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Personalized medicine: selected Web resources

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Abstract

Information about personalized medicine abounds, yet it is difficult to comprehensively search for information on this topic due to the broadness of the term "personalized medicine," the variety of terms that are used to describe this concept, the vast amount of pertinent journal articles and Web sites, and the fast pace of developments in this field. A selected list of Web sites is provided as a starting place for information about concepts, terminology, projects, databases, tools, and stakeholders related to personalized medicine.

Since the completion of the Human Genome Project in 2003, interest in "personalized medicine" and the quantity of journal literature and Web resources related to this topic has been burgeoning. Former US Department of Health and Human Services (HHS) Secretary, Michael O. Leavitt, made personalized medicine one of his priorities, and the US President, Barack Obama, was the author of the Genomics and Personalized Medicine Acts of 2006 and 2007. The attention and energies of these two high-level officials, as well as many others, have contributed to the continued US support for this research agenda. Kathleen Sebelius succeeded Michael O. Levitt as HHS Secretary on April 28, 2009. On May 5, 2009, a coalition representing more than a hundred genetic testing laboratories, patient advocates, investors, and health policy researchers sent the Secretary a letter describing their issues and concerns regarding personalized medicine.

As stated on the HHS personalized health care Web site, "Virtually every agency in the US Department of Health and Human Services participates actively in initiatives that are working toward the long-term goals of personalized health care. The integration of these efforts will act as a powerful force to achieve personalized patient care." The HHS issued two reports on US efforts related to personalized medicine. The first report (2007) "included summaries of federal efforts in the areas of expanding the science base for personalized health care; supporting health information technology; regulatory responsibilities; implementing personalized medical products and services in clinical practice; and ethical, legal and social issues." The second report (2008) "seeks to bring into focus a sampling of activities that are now underway in different parts of the private and academic health care sectors toward integrating personalized health care into clinical practice."

- HHS Personalized Health Care Initiative, US Department of Health and Human

  Services

  [http://www.hhs.gov/myhealthcare/]

- HHS Secretary's Advisory Committee on Genetics, Health & Society (SACGHS)

  [http://oba.od.nih.gov/sacghs/sacghs_home.html]

- "Personalized Health Care: Opportunities, Pathways, Resources," US Department of

  Health and Human Services, September 2007

  [http://www.hhs.gov/myhealthcare/news/phc-report.pdf]

- "Personalized Health Care Pioneers, Partnerships, Progress," US Department of Health

  and Human Services, November 2008

  [http://www.hhs.gov/myhealthcare/news/phc_2008_report.pdf]

- Letter to HHS Secretary, Kathleen Sebelius, May 5, 2009

  [http://www.dnapolicy.org/resources/LtrtoSecSebeliusrePersonalizedMedicine.pdf]

Another important milestone on the road to attaining personalized medicine was the passage of

the US Genetic Information Nondiscrimination Act (GINA) which was signed into law in May

2008, and was designed to prohibit the improper use of genetic information in health insurance

and employment.

- H.R. 493, Genetic Information Nondiscrimination Act of 2008

  [http://thomas.loc.gov/cgi-bin/bdquery/z?d110:HR00493:@@@L&summ2=m&]

Terminology

How can researchers and clinicians sift through the petabytes of information on the internet to find relevant information about personalized medicine? At the time of writing, a keyword search for "personalized medicine" in PubMed reveals hundreds of articles published in the last year alone, and that reflects just a tiny percentage of the articles on this topic. A Google search for the phrase "personalized medicine" now reveals over 500 000 results and that, too, is just the tip of the iceberg. Why are these search engines finding such a small percentage of the available information? The concept of personalized medicine is a broad one, and one that can be represented by many different terms and spellings such as personalized medicine, personalised medicine, personalized health care, personalized healthcare, individualized medicine, etc. In addition, there are many narrower topics, or related topics, covered by this umbrella term such as pharmacogenomics, biomarkers, neuromarkers, microarray analysis, single nucleotide polymorphism (SNP) profiling, electronic health records, and many more. The proliferation of "-omics" terms such as genomics, pharmacogenomics, proteomics, epigenomics, nutrigenomics, agrigenomics, metabonomics -- even neurogenomics -- is one signal of the infiltration of genomics into many different fields.[1] Another indicator is the number of recently published journals specifically dedicated to this topic that were started after 2002 (eg, Personalized Medicine, Current Pharmacogenomics and Personalized Medicine, Human Genomics and Proteomics, Genome Medicine, Genomic Medicine, BMC Medical Genomics, The Open Genomics Journal, etc.).

