Family History in a Flash

News flash, November 22, 2007: Inexpensive, non-invasive genetic test detects 2-3 fold risk increase for diabetes, cardiovascular disease, stroke, colorectal cancer, breast cancer, and ovarian cancer. In some individuals the test performs even better, detecting much higher disease risk for less common disorders like hemochromatosis, thrombophilia, and alpha-1 antitrypsin deficiency. Total turnaround time for results: 20 minutes. This test is available on demand in your office or on the internet, and you can be reimbursed for performing it. And, this test is covered by all third party payers – since it costs absolutely nothing! The holy grail of the genomic era? Gene chip created by an up and coming biotech? Snake oil from late night TV? No. The U.S. Surgeon General's family history tool: My Family Health Portrait (see https://familyhistory.hhs.gov).

My Family Health Portrait (MFHP) is designed to be completed at home by patients, and can provide the health care provider with a bonanza of succinct, legible, organized family health information. To promote public awareness of the usefulness of family history as a health tool the U.S. Surgeon General has designated every Thanksgiving starting in 2004 as a day to remember and discuss family history. The National Human Genome Research Institute of the National Institutes of Health (NIH), as well as the Centers for Disease Control (CDC), the Health Resources and Services Administration (HRSA), and the Agency for Healthcare Research and Quality (AHRQ) have partnered with the Surgeon General's office on this project since its inception.

Many experts think that family history is truly the single best 'genetic test' available to primary care. Certainly it is the most readily accessible and cheapest. In addition to providing estimates of heritable disease risk, a careful family history can provide insights on family dynamics, shared environmental factors, and patient health concerns. For some providers, the family history serves as a jumping-off point for a broader discussion of disease screening, e.g. "even though you don't have a family history of colon cancer, you are over 50 and guidelines suggest you consider some form of colon cancer screening." Outside of the screening environment, family history data can be used to guide differential diagnosis and even, occasionally, to inform prognosis.

Those of you grounded in evidence-based medicine may ask if there is evidence that taking a generic family history during routine health maintenance visits improves health outcomes. Surprisingly, few studies have directly addressed this question. As with much in clinical medicine, the action of clinicians is guided largely by tradition – e.g., how many studies address the value of taking a past medical history, but would you care for a conscious patient without one? However, tradition is no longer an adequate basis to continue a practice in the setting of competing medical priorities. What is the evidence for the use of family history in primary care and what are the gaps in our understanding of the use of family history? To answer such questions, the NIH is sponsoring an extensive examination of the literature and impartial review of the evidence through an upcoming "State-of-the-Science" conference. However, one does need to await such a review to know that, at least for certain disorders – including such

common ones as heart disease, diabetes, and colon cancer - the benefits of utilizing family history information can be great.

You say family history isn't very flashy? What if a family history tool could be completed by the patient at home and then be used to populate your electronic medical record (EMR) via secure internet connection? What if when you opened the data for review, the EMR presented you with an expert interpretation of the data along with guideline-based management options? What if the family history data was automatically integrated with genetic test data to bolster the predictive value of the information, and point to interventions tailored to the patient's individual genetic makeup? Surprisingly, this level of capability in the EMR is not that far-fetched. For a number of months, multiple federal and private organizations have been working collaboratively through the Personalized Health Care workgroup of the American Health Information Community (http://www.hhs.gov/healthit/ahic/healthcare/) to develop standards for healthcare information technology to make this futuristic vision a fast approaching reality. At least some EMR systems should have this type of advanced capability within the next five years.

In a time of microarrays, '-omics', and talk of full genome sequencing, family history remains a cornerstone of the concept of truly personalized medicine – and seems likely to be only more useful, not less, in the near future.