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JAMA. 2008;299(11):1356-1358 (doi:10.1001/jama.299.11.1356)

<http://jama.ama-assn.org/cgi/content/full/299/11/1356>

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Key Internet Genetics Resources for the Clinician

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VIRTUALLY ALL PHYSICIANS, WHETHER PRIMARY CARE OR specialists, care for patients with health issues related to genetics. While the relationships between genes and disease have not always been apparent, the use of new tools such as genome-wide association studies and expression arrays means that the genetic aspects of prevention, diagnosis, therapeutics, and patient management will increasingly find their way into the clinic. Many patients want to know their risk for conditions based on family history, health history, age, or test results. Other patients, whether they have rare or common conditions, seek guidance for management decisions. As genomic information and genetic tests become more integrated into medical care, physicians need to know how to access genetic information and resources for their patients.

More than 10 years ago, *JAMA* published an article on Internet genetic resources.¹ Since then, the Internet has become more integrated into daily life, and genetic resources on the Web have proliferated.² The free resources listed in the BOX, while not an exhaustive list, are generally those that have existed for several years and that the genetics community commonly regards as key and credible. These references can be used to find information about genetic factors in health and in specific diseases, supportive resources for patients, family history tools, genetic testing, state laws on use of genetic information for insurance and employment, and directories of genetics clinics and genetics professionals.

Disease-Specific Information

Given the current pace of advances in identifying genes and their functions, the most up-to-date information on a specific condition often comes from a PubMed/MEDLINE search³ or use of a medical literature review program. Key Internet genetics resources are listed in the Box. GeneReviews is considered a go-to site for obtaining clinically oriented summaries about conditions including clinical features, differential diagnoses, management, inheritance, and genetic testing. Genetics Home Reference, geared for patients, provides brief summaries of conditions and links to resources and is also useful for physicians. Users of OMIM (Online Mendelian Inheritance in Man) can generate differential diagnoses by placing key clinical features in quotations and applying the search function; obtain a systems review of clinical fea-

tures for conditions in the Clinical Synopsis section; and access gene databases.

Patient Information and Support Groups. It takes only a few minutes to print contact information for a support group for patients. Many disease support groups have published educational resources for patients and their families and also have specifically developed materials for health care professionals. The Box includes Web sites for support group directories, genetic condition information for patients, and general genetics resources for patients who want to learn more about genetics and inheritance.

Family History Tools. In 2004, the US Surgeon General's Family History Initiative was launched, with Thanksgiving declared as National Family History Day. Information about this initiative and downloadable family history tools (in both English and Spanish) for the general public, along with other family history resources, are listed in the Box.

Genetic Tests. When caring for a patient who has concerns about genetics issues, a clinician's first response may be to determine whether a genetic test is available. For many conditions, genetic testing remains an evolving and moving target. More than 1000 genetic tests are now available,⁴ and hundreds more are moving through the research pipeline to clinical application. Determining the appropriate genetic test is critical, given that laboratories may offer different types of tests and use different methods to test for the same condition. In addition, genetic tests can be costly. Potential expense is an important consideration, particularly when genetic testing is ordered for asymptomatic patients; these tests are less likely to be fully covered by insurance than those ordered for symptomatic patients.^{5,6} Even when a patient is symptomatic, it can be difficult to obtain insurance coverage for a genetic test if the results will not significantly affect management and treatment.⁵

The interpretation and implications of genetic tests are often less straightforward than for more standardized medical tests, so the clinician needs to be particularly thoughtful in ordering them. It also may be prudent to consult a genetics professional when considering genetic testing that is not considered routine or standard care.

Genetic tests include chromosome-based, DNA-based, and biochemical tests. The online directories in the Box are

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Box. Key Internet Genetics Resources With Web Site Addresses**Directories of Genetics Clinics and Clinical Genetics Specialists**

GeneTests (select "Clinic Directory"): <http://www.genetests.org>
 American College of Medical Genetics (select "Find a Geneticist"): <http://www.acmg.net>
 National Society of Genetic Counselors: <http://www.nsgc.org/resourcelink.cfm>
 American Society of Human Genetics^a: http://www.ashg.org/pages/member_search.shtml
 Cancer Genetics Services Directory: http://www.cancer.gov/search/genetics_services/
 Genetic and Rare Diseases Information Center: <http://rarediseases.info.nih.gov/>