When searching PubMed for articles, it is often useful to search using National Library of Medicine Medical Subject Headings (MeSH) which are used to consistently categorize article references, and bring together references on a topic. If there is a good MeSH term (or terms) for a particular topic, researchers do not have to think of every single keyword and synonym that authors might have used to describe that concept. However, there is not a single MeSH term that covers the broad topic of personalized medicine, and existing MeSH terms such as "Pharmacogenetics," "Patient-Centered Care," "Genomics," "Genome, Human," "Genetics, Medical," "Proteomics," "Biomarkers," and "Medical Records Systems, Computerized" vary in how consistently they are applied.

Definitions of particular terms vary, also. It is hard to find a standard definition of many terms, including "personalized medicine," and sometimes people confuse one term with another (eg, "pharmacogenetics" vs. "pharmacogenomics").[2] The issue of terminology concerns everyone: researchers, clinicians, public policy decision makers, bioinformaticists, laypeople, as well as other stakeholders. The US National Human Genome Research Institute and US National Cancer Institute have created useful glossaries:

- Talking Glossary of Genetic Terms, US National Human Genome Research Institute
  [http://genome.gov/10002096]
- Dictionary of Cancer Terms, US National Cancer Institute
  [http://www.cancer.gov/dictionary/]

There is an excellent glossary in the new, comprehensive, two-volume book set, Genomic and Personalized Medicine, which was published by Elsevier/Academic Press in 2009.[3] This book set, available in print or electronically through ScienceDirect, is an excellent starting place for people who are trying to get an understanding of the many concepts and issues that comprise personalized medicine. The former US HHS Secretary, Michael O. Leavitt, wrote the foreword to this book. Section 12 of this book, titled "Neuropsychiatric Disease Genomic Medicine," includes eight chapters that discuss dementia, Parkinson's disease, epilepsy, ophthalmology, neuromuscular disorders, psychiatric disorders, depression, and bipolar disorder.

- Genomic and Personalized Medicine

    [http://www.science-direct.com/science/book/9780123694201]

In short, the terminology that is used in article databases such as PubMed as well as on various Web sites is wide-ranging and makes it difficult to pull all of the relevant information on this topic together.

Additional Web resources

There are thousands of Web sites that pertain to personalized medicine and its subtopics. Any collection, especially one in a "brief report" such as this, is necessarily a "selected" list. The following Web sites are provided as a sample of the range of projects and Web sites that are available.

- National Human Genome Research Institute (NHGRI)

  [http://genome.gov]

- PhenX Toolkit, NHGRI

  [https://www.phenxtoolkit.org/]

- ENCODE Project: ENCyclopedia of DNA Elements, NHGRI

  [http://www.genome.gov/ENCODE/]

- Ethical, Legal, and Social Implications ELSI Research Program, NHGRI

  [http://www.genome.gov/10001618]

- Human Genome Project, NHGRI

  [http://www.genome.gov/10001772]

- Pharmacogenetics Research Network, National Institute of General Medical Sciences

  (NIGMS)

  [http:www.nigms.nih.gov/pharmacogenetics]

- Environmental Genome Project, National Institute of Environmental Health Sciences

  (NIEHS)

  [http://www.niehs.nih.gov/research/supported/programs/egp/]

