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From e-Health to i-Health

Traversing Tomorrow's Healthcare Frontier

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Technology is a curious creature. The most meaningful changes not only descend upon us, seemingly out of the blue, but also completely shake up the way we live and work. In addition, what may appear to be The Next New Thing often masks something else, which offers the same promises but does so in a far more effective and profound manner.

New technologies rarely arrive by leaps and bounds. Indeed, most breakthroughs seem to have been shaped by stealth. And yet, once upon us, they have turned our world upside down – achieving far more than their inventors dreamed of. Consider the printing press and electricity, the motor car and the telephone, computers and the Internet - or the impact of birth control pills and antibiotics.

On the other hand, not infrequently, as one major new frontier of technology seems to be just settling in, another – close on its heels – rides upon its back, opening up a vaster vista beneath. Remember cell phones instead of pagers, DVDs against video-cassettes, the Web rather than CompuServe, or the personal computer versus Sinclair's ZX-81.

Necessity – The Mother of Reinvention, and Relevance

In recent decades, the most enduring new technologies have also been those which are future proofed, continuing to find additional uses and juxtaposing near-seamlessly with ever-newer breakthroughs. Think of the novel/still-emerging applications for personal computers after the inception of the Internet, and the use of the mobile phone for both computing and Web surfing.

Healthcare too will not be exempt from such trends, of technology forced to reinvent itself and grow, in order to meet real needs, and thus itself remain relevant and survive.

Indeed, even as e-Health programmes seemingly flourish across the globe, they may simply be concealing a more powerful and pervasive phenomenon.

This concerns the emerging era of personal and individual healthcare, or what can be termed i-Health. It will be driven digitally for you, me and everyone else.

Of Supply and Demand, from Concept to Experience

The differences between e-Health and i-Health are significant. While e-Health is largely about concepts, policy and infrastructure, i-Health will be about use. The first is pushed on the technology supply side, while i-Health is going to be demand-led, pulled by need and finessed by experience.

Most crucially (if subtly), i-Health is more about patients than physicians. It is about reality rather than potential, about largescale implementation at affordable prices, instead of the bells and whistles of pilot projects.

Such developments will demand considerable advance attention from both healthcare policymakers and IT managers.

In 2008, a major EU-funded study called 'Financing eHealth' (see page 12) found an over-emphasis on e-Health technology (for its own sake), at the cost of organisational change and real, lasting benefits for health systems. The study also highlighted inadequate financing for the e-Health

investment lifecycle, as well as an absence of long-term plans focusing on the entire e-Health user chain – in other words, all the way to what we call i-Health. Indeed, many seem to believe that getting e-Health projects started up was enough.

Driving Down the e-Health Highway

As a result, it is i-Health which is going to give flesh and blood to the e-Health skeleton. New forms of digital healthcare systems, fashioned through use, and continuously reshaped in the interests of users, is what the future will be about. The choice of terminology (i-Health rather than e-Health) may be one of the keys to adapting mindsets.

And yet, it remains vital to underline that i-Health would not be feasible without e-Health. Indeed, like other now-familiar technologies, i-Health is fuelled by the booming infrastructural spend on e-Health (from wireless/mobile systems and RFID to miniaturised sensors and more), as well as a steady fall in the enabling systems that drive both.

To use a popular analogy, good i-Health will be what drives along the e-Health highway. Both have their uses.

i-Health: The Symbols

So what then are going to be the main pillars of i-Health? There are two: consumer genomics and the electronic health record. On its part, healthcare IT both fuels and straddles these two technologies.

Consumer Genomics

The most potent, if still incipient, i-Health trend is that of consumer (or personal) genomics. Today, genomics technology is coming off the drawing board and close to making a mark on more and more people's lives.

A score of genomic vendors already offer a range of services from selective screening for some hundred-odd major disease genes to complete sequencing of a person's genome.

Dizzying Price Falls, Special Promotions

Costs, once prohibitive, are being driven down in something akin to a price war. They seem set to drop further in the future.

