperability, etc., we are yet to see how Biomedical Informatics is included in the landscape of digital medicine.

The journal Nature Biotechnology has recently initiated a series of commentaries and articles jumping on the bandwagon of digital medicine. In the introductory article, the authors define digital medicine as “technology and products that are undergoing rigorous clinical validation and/or that ultimately will have a direct impact on diagnosing, preventing, monitoring or treating a disease, condition or syndrome” whereas the concept of digital health is defined more broadly encompassing products that might not be FDA approved and target a broad audience from patients to researchers [9]. Interestingly, although they delve deep into the subject, claiming that digital medicine is going to transform biomedical research, clinical practice and the commercial sector, the word ‘informatics’ is not once mentioned. Even as the authors suggest that in the next few years we will see the creation of new PhD programs in digital medicine, it is not even considered that these programs have already been in existence for more than three decades under the realm of Biomedical Informatics.

In our research group at the University of Melbourne, we believe digital medicine to be at the intersection between Biomedical Informatics, Participatory Medicine and Precision Medicine (as illustrated in Figure 1), where we identify the latter two as main areas of application for our Biomedical Informatics research [10]. Participatory medicine includes self-quantification and the use of social media, whereas we consider precision medicine an approach to process, integrate and analyse data originating from the human genome, phenome and exposome.

3. Key Research Areas in Biomedicine

3.1 Precision Medicine

We are now aware that having access to the sequence of the human genome is not enough to prevent, diagnose or cure the most prevalent diseases [11]. As Figure 2
illustrates, it simply cannot be used as an exclusive source of information to analyse human disease. Although an individual's genome sequence is important, its interactions with the genomes of the microbes living inside our body, (that is, the microbiome), should also be given heavy value in terms of affecting the risk and predisposition to disease [12].

There are tremendous inter and intra-individual genetic variations, such as those found in a tumour, that also account for different disease phenotypes [13, 14]. Phenotypic data must be collected at different levels, not only as individual clinical data, but also as molecular information, including transcriptomics, proteomics and metabolomics [15]. These latter are extremely relevant in understanding genome regulation, which is responsible for the synthesis of specific gene products [16].

Furthermore, there is a strong relationship between the environment and our genetic information, which we now better understand through epigenomic studies [17]. With all of these factors now in our perspective, we are starting to increasingly recognize the importance of the role that the exposure to environmental risk factors (exposome) plays in the development of many, if not all diseases [18].

With this in mind, it is not surprising that health care professionals have, in the last two decades, started to base their decisions for the best course of treatments and preventions on the patient's specific health data and needs, thereby ‘personalizing’ the patient's clinical experience. This is where personalized medicine, a medical practice based on a patient's genetic profile comes into play. This term, which was coined in 1999 by two journalists from the Wall Street Journal [19] is associated with the most recent concept of precision medicine. There is however a significant difference between the two.

Whereas personalized medicine could be suggested to deal, in its majority with genomic and phenotypic data, such as that included in clinical records, precision medicine aims to combine the knowledge of the patient's personal characteristics with the traditional medical records, in addition to including any relevant environmental factors or risks [20]. Personalized medicine revolves around a kind of static or snapshot view of the patient's genetic information, in contrast with precision medicine, which allows us to create dynamic stratifications of patients through modeling the individual's life journey, taking time into account. More importantly, precision medicine does not only rely on genomic data and clinical parameters, but also integrates new and important sources of information, such as non-genomic biological data and the patient's lifestyle patterns, or in other words, exposomic, metabolomic, microbiomic or epigenomic data. These differences are illustrated in Table 1.