- NIH Chemical Genomics Center

  [http://www.ncgc.nih.gov/]

- NIH Data Sharing Policy for Genome Wide Association Studies (GWAS)

  [http://grants.nih.gov/grants/guide/notice-files/NOT-OD-07-088.html]

- NIH Roadmap for Medical Research

  [http://nihroadmap.nih.gov]

NIH neurosciences-related resources

- NIH Blueprint for Neuroscience Research

  [http://neuroscienceblueprint.nih.gov/]

- National Institute of Neurological Disorders and Stroke (NINDS): Clinical and

  Translational Resources

  [http://www.ninds.nih.gov/research/scientific_resources/clinical/]

- Neuroscience Information Framework (NIF)

  [http://www.neuinfo.org/]

National Center for Biotechnology Information (NCBI) databases

- Entrez - All Databases

  [http://www.ncbi.nlm.nih.gov/sites/gquery]

- Human Genome Resources

  [http://www.ncbi.nlm.nih.gov/projects/genome/guide/human/]

- Online Mendelian Inheritance in Man (OMIM)

  [http://www.ncbi.nlm.nih.gov/omim/]

- Entrez Gene

  [http://www.ncbi.nlm.nih.gov/sites/entrez?db=gene]

- Database of Genotypes and Phenotypes (dbGaP)

  [http://www.ncbi.nlm.nih.gov/sites/entrez?db=gap]

- Gene Expression Omnibus

  [http://www.ncbi.nlm.nih.gov/geo/]

- PubChem

  [http://pubchem.ncbi.nlm.nih.gov/]

- NCBI BioSystems

  [http://www.ncbi.nlm.nih.gov/biosystems/]

Other US federal government agency resources

- HHS Personalized Health Care: Federal Activities

  [http://www.hhs.gov/myhealthcare/activities/]

- HHS CDS Collaboratory

  [http://healthit.hhs.gov/portal/server.pt?open=512&objID=1230&parentname=

  CommunityPage&parentid=1&mode=2&in_hi_userid=10741&cached=true]

- CDC Public Health Genomics

  [http://www.cdc.gov/genomics/]

- CDC Evaluation of Genomic Applications in Practice and Prevention Initiative

  [http://www.cdc.gov/genomics/gtesting/EGAPP/about.htm]

- CDC Human Genome Epidemiology Network (HuGENet)

  [http://www.cdc.gov/genomics/hugenet/default.htm]

- CDC HuGE Navigator

  [http://www.hugenavigator.net/]

- CDC Genomics Workforce Competencies

  [http://www.cdc.gov/genomics/training/competencies/]

Nonprofit organizations

- Personalized Medicine Coalition

  [http://www.personalizedmedicinecoalition.org/]

- Genetic Alliance

  [http://www.geneticalliance.org/]

- Coalition for 21st Century Medicine

  [http://www.twentyfirstcenturymedicine.org/index.shtml]

Resources for the general public

- Genetics Home Reference

  [http://ghr.nlm.nih.gov/]

- "From Genes to Personalized Medicines," National Institutes of Health

  [http://www.nih.gov/about/researchresultsforthepublic/Genes_PersonalizedMed.pdf]

- "Genetics, Disease Prevention and Treatment," National Human Genome Research

  Institute (NHGRI)

  [http://www.genome.gov/19016938]

- MedlinePlus: Genes and Gene Therapy

  [http://www.nlm.nih.gov/medlineplus/genesandgenetherapy.html]

Commercial companies targeting the general public

- deCODEme

  [http://www.decodeme.com/]

- 23andMe

  [https://www.23andme.com/]

- Navigenics

  [http://www.navigenics.com/]

Miscellaneous resources

- Morningside Initiative, American Medical Informatics Association (AMIA)

  [http://www.amia.org/inside/initiatives/cds]

- "The Roadmap for National Action on Clinical Decision Support," AMIA

  [http://www.amia.org/files/cdsroadmap.pdf]