Such a pattern has been seen with other technological novelties, where unit prices fall to grow the user base, and lower prices then rapidly fuel further market penetration in a virtuous cycle.

Compared to the 3 billion dollar tag of the pioneering 2003 (Human) Genome Project (which furnished the first composite map of human DNA code), the plunge in prices for genome mapping is dizzying.

Privately-held American start-up 23andMe has been offering an analysis of DNA markers on 26 diseases for which there is an accepted genetic association (ranging from prostate cancer and Parkinson's to diabetes and Crohn's Disease), as well as another 72 where genetic factors are suspected (if not yet scientifically accepted). The cost: just 399 dollars – thanks to a special promotion which slashed prices from the previous 999 dollars.

European firms are also in the fray. Iceland's deCode Genetics has been offering its deCodeme personal genomic scanning service for just under 1,000 dollars (analysing about 1 million genetic variations, compared to 600,000 by 23andMe). More recently, in January 2009, deCode announced the launch of two scans – the first to assess major cardiovascular diseases (including heart attack, stroke, atrial fibrillation and peripheral artery disease) and the second aimed at common cancers (breast, prostate, lung, bladder, skin and colorectal). The cardiovascular conditions scan is priced at 195 dollars, and the one on cancers at 225 dollars.

Like the special promotion price from 23andMe, deCode also has taken account of marketing to galvanize demand; both scans can be ordered in a 'bundle' for 350 dollars – about equal to that of a higher-end mobile phone.

Map Your 6 Billion Base-Pair Genome for 1,000 Dollars

Prices for more comprehensive mapping remain high, but are also falling.

In 2007, the genome of Nobel laureate James Watson (one of the two scientists credited with the discovery of DNA) was decoded for approximately 1 million dollars.

By late 2008, American start-up Knome was offering to decode a person's entire 6 billion base-pair genome at 100,000 dollars (down sharply from 350,000 dollars at the middle of the year). The closest contender for broader appeal, however, is another US firm called Complete Genomics, now reported to be readying a whole-genome analysis for 5,000 dollars.

Industry experts foresee a trend towards a 1,000 dollar price point for decoding entire human genomes. This would make it a near-mass market technology, and is expected to be reached sometime early in the next decade.

Personal Medicine and à la Carte Insurance

The impact of consumer genomics will be sweeping. An especially striking new frontier will be personal medicine, where drugs, medicine combinations and dosages are prescribed according to a patient's specific genetic background rather than one grouping tens of thousands or more roughly approximate cases. Further down the horizon is an end to several inherited diseases.

A side-effect of personal medicine could be the emergence of 'à la carte' health insurance, providing choice of cover and premium based on a person's particular disease risks and (eventual) treatment requirements, rather than loading the highestrisk beneficiaries atop the lower-risk ones.

Not Yet, but Only a Few Years

In spite of warnings – from healthcare professionals to genomics companies themselves – about the danger of putting too much faith, too quickly in the technology, few doubt that it is a question of time before consumer genomics and i-Health become an everyday fact of life. While vendors of screening tools routinely warn patients that their services do not constitute 'medical advice', the Head of Genomics at the US Centers for Disease Control and Prevention notes in a review in the 'New England Journal of Medicine': "For the patient asking whether these services provide information that is useful for disease avoidance, the prudent answer is 'Not now — ask again in a few years'."

The best illustration of the emergence of genetics-based, personal mass medicine was a January 10, 2009 feature in 'The London Times'. This reported that the prestigious University College London (UCL) was, for the first time, offering genetic tests to track the risk of breast, ovarian and prostate cancer in people without a family history of these diseases.

The UCL programme is expected to eventually cover more conditions (e.g heart disease and diabetes) as well as screening of embryos by parents who carry a defective gene. Indeed, 'The London Times' story about the UCL screening accompanied news reports about the birth at the hospital of one of the world's first babies selected to be free of a genetic risk of breast cancer, in the shape of the BRCA1 gene; this gives a woman an 80 percent chance of developing breast cancer, and also raises the risk of ovarian and prostrate cancer in her offspring.