Disease-Specific Information for Health Care Professionals

GeneTests (select "GeneReviews"): <http://www.genetests.org>
 OMIM (Online Mendelian Inheritance in Man): <http://www.ncbi.nlm.nih.gov/sites/entrez?db=OMIM>
 Genes and Disease (NCBI) (organized by parts of the body affected): <http://www.ncbi.nlm.nih.gov/books/bv.fcgi?rid=gnd>
 Chromosomal Variation in Man: <http://www.wiley.com/legacy/products/subject/life/borgaonkar/access.html>

Patient Information and Support Groups

Genetics Home Reference: <http://www.ghr.nlm.nih.gov/>
 Genetic Alliance: <http://www.geneticalliance.org>
 National Organization for Rare Disorders: <http://www.raredisorders.org>
 GeneTests (search for condition, "Resources" link): <http://www.genetests.org>
 Genetic and Rare Diseases Information Center: <http://rarediseases.info.nih.gov/>
 Office of Rare Diseases, National Institutes of Health: <http://rarediseases.info.nih.gov/>
 Chromosome Deletion Outreach: <http://www.chromodisorder.org/CDO/>
 National Human Genome Research Institute (Specific Genetic Disorders page): <http://www.genome.gov/10001204>

Pregnancy, Birth Defects, Effects of Medications

March of Dimes: <http://www.marchofdimes.com/pnhec/4439.asp>
 Organization of Teratology Information Specialists (select "Fact Sheets" for medication information): <http://www.otispregnancy.org/>

Family History Tools

US Surgeon General's Family History Initiative: <http://www.hhs.gov/familyhistory/>
 Centers for Disease Control and Prevention (family history fact sheets, tools, resources): <http://www.cdc.gov/genomics/public/famhist.htm>
 American Medical Association (brochure and questionnaires for prenatal, pediatric, and adult patients): <http://www.ama-assn.org/ama/pub/category/2380.html>
 National Society of Genetic Counselors: <http://www.nsgc.org/consumer/familytree/index.cfm>
 Genetic Alliance (family history tools and resources for consumers): http://www.geneticalliance.org/ws_display.asp?filter=fhh

Genetic Tests

GeneTests (select "Laboratory Directory"; for DNA banking, select "Services"): <http://www.genetests.org>
 Biochemical Genetics Tests (biochemical genetic tests): <http://biochemgen.ucsd.edu/>
 National Newborn Screening & Genetics Resource Center (newborn screening tests, state newborn screening, and genetics programs): <http://genes-r-us.uthscsa.edu>
 National Society of Genetic Counselors—DNA banking information, patient brochure: <http://www.nsgc.org/dnabanking.cfm>

Guidelines for Genetic Testing

American College of Medical Genetics (select "Practice Guidelines" and "Policy Statements"): <http://www.acmg.net>
 American Society of Human Genetics (select "Policy/Advocacy"): <http://www.ashg.org>
 National Society of Genetic Counselors (select "Practice Guidelines" and "Position Statements"): <http://www.nsgc.org>
 National Guideline Clearinghouse (search for "genetic(s) and name of condition"): <http://www.guideline.gov>

Genetic Information—Implications for Insurance and Employment

National Human Genome Research Institute Policy and Legislative Database: <http://www.genome.gov/PolicyEthics/LegDatabase/pubsearch.cfm>; Policy & Ethics page: <http://www.genome.gov/PolicyEthics/>
 National Conference of State Legislatures—Genetics Laws and Legislative Activity: <http://www.ncsl.org/programs/health/Genetics/charts.htm>
 Genetic Alliance: http://www.geneticalliance.org/ws_display.asp?filter=policy.discrimination
 National Society of Genetic Counselors: http://www.nsgc.org/consumer/genetic_discrimination_resource.cfm

General Genetics Resources

National Human Genome Research Institute: <http://genome.gov/Education/>
 American Society of Human Genetics (select "Education"): <http://www.ashg.org>
 Dolan DNA Learning Center: <http://www.dnalc.org/>
 Genetic Science Learning Center: <http://learn.genetics.utah.edu/>
 Genetics Education Center: <http://www.kumc.edu/gec>