As Clayton Christensen explained in 2008, “precision medicine is a new approach to discover and develop medicines, vaccines, or other interventions that enable disease prevention and deliver superior therapeutic outcomes for patients” [21]. This concept was publicly adopted in 2011, following the publication of the report “Toward Precision Medicine: Building a Knowledge Network for Biomedical Research and a New Taxonomy of Disease” by the US National Academy of Sciences [22]. According to this document, precision medicine could be defined as an emerging model that customizes health care tailored to each individual, by developing a new and redefined taxonomy for disease classification. In doing this, we are using genomic, clinical, environmental and behavioural information to understand the biological basis and causes of a disease. In turn, this should lead to a better selection of disease targets, as well as to the identification of patient populations that demonstrate improved clinical outcomes to novel preventative and therapeutic approaches [23].

### 3.2 Participatory Medicine

Another principal research avenue in biomedicine is participatory medicine, an idea related to the rise of the electronic patient, known as the e-patient. This notion was long ago explored in 1973 by Shenkin and Warner’s New England Journal of Medicine article “Giving the patient his medical record: a proposal to improve the system” [24]. Tom Ferguson, an American scientific author and physician originally created the term ‘e-patient’ and was successful with his book “e-patient: how they can help us to heal healthcare”. Empowering patients to take control over their own health is progressively becoming a catalyst to enhancing our healthcare system, and the evidence in the literature strongly supports this [25].

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<th>Criteria</th>
<th>Personalized medicine</th>
<th>Precision medicine</th>
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| Main goal          | Improving tailored therapy  
• Looking for the right drug for the right people | Improving diagnosis, therapy and predictive models  
• Looking for the right drug for the right disease  
• New taxonomy of disease and disease reclassification |
| Decision support   | Focused on individual data + evidence                                                  | Based on large population resources                                                 |
| Products           | Companion diagnostics to chose best therapy                                           | New/refined diagnostics methods                                                     |
| Data sources       | Bi-dimensional – use of genomics and clinical data                                     | Multidimensional – use of genomics, clinical, molecular (multi-omics) and other  
(i.e. exposome) data sources                                                        |
| Patient monitoring | Discrete, static – “Snapshot”                                                         | Continuous, dynamic stratification – Modeling patient journey                       |
| Approach           | Reductionist                                                                         | Integrative, system thinking                                                        |
| Role of patient    | Passive                                                                              | Active through consumer health devices – patient generated data                      |
Moreover, in 2009, the Society for Participatory Medicine was established, which defined this concept as "a movement allowing patients to be empowered, engaged, informed and involved in both decision making, management, sharing and learning about their own health" [26]. In addition, this society has created the Journal of Participatory Medicine, and through this, e-patients have been able to reach an extremely high level of public visibility by sharing their experiences and opinions related to healthcare systems and various health issues [27].

Patient advocates like Regina Holiday, Hugo Campos, e-patient Dave or Salvatore Iaconessi, amongst others, are also attempting to convey the significance of the active participation that patients should have in improving health care [28]. They have radically done this through the creation of slogans like 'give me my damn data', 'the patient will see you now', 'let patients help', or 'nothing about me without me'.

However, why exactly is participatory medicine relevant for biomedical informatics? Participatory medicine is the result of the convergence between self-tracking devices, social networks, mobile devices, sensors, personal health records, games and the many other tools that currently impact this area of digital medicine. Patients can now access and store personal testing data, diagnosis and treatment options through different online services; they can manage any medical images and health information online and make it available to others for feedback or advice. They can additionally choose to share it through social media, and for instance, initiate their own clinical trials in platforms such as 'Patients like me' [29]. More evidently, patients can monitor themselves through the use of health-related devices and sensors, such as those promoted by the movement known as 'The Quantified Self' [7, 30], and enter their data into personal health records. And not surprisingly, customers now can even have their own personal genome and microbiome available through direct consumer services such as 23andMe or Mbiome, respectively. The importance of patient-reported outcomes and shared decision making between patient and physician, is only increasing from here.

Importantly, the common element to all of these aspects of participatory medicine is the management of individual or patient-generated health information, which clearly belongs to the field of biomedical informatics, in particular, to personal or consumer health informatics.