Genomics and the EHR Synergies from Healthcare IT

Consumer genomics will be a powerful new catalyst for the electronic health record (EHR). In turn, healthcare IT is seen as the only means to link and provide substance to both.

In the most basic terms, inclusion of genomic test results in an EHR is clearly the best way to personalize healthcare decision making. The immediate advantages – from avoidance of adverse reactions to a choice of optimal interventions – are clear. However, the longer-term objectives impact upon the broader foundations of healthcare culture itself. In the words of the American Health Information Community (AHIC), a genomic EHR would begin "the transition of the health care sector from a reactive to a predictive enterprise."

New Protocols and Standards

Given the novelty of genomic screening for healthcare professionals and the wide variety of tests and technologies in the field (within both the US itself and Europe), it is likely that the development of standards will take some time. Acceptance by physicians of genomics-based decision-support tools (known as Clinical Decision Support or CDS functionality) will take even longer.

In the meanwhile, efforts in terms of linking genomic data to EHRs are likely to remain focused on gaps in IT protocols, metrics and standards (covering terminology, coding, messaging etc.). Efforts in these fields are under way both in Europe and the US.

The LIS Cornerstone of Personalized Medicine

A cornerstone of personalized medicine will consist of Laboratory Information Systems (LIS), which capture data from genomic screening. Robust standards to address communication between LIS and EHRs are crucial to guarantee two-way transfer of information in the pre- and post-analytic phases, especially by extension to the Harmonized Use Case for EHRs (Laboratory Results Reporting).

The key goal, from the viewpoint of healthcare IT, is the need to ascertain that genomic data is not only well structured but that its integrity is ensured, and that it is transferred via secure, unified user and system interfaces to bioinformatics and clinical decision support personnel as well as other concerned parties.

Intelligent Reporting, CDS and the EHR

In the era of personal genomic medicine, the challenge of data design will be accompanied by a growing requirement for intelligent reporting. Most physicians are not trained to assess the significance of genomic data from their patients. As a result, streamlining and automating findings is likely to become crucial, as is the structuring of interpretations, to proactively assist physicians in the use of genomic data.

Automated CDS functionalities are therefore likely to be one of the key new challenges for healthcare IT.

Eventually, genomic reports are likely to be delivered to the clinician via an EHR. The key purpose of the latter would be to store and update genomic data (and its interpretations) in an organised manner and provide real-time access to authorised clinicians.

IT will again hold the key to the standardisation of results reporting and the reduction of variability in their interpretation – before they are transferred to an EHR.

The Future of i-Health: Challenges for Healthcare IT

The significance of the above factors cannot be under-estimated. Even today, most genomic data provides no more than probabilities of particular diseases (related to an average in a sample population). Thus, a 15.3 percent risk of getting prostate cancer before a patient turns 80, compared with an average risk of 20.4 percent in a specific sub-group, means little on its own.

One cannot speak of someone suffering from 15.3 percent prostate cancer. The need for correlating such a finding with other risk data and then making an interpretation will be a major challenge.

To make all this happen will require rapid growth in the number of both patient case histories and genomic databases. Such growth will in turn pose additional challenges for healthcare IT.

Most immediately, the sheer volume of information generated by genomics will drive a need for petabyte (and larger) databases. Grid computing architectures hosted on supercomputers or clusters and driven by highly sophisticated collaborative computing software are likely to become common, given the huge mass of historical data in EHRs and the need to both update it with even larger masses of new genomic information – and make all this, along with its intelligent interpretation - accessible in a distributed fashion.

As e-Health moves inexorably towards i-Health, the challenge of genomics will eventually shift from testing and data collection to ITmediated interpretation. One of the key challenges for policy makers today is to find ways to encourage such skills and provide incentives for adequate compensation and benefits.

As discussed elsewhere in this issue (see I Have a Dream – Barack Obama’s Healthcare IT Vision, page 25), one of the major challenges foreseen in the US in the near term is a shortage of IT skills to make Obama’s healthcare reforms a reality. This reflects a similar situation during the Silicon Valley boom years in the 1990s.

