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The Role of National Library of Medicine® Web Sites in Newborn Screening Education

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Expanded newborn screening programs and subsequent detection of rare genetic disorders challenge parents and their medical providers to learn about the treatment and management of these disorders. Many people seek medical information on the Internet but may encounter requests for registration or fees, or find that resources are out of date, difficult to understand, or buried in advertisements. The U.S. National Library of Medicine (NLM), a component of the National Institutes of Health, provides web-based resources that address the challenges of newborn screening education. These resources include MedlinePlus®, Genetics Home Reference™, ClinicalTrials.gov, and PubMed®. NLM websites are not commercial, do not require registration or fees, and provide varied levels of information for a continuum of audiences from low-literacy consumers to health professionals. Using phenylketonuria as an example, this study describes the information that parents and their medical providers can find through NLM resources. NLM has embraced the digital age and provides the public with reliable, accurate, and up-to-date educational materials.

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Newborn screening programs in the United States began in the 1960s with a single test for phenylketonuria (PKU). In small increments, states expanded testing for additional disorders. By 1999, most states screened for fewer than 8 genetic disorders [National Newborn Screening and Genetics Resource Center, 1999]. The introduction of tandem mass spectrometry for newborn screening in the 1990s, however, allowed detection of 30 or more metabolic disorders from a single specimen [Chace et al., 2003]. This technological advance, coupled with recommendations for expanded newborn screening [Newborn Screening Task Force, 2000; Health Resources and Services Administration, 2005], accelerated the increase in the number of disorders that are included in state newborn screening programs. By 2005, half the states screened for 25 or more disorders [Therrell, 2005].

The challenge of educating healthcare providers and parents has intensified with expanded testing. Many of the disorders included in expanded screening programs are rare and may not be well understood by medical professionals who provide treatment and educate parents [Newborn Screening. . . , 2005]. The U.S. National Library of Medicine (NLM), a component of the National Institutes of Health (NIH), provides resources that address the challenges of newborn screening education.

THE U.S. NATIONAL LIBRARY OF MEDICINE—A BRIEF OVERVIEW

For more than 170 years, NLM has served the medical, research, and library communities. NLM's audience began to expand in 1997, when access to the biomedical literature database MEDLINE® was made freely available on the Web. As a consequence of free access, this resource—once used almost exclusively by medical librarians, scientists, and health professionals—was discovered by the public [Lacroix and