4. The Role of Biomedical Informatics in Digital Medicine

Biomedical informatics is a vital discipline for digital medicine, as it can enable the progress of these two new research opportunities (precision medicine and participatory medicine), increasing the evidence base, guaranteeing high quality data, improving systems usability and facilitating change management that pays close attention to the information issues at hand. The following aspects represent three main examples of this.

4.1 Therapeutic Affordances of Social Media

It is widely known that chronic disease management is one of the main focuses of medicine today, and its rise within the ageing population continues to bring a tremendous burden on the health care system. It has also become evident that many of the sufferers of chronic diseases have taken to social media as a manner of coping with the psychological and social impacts of illness. With this in mind, the University of Melbourne has been developing a project called "Therapeutic Affordances of Social Media" conducted mainly by PhD student Mark Merolli, under the co-supervision of Dr Kathleen Gray and Prof Martin-Sanchez. It was our aim to analyse why and how patients with chronic disease are currently using social media, and to develop a framework to generate evidence useful for the design of social media interventions for chronic disease patients [31]. This led us to our research question: how can we explain social media’s effect on the health outcomes of people with chronic disease?

Our research project had three phases. We firstly undertook a literature review to identify the existing evidence in the use of social media by chronic disease patients [32]. Secondly, we conducted a global online survey using PROMIS (Patient Reported Outcome’s Management Information System) [33] as a way to measure beneficial outcomes from those interventions from the perspective of the patient, using a standardised set of questionnaires [34, 35]. Finally, we carried out a pilot clinical study in people with chronic pain to evaluate how using social media was improving their health [36].

Through our literature analysis we identified a few of the affordances of social media: self-presentation, connection, exploration, narration, and adaptation [32]. Affordance is a concept borrowed from design, and refers to those aspects that make people use an object in a particular way. Social media allows patients to anonymise themselves through self-presentation, connect to others, explore more about their disease, narrate their story, and many platforms also adapt to a patient’s disease status. All of these affordances can be provided by not just one, but by various social media platforms. Platforms change very often, yet the way in which patients interact with them will always be relevant [34].

Upon deeper analysis of our survey and pilot study, we were also able to see how those affordances were statistically correlated with good health outcomes [35]. We found that several outcomes were having significantly positive effects, in the social, psychological and cognitive dimensions of the chronic patients’ health [36].

4.2 Self-Quantification

A Biomedical informatics approach can also contribute to the improvement of health outcomes through the use of digital medicine technologies, such as self-quantification. One of our other PhD students Manal Almalki, has been actively working on this area for three years now under the co-supervision of Prof Martin-Sanchez and Dr Kathleen Gray.

The concept of self-quantification was developed by the Quantified Self community [30], an international collaboration of users and developers who share an interest in self-knowledge through the use of
sensors, personal devices, and apps to self-monitor and measure aspects of their health.

In 2011, we were successful in getting a project Self-Omics funded by the Institute for a Broadband-Enabled Society in Australia [37]. This project aimed to address the information and communication needs of the ‘quantified individual’ for enabling participatory and personalised medicine. Through this funding we were able to purchase most of the self-monitoring devices that were available at that time in Australia (over 50) and set up a Quantified-Self (QS) Lab. This project provides a clear representation of the role that biomedical informatics can play to improve health outcomes derived from self-quantification.

Through the use of the (QS) Lab we investigated how information flows from the self-monitoring device to the smartphone, from the smartphone to the cloud, and the different architectures for information management, communication protocols and user interfaces in these devices. Immediately, we realised that to measure an aspect such as cardiovascular risk, the use of several devices was needed (to measure weight, blood pressure, sleep pattern, physical activity and calories burnt). However, the need to use up to five different devices, makes it difficult to integrate the data into one location and support decision making and behaviour change by users. However, as we discovered, the integration can actually been done in one of three different ways: through Personal Health Records as we did (with Microsoft HealthVault), manually (e.g. using Excel files), or through integrated platforms such as Google Fit, HealthKit, WebMD Healthy Target, Samsung S.A.M.I, Philips-Salesforce, Qualcomm Life 2net, Open Humans, Human API and Validic.