- National Coalition for Health Professional Education in Genetics (NCHPEG)

  [http://www.nchpeg.org]

- BIG Health Consortium

  [http://bighealthconsortium.org/]

- J. Craig Venter Institute

  [http://www.jcvi.org/]

- UCSC Genome Bioinformatics

  [http://genome.ucsc.edu/]

- GenMAPP: Gene Map Annotator and Pathway Profiler

  [http://www.genmapp.org/]

- Bioconductor: Open Source Software for Bioinformatics

  [http://www.bioconductor.org/]

- Brain Research and Integrative Neuroscience Network (BRAINet)

  [http://brainnet.net/]

- Allen Brain Atlas, Allen Institute for Brain Science

  [http://www.brain-map.org/]

Although this article focuses primarily on US projects and Web sites, progress towards making personalized medicine a reality is an international effort, as is reflected by this sample of project and tool Web sites:

International and non-US resources

- 1000 Genomes Project

  [http://www.1000genomes.org/page.php]

- Human Variome Project

  [http://www.humanvariomeproject.org/]

- International HapMap Project

  [http://www.hapmap.org/index.html.en]

- Structural Genomics Consortium

  [http://www.thesgconline.org/]

- GeneCards

  [http://www.genecards.org/]

- Ensembl Human Genome Browser

  [http://www.ensembl.org/Homo_sapiens/Info/]

- ArrayExpress Database

  [http://www.ebi.ac.uk/microarray-as/ae/]

- International Sequencing Consortium

  [http://www.intlgenome.org/]

- European Bioinformatics Institute

  [http://www.ebi.ac.uk/]

- Swiss Institute of Bioinformatics

  [http://www.isb-sib.ch/]

- Max Planck Institute for Molecular Genetics

  [http://www.molgen.mpg.de/]

- Nationales Genomforschungsnetz (NGFN)

  [http://www.ngfn.de/]

- Riken Genomic Sciences Research Complex (GSC)

  [http://www.gsc.riken.go.jp/]

- Kyoto Encyclopedia of Genes and Genomes

  [http://www.genome.jp/kegg/]

- ExPASy Proteomics Server

  [http://expasy.org/]

- European Proteomics Association (EuPA)

  [http://www.eupa.org/]

- HUPO: Human Proteome Organisation

  [http://www.hupo.org/]

- HUPO Brain Proteome Project

  [http://www.hbpp.org/5602.html]

Conclusion

Learning about the plethora of concepts, terminology, projects, databases, tools, and stakeholders involved in personalized medicine is a difficult task. For an overview with both breadth and depth, consulting the book by Willard and Ginsburg (mentioned above)[3] is highly recommended. Keeping up with new literature and other developments in specific areas of personalized medicine is also challenging. It is possible to follow new journal literature, in PubMed for example, by setting up search alerts for topics of interest, or alerts for tables of contents from particular journals. Another strategy is to create Google alerts, or to arrange with government agencies or other organizations to receive their news alerts. While e-mail alerts are one way to receive this information, an RSS (Really Simple Syndication) reader such as Google Reader (http://reader.google.com) is another way to easily and efficiently read and manage

alerts. Researchers at institutions that are fortunate enough to have a librarian or other information professional should consult them for advice on searching for information, managing what they find, and keeping informed about new developments, especially in fast-paced fields such as personalized medicine.

References

1.   Ozdemir V, Suarez-Kurtz G, Stenne R, et al. Risk assessment and communication tools for genotype associations with multifactorial phenotypes: the concept of "edge effect" and cultivating an ethical bridge between omics innovations and society. OMICS. 2009; 13:43-61.

2.   de Leon J. The future (or lack of future) of personalized prescription in psychiatry. Pharmacol Res. 2009; 59:81-9.

3.   Willard HF, Ginsburg GS, eds. Genomic and Personalized Medicine. 1st ed. Amsterdam, the Netherlands; Boston, MA: Elsevier/Academic Press; 2009.