Such a comparison is not coincidental. “Genomics today is where the computer industry was in the 1970s,” according to Randal Scott, a former Chairman of Incyte Pharmaceuticals, Inc., which went on to become one of the first biotech firms to set up a dedicated genomics business.

And the i-Health wave is also irreversible. Leading US business magazine ‘Forbes’ foresees genomics technologies directly or indirectly contributing to about 20% of U.S. GDP by 2030. Not surprisingly, Time Magazine named 23andMe's Personal Genome Service (referred to previously) as the ‘Invention of the Year’ for 2008.

Genomics and EHRs: The US Case

In the US, a major step to build an official genomics framework for EHRs dates back to 2006, when the government's American Health Information Community (AHIC) set up a Personalized Healthcare Workgroup. The Workgroup, which consists of healthcare providers, drugs and diagnostics firms, laboratories and universities, patient advocacy groups and government agencies, had a mandate to find means to develop "standards for interoperable integration of genomic test information into personal e-health records."

Shortly before the Workgroup's establishment, the US Health Secretary observed that "…genomics will play an increasingly larger role in medicine, and now is the time to figure out how best to incorporate genetic information into e-health records, before multiple non-standard approaches take hold." The Workgroup is focusing on four issues:

Ó Genomic Tests

Ó Family Health History

Ó Clinical Decision Support

Ó Confidentiality, Privacy, and Security.

Genomics IT Infrastructure: An Example from Europe

Europe's EGCT (Advancing Clinico-Genomic Trials on Cancer) project is developing a unified technological infrastructure to facilitate secure, seamless access to genomic and clinical data, enriched by knowledge discovery operations and services. The project is already directed at an application area – to support multi-centric, post-genomic clinical trials, initially focused on cancer but evidently extensible to other areas.

The EGCT project has conceived an overall architecture for an integrated genomics platform. Its infrastructure uses a common set of services and service registrations for the entire cancer clinical trials community.

Key points of emphasis are:

Ó Standards and models for exposing web services (semantics)

Ó Modelling and simulation tools from the molecular to the systems biology level

Ó Data mining tools for knowledge discovery

Ó Domain-specific ontologies and data representation models to permit meta-analysis

Ó Customisable discovery workflows for design and secure execution by researchers

The EGCT has also set up cross-disciplinary task forces to propose guidelines on issues such as data sharing (including legal, regulatory, ethical and IP issues) and advanced security tools for data anonymisation. It is also developing enhanced standards for data protection in a web (grid) services environment.

The European Bioinformatics Institute

The European Bioinformatics Institute (EBI) is a centre for R&D in genomics and bioinformatics at Hinxton, Britain.

The roots of the EBI lie in the European Molecular Biology Laboratory (EMBL) Nucleotide Sequence Data Library set up in 1980 at Heidelberg, Germany. This was was the world's first nucleotide sequence computerized database of DNA sequences. The scope of the EMBL mandate grew, as direct electronic submissions of data began after the launch of genome projects, and the two (soon conjoined) projects soon found a new home in Britain at the Wellcome Trust Genome Campus, with funding provided by the latter as well as the EU Commission, the US National Institutes of Health, and 20 national governments.

The Hinxton facility (now known as EMBL-EBI) hosts two databases, one for nucleotide sequences and a second for protein sequences. In recent years, it has become a centre for excellence in bioinformatics.

The facility hosts a wide range of IT-related workgroups, one of the most crucial of which is the Database Research and Devel opment Group , which focuses entirely on the massive database-related challenges (size, complexity, and real-time interconnectivity) referred to elsewhere in this article.

Other workgroups with a specific IT focus include the External Services Group which develops Web Services APIs for EMBL-EBI tools, including a specialised Search Engine, servers etc.

Finally, a group headed by Peter Rice is investigating and advising on grid technology requirements (including requisite middleware) as well as application development (for virtual, collaborative applications) and participation in standards development.

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