We also delved into how the connection between the activity theory from Information Systems and the theory of patient activation with the aim of determining how the complexity of the integration of data affects patient engagement.

In a white paper that we published at the end of this project, titled “Self-Quantification: The Informatics of Personal Data Management for Health and Fitness” [38], we proposed a classification for self-quantification systems (devices and software) and a classification for activities, such as sleep or physical exercise, that can be measured with self-monitoring devices, based on the WHO International Classification of Functioning, Disability and Health (ICF) [39].

These taxonomies could allow biomedical informatics to then be used to address the need there is now to standardise how we annotate and describe experiments in this area, leading us to higher quality data. This is similar to what the bioinformatics community did 15 years ago, with the development of the first minimum information standards that were applied into genomics and proteomics experiments. One clear example of this is MIAME: the Minimum Information About a Microarray Experiment [40].

As we had already developed the classification for the devices and aspects that we wanted to measure, it was not difficult to conjure up a minimum information standard [41]. This standard, like a template, will allow self-monitoring experiments to be annotated, and the sample, device, variable being measured, as well as the method of generating data to be declared clearly. This standard is called MISME (Minimum Information About a Self-Monitoring Experiment) and is still being worked on today, although an initial version has been already uploaded into Biosharing portal [42].

More importantly, we wanted to study how the data coming from these self-monitoring devices can be used to obtain behavioural changes, such as when an individual quits smoking. By doing this, we may be able to find out how we can support these changes and search for methods that allow patients to adhere to these changes in behaviour; perhaps through more engaging apps, personalised SMS messages, or even risk prediction algorithms.

### 4.3 Exposome Informatics

In a recent paper about Exposome Informatics [43], we referred to the definition of the exposome as the lifelong exposure to environmental risk factors of an individual, complementing the genome. This is extremely relevant to consider, as we know that the environment can hugely influence the health trajectory in the life of a patient. In this article, we also explored the impact that the exposome can have on Biomedical Informatics, since the consideration of...

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**Figure 3** Connecting the genotype to the phenotype through environmental data requires the contribution from Bioinformatics and Health Informatics (adapted from [46]).
Self-Quantification as an interface to the individual exposome, places this area very nicely at the intersection between participatory and precision medicine.

The interplay between the genotype and the environment (expotype) explains an individual's phenotype, i.e. whether it is healthy or diseased. In terms of the development of a complex disease, environmental and behavioural factors can be seen as decisive a factor as the combination of several genes [44, 45]. With this knowledge in hand, it is useful to remember that our final objective as biomedical informaticians is to be able to understand how diseases are developed, contributing to explaining their underlying causes. In order to do that, we need to take into account how the environment interacts with genomic information and produces a particular phenotype.

Therefore, it is vital to ensure that there is a collaboration between Bioinformatics, which has a heavier focus on the molecular side, i.e. genomic, gene expression, proteomics, metabolomics etc., and Health Informatics, which provides methods to deal with clinical and environmental data, i.e. patient generated data, clinical data and population level data as shown in Figure 3.

4.4 The Changing Role of Biomedical Informatics

Twenty years ago, when biomedical informatics was only beginning to be properly defined, the approach that is based on the different levels of complexity of living beings proved to be advantageous. Such as when considering a hierarchy ascending from the molecule, to the cell, to the tissue, to the individual, and then to the population, for instance. At the molecular and cellular level, we can talk about bioinformatics, at the tissue and organ levels, we can talk about imaging informatics, at the individual level, we reach clinical informatics, and at the population level, it becomes public health informatics. This model allowed biomedical informatics to embrace the new data sets coming from genomics and the human genome project because it is clear that in order to understand disease, we need to take into account every layer of information, all of which output different data that requires integration [47]. However, the problem is that the environment is not clearly represented in the aforementioned approach, and the environment does affect every level of data. Although the population level has much to do with the environment, we now finally have the tools to assess the influence of environmental risk factors on individuals, not just in terms of larger communities. We can now collect the information we need (genomic, environmental, and phenotypic) from distinctive individuals. Moreover, not only are we able to do this with the aid of the most sophisticated equipment known to laboratories and clinics alike, but this collection of data is also possible with the use of affordable and ubiquitous technologies such as mobile phones and sensors (participatory health and self-quantification). Furthermore, now thanks partly to the role of biomedical informatics, all of the data obtained can be shared through social media, personal health records, or other clinical systems. This flow of information is depicted in Figure 4.

