



A genome's home: Genomics and health care reform

The coming year promises to usher in important reforms to the delivery of health care in the United States. Rising costs, questions about health care quality, and a business community rendered uncompetitive at least in part by the high cost of employee benefits have aligned strange bedfellows with significant political resources to affect change. The population at large seems ready for reform. *Prevention, personalized medicine, and the medical home* have become societal buzzwords, with much debate regarding the merits of each concept. The medical home model seems to be growing in currency among multiple segments of the health care ecosystem and has been endorsed by several influential health care provider groups as an essential part of any reform package. Though the exact definition of a *medical home* is slippery—one is reminded of the parable about the blind men and the elephant—one can make a strong argument that genomic information should have a room reserved in any model of a medical home that gains acceptance.

Tremendous progress has been made over the past 2 years in understanding the genomic underpinnings of many of the disorders most costly to our society. At the same time, the cost of obtaining genomic information has plummeted. Currently, an individual can obtain a “genome-wide scan” for less than \$500; such scans contain more than 500,000 bits of information from the genome with potential relevance to health and disease. As of the end of 2008, full genome sequencing

costs have decreased more than 10,000-fold from the costs of only 8 years ago—a rate of decline exceeding that of Moore’s law for decreasing cost of semiconductors in the computer industry. There is no evidence that the rate of decline is diminishing—many feel that complete genome sequencing will be available for a price comparable to that of a CT scan within the next 5 to 10 years. Though the rate of accumulation of data is currently outstripping our knowledge of what to do with it,

Second, the number of medications for which pharmacogenetic testing is potentially helpful is growing rapidly—and for a handful such testing is imperative. The availability of a repository of genomic sequence data in the context of a medical home seems likely to become a means to economically select the right drug at the right dose for the patient in question.

Third, numerous guidelines currently suggest that patients considering having children be offered the option

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this will not remain the case for long; and failure to develop health care systems that are capable of dealing with genomic information now will do future generations a disservice.

Consider four aspects of care delivery that might logically be part of an individual’s medical home and may potentially be greatly affected by the availability of low-cost, full-genome sequence information: 1) newborn screening; 2) pharmacotherapy; 3) reproductive counseling; and 4) disease risk management.


First, newborn screening panels are the earliest “genetic test” most people encounter. Currently most states test newborns for more than 29 disorders using biochemical measurements, some with very low positive predictive values. It seems likely that many of these tests could be enhanced by having full genome sequence information available from an early age either to replace the biochemical screen or to aid in the interpretations of ambiguous results.

of genetic testing for carrier status of a variety of conditions with implications for their potential offspring. Patients choosing to access such genomic sequence information in their medical home could do so without need of redundant testing.

Finally, a long and growing list of known genomic variants contribute to a person’s risk of developing common complex conditions like diabetes, heart disease, and cancer. Many of these variants are already being incorporated in disease risk assessment algorithms, and at least some are likely to prove to be helpful in improving both screening and prevention efforts. The availability of full genome sequence information in the context of the medical home could allow patients to access relevant genetic risk information of their choosing at the most appropriate point in life.

The above examples also illustrate that achieving the maximum potential of a genome-enabled (or, in fact, any) medical home will require continued progress in another major technologi-

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cal area—health information technology. One major issue that needs immediate attention, regardless of the ultimate fate of the medical home model, is that genomic data lack interoperability between the majority of electronic medical record systems. The Personalized Health Care Workgroup of the American Health Information Community initiated by former US Department of Health and Human Services Secretary Michael Leavitt has made a start towards addressing this point over the past several years. Early indications are that the Obama administration views both personalized medicine and the electronic medical record as important to the nation's health, but it is too early to know what initiatives may be put forward in these areas.

A somewhat more pedantic but potentially very costly problem is that billions of bits of genomic data on millions of individuals will overwhelm the storage capacity of current health information technology systems. In the wash of genomic data, information security will be another critical consideration.

Physician assistants are well-positioned to be co-architects of any emerging health care reform. The American Academy of Physician Assistants supports the idea of the patient-centered medical home. For the upcoming 111th Congress, the AAPA has made the medical home concept, and clear delineation of the PA role within it, part of their agenda (see www.aapa.org/gandp/pdf/AAPA_HC_Discussion%20Ltr_to_TDaschle_1208.pdf for more information).

As evidenced by this Genetics in Medicine column and numerous other educational activities, the PA community has also been a leader in adopting and enacting competencies for genetics and genomics education for its members. Continued vision and leadership by PAs will help to ensure that the genome has a home in the future of medicine. [JAAPA](#)