Gateways to Clinical Genetics Resources

Genetic Disorder Information on the Web: http://www.ornl.gov/sci/techresources/Human_Genome/posters/chromosome/diseaseindex.shtml
 Information for Genetic Professionals: <http://www.kumc.edu/gec/geneinfo.html>
 Office of Rare Diseases, National Institutes of Health: <http://rarediseases.info.nih.gov/>
 National Human Genome Research Institute Online Health and Support Resources: <http://www.genome.gov/11510197>

Resources to Help Clinicians Integrate Genetics Into Patient Care

National Coalition for Health Professional Education in Genetics: <http://www.nchpeg.org>
 GeneTests (select "Educational Materials" and "Genetic Tools"): <http://www.genetests.org>
 March of Dimes—Genetics and Your Practice: <http://marchofdimes.com/gyponline/index.bm2>

Clinical Genetics Specialists Professional Organizations

American College of Medical Genetics: <http://www.acmg.net>
 National Society of Genetic Counselors: <http://www.nsgc.org>
 International Society of Nurses in Genetics: <http://www.isong.org>
 American Board of Medical Genetics: <http://www.abmg.org>
 American Board of Genetic Counseling: <http://www.abgc.net>

^aCan be searched by name, city, or state but does not currently differentiate genetics researchers from clinicians, unless one cross-checks individual entries with certification status.

searchable by condition and generally provide direct links to laboratories offering the genetic test.

Guidelines for Genetic Testing

Guidelines and position statements are available regarding genetic testing of minors,⁷ prenatal and childhood genetic testing for adult-onset conditions,^{7,8} testing of children being placed for adoption,⁹⁻¹¹ professional disclosure of familial genetic information,¹² duty to recontact,¹³ and genetic screening or testing for different conditions¹⁴⁻¹⁷ (Box).

Direct-to-Consumer Internet Marketing of Genetic Tests. An increasing number of Web sites offer direct-to-consumer marketing of genetic tests. Some tests are directly applicable to health care, such as cystic fibrosis carrier testing. Others tests, at least currently, are less applicable to health care, such as nutrigenomics, ancestry, or paternity testing. Even when a clinician can easily discern whether a Web-based test is applicable to health care, a layperson may not. Similarly, it may be difficult to distinguish a credible site from one that looks good but actually overstates and misrepresents genetic testing information. Genetics organizations have issued policy statements that express concern and suggest guiding principles for direct-to-consumer genetic testing.¹⁸⁻²¹

Genetic Information—Implications for Insurance and Employment. Patients often have concerns about how genetic information can affect insurance coverage, including health, life, disability, and long-term care insurance. Currently, a patchwork of state laws differ widely in their content and extent of protection against genetic discrimination in insurance and employment. Despite significant efforts over the past decade to pass federal legislation on genetics discrimination, no such federal law exists as of this writing. The Box includes useful resources to find information about genetics laws in each state as well as information about genetic privacy, informed consent, and discrimination issues.

Directories of Genetics Clinics. Every state has one or more standing genetics clinics or provides periodic access to outreach clinics. Depending on location, however, a clinic in a neighboring state may be closer for a patient. The Box includes directories of genetics clinics and Web sites for professional and certifying organizations for clinical genetics specialists.

Resources to Help Clinicians Integrate Genetics Into Patient Care. The Box also presents gateways to available clinical genetics and educational resources that can help clinicians integrate genetics into patient care. The National Coalition for Health Professional Education in Genetics promotes health professional education and access to genetics resources. Its site includes core principles and competencies in genetics essential for all clinicians, a “Genetic Applications in Practice” newsletter, and a search engine for genetics resources. GeneTests’ “Educational Materials” and “Genetic Tools” sections include information about genetic services, family history “red flags,” genetic testing, and resources aimed at primary care physicians. Educational modules on these topics have also been developed by the March of Dimes and other groups.

Conclusion

It is challenging for clinicians to keep up with advances in their own areas of medicine, let alone in other specialties. As genetic information and genetic testing become more widely integrated into medical care, all clinicians will need to know how to access information and resources for their patients and themselves and how to refer patients to genetics specialists when indicated. Through the use of Internet genetics resources, clinicians can efficiently obtain valuable information to help them care for patients and help patients address genetic issues in their lives.

Financial Disclosures: None reported.

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