If we were to collect this data systematically, which a few individuals have labelled

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**Figure 4**
Collection of individual environmental, phenomic and genomic data is now possible, not only using sophisticated equipment, but also with affordable devices available to citizens.
the new human genome project: the Human Exposome Project [48], the idea of the exposome should thus definitely influence the design and development of future biomedical research information systems.

At the moment, there are in fact many exposome-related projects taking place around the globe. For instance, in the US, Hercules [49], a joint centre between Emory University and the Georgia Institute of Technology has been funded by the National Institute for Environmental Health Sciences. In addition to this, the European Commission has also supported several projects for large collaborative research networks, including HELIX [50, 51], Exposomics [52], Heals [53] and New Generis [54]. All of these are dealing with different aspects and approaches to collect, integrate and analyse data from the Human Exposome.

The development of large-scale data repositories represents an important element for these new approaches in precision medicine. In this regard, biobanks, such as the UK Biobank [55] or more recently the “1 Million Genomes Cohort” included in the US White House’s Precision Medicine Initiative [23] represent critical resources providing well-annotated sample repositories for research. It is also worth highlighting that in the last couple of years several large scale studies, different to the clinical trials that we have become accustomed to, have taken place. Clinical trials customarily deal with a particular disease or health problem, and the subjects are often ill patients. However, we are now witnessing the development of very large-scale studies involving healthy individuals. For instance, the Health eHeart study [56, 57], the Google Baseline Health Project [58], the Health Data Exploration Project [59] and the 100 People Wellness Project people [60]. These studies are being conducted as we are now realising the importance of collecting data from all of the genome, exposome, and phenome. They are trying to identify the baseline, and the potential variables that could be vital in detecting disease even before the first symptoms appear. These studies could lead us to the core of preventive medicine.

From the perspective of Biomedical Informatics, it could be beneficial to consider a new way of structuring our skills, methods and expertise such that Biomedical Informatics is placed as the discipline that can deal with all three types of data: from the genome, from the exposome, and from the phenome, as well as with their interplay. Hence, our relevance and importance should clearly be maintained in projects and endeavours like the aforementioned.

5. Perspectives

In recent years, Biomedical Informatics has been facing competition from other disciplines that have approached our area of research, dressing it with newer and more appealing names, catching the attention of society, the media, and in some cases even funding agencies. Some of these areas, which seem to overlap with several of the traditionally considered core knowledge domains of Biomedical Informatics include “(Big) Data Science”, “Digital Health” or “In Silico Medicine”.

The rise of these evolving fields has been demonstrated through a Google Trends search for the last five years, showing the pattern of interest over time for the term Biomedical Informatics, in contrast with one of these emerging areas, Digital Health. As seen in Figure 5, whilst Biomedical Informatics has a slightly decreasing presence in the news, it is evident that the term Digital Health is increasingly becoming more popular.

A similar pattern is seen to be taking place with the terms (Big) Data Science and In Silico Medicine. This can notably be seen in the case of the US National Library of Medicine (NLM), which has supported training and research in Biomedical Informatics for many years, but yet has recently published its long-term scientific vision, citing the need to position itself as the “epicentre for biomedical Data Science” [61]. Stanford University has also recently announced the creation of a new Department with the name of Biomedical Data Science [62, 63]. Moreover, In Europe, particularly during the 7th Research Framework Programme (2007–2013), the Commission decided to focus most, if not all, of the research funded in ICT for Health on what is known as “Virtual Physiological Human”, or “In Silico Medicine” [64, 65]. This is a technological area based on multi-scale modelling and simulation of the human anatomy and physiology. Whilst In Silico Medicine should be considered a subset of Biomedical Informatics, the European Commission gave all importance and support to this new field.

5.1 Upcoming Trends and Associated Risks for Biomedical Informatics

5.1.1 Biomedical Informatics in Australia

Australia provides a clear example of a country in which the challenges posed by the aforementioned ‘new players’ in the biomedical information domain have begun to threaten the consolidation of biomedical informatics as a recognised discipline.

In Australia, Biomedical Informatics is not even the standard term used to describe this discipline. Instead, the terms Health Informatics, or e-health, have become predominant. More disturbing, however, is that Health Informatics still holds a substandard presence, at best, in those formal systems (Government coding and classification systems) describing education, no presence in systems in charge of occupations, and insufficient recognition in the areas defining research. A clear example of this is the fact that the main Australian and New Zealand Field of Research Coding System (ANZSRC) still identifies Health Informatics as a subgroup of Information and Computing Sciences research, under the subdivision 0807 Library and Information Studies [66].

The Australian member society of IMIA, HISA (Health Informatics Society of Australia) has prioritised the embracement of the domain of Digital Medicine, as seen through the organisation of several events such as the Personalised Medicine 2013, Big Data 2013 and Participatory Health 2014 conferences, chaired by Prof Martin-Sanchez, as well as through their focus on digital health in the recent HIC 2015. Nevertheless, they maintain a strong, focused presence as a Health Informatics or-
ganisation. This is clearly evident not only in their own name (HISA), but also in the title of their major annual congress HIC (Health Informatics Conference).

5.1.2 Biomedical Informatics in Spain

Spain is yet another case of the transition and development that is taking place between all of these related areas that could be pushing the discipline of Biomedical Informatics into redundancy. Spain has had a National Society of Health Informatics (SEIS) for more than thirty years [67]. The society is affiliated with both EFMI and IMIA. However, in recent months two new partnerships have risen that are clearly competing for the same space. First, the Digital Health Association (ASD), which was born with the aim “to share knowledge and create opportunities for debate on the use of information and communications technology (“ICT”) in the field of medicine and health” [68]. Secondly, the Association of Researchers in eHealth (AIES) has also been created. The AIES is a “non profit organization, aimed at the promotion and dissemination of eHealth in both Spain and Latin America” [69]. Once more, although the leadership of these new partnerships is mostly composed of individuals with experience in the health sector (clinicians, journalists, economists, consultants), the presence of biomedical informaticians is very limited.

It may be possible that these new associations were founded to fill a void left by SEIS, and it will be interesting to see how they evolve and if any approach or collaboration occurs between them. However, when these organisations hold annual events labelling them with terms such as eHealth, digital health, or health informatics, the lack of distinction between disciplines seems unlikely to contribute positively to strengthen the academic and professional situation of BMI.

5.2 Recommended Strategies for Biomedical Informatics

Although there might be different reasons explaining the incursions from these domains, they are supported quite often with the use of vague definitions that oversee their direct application in the realm of health information and the health environment, without considering its peculiarities. In addition, they generally ignore the presence of a previously existing discipline with its own body of knowledge and that is focused on the analysis and management of information and knowledge in human health related applications. The rise of these fields could perhaps also be at-
tributed to the possible perception of Biomedical Informatics as a rigid discipline focused only on particular aspects and lacking flexibility to efficiently deal with the challenges associated with new technologies and approaches. These new, and not so new, domains are becoming a real threat, which could rip and tear our discipline, stripping it away from some our most relevant areas of work.

Before we allow this to happen, we need to question the capacity of these new entities (that do not prioritise biomedical informatics) to address, from a scientific perspective, the informational problems that new research areas, such as participatory and precision medicine introduce. As appealing as they may sound, how will these new associations deal with issues such as system usability, change management, interoperability or standards, without a strong presence of professionals that have formal training in biomedical informatics?

As informaticians, we know how to deal with every kind of data there is: big, small, fast, smart. We have the tools and knowledge to implement and integrate standards for data collecting and annotation. As we venture into these new areas of research, we hold the title of experts in system design and human-computer interaction. We can apply our methods and knowledge to the creation and design of digital health solutions, such as devices, sensors and self-monitoring tools. Who better, then, to facilitate the empowerment of patients through the use of new technology, than us?

The Electronic Health Record is one of the best tools available to patients to allow them more control over their health, and the ability to integrate data coming from multiple different devices and wearables remains more than within our reach. This is an issue that we, as informaticians, could collectively address, as we can ensure adequate levels of interoperability and use of standards for digital solutions. Addressing cognitive issues, facilitating behaviour change, and supporting decision making by clinicians and patients is also well within our capabilities as biomedical informaticians.

Beyond this, the amount we could contribute to areas such as data analytics, data visualisation, and data presentation is infinite. We could add immensely to the foundation of scientific evidence for digital medicine, and allow it to advance into even greater territories.

It is also well within our expertise to handle potential privacy and security issues. Therefore, we should be the ones in charge of the management of organisational change and the professionals involved in implementing information systems. For instance, by beginning to consider how we could support n-of-1 trials, so that we may run clinical trials with solely one subject, over time we would open the door to a world of new research opportunities.

Are we sure that these emerging industries surrounding ‘digital wealth’ are truly concerned with improving the outcomes, efficiency and safety of clinical care? We firmly believe that these new areas of Digital Health, (Big) Data Science, and In Silico Medicine, need the expertise of biomedical informaticians in order to continue contributing to improving the health of our society.

6. Conclusions

In this paper, a few plausible definitions for the term Digital Medicine have been suggested. We have also delved into the basic concepts behind two of the main research avenues that show promise and importance in health care today: precision and participatory medicine. Through the use of several examples of current research projects, including affordances in social media, self-quantification and exposome informatics, it has been demonstrated that BMI plays a key role in the new area of Digital Medicine. Thus, the potential impact and associated risks for Biomedical Informatics, due to the development of Digital Health and other recent fields, such as (Big) Data Science or In Silico Medicine, was highlighted and explored.

In doing so, one important conclusion was reached: Medicine is now at a cross-road. From where we stand now, the challenges we face are endless. We need to be able to make earlier diagnoses, create more personalised therapies and clinical trials that are faster and more effective. We need to improve how we classify different diseases and we are lacking a good enough model of risk profiling for disease prediction and prevention. We need to control the costs of healthcare systems and push patients to take more responsibility for the maintenance of their own health. However, the most important thing that we need is to produce a system that is proactive. We need to emphasise prevention over cure.

Precision and participatory medicine both constitute ideas that contribute towards the more ideal concept of preventive medicine. Nevertheless, no matter what approach we adopt from whichever direction, the most decisive factor of progress is the proper use of data, information, knowledge, and evidence. This is where we, as biomedical informaticians, come in. This is what we have been trained to do. The future of preventative medicine is ripe with opportunities for biomedical informatics, and it is up to us to embrace these, and collaborate with other new emerging areas, to advance the health of our society.

This will not be easy. In venturing into these new territories of medicine, we do run the risk of diverting subconsciously back to our traditional areas of work within our comfort zone. There are no shortcuts on the road to success. We will need to constantly keep trying and keep pushing, keeping in mind that these new areas of study are already affecting how we design our educational programs to train the future generation of biomedical informaticians, and how we implement and evaluate our systems. If Biomedical Informatics does not start to significantly embrace and complement the emergence of these new disciplines, we could be in grave danger of becoming irrelevant. The cost of this to our patients and our society could be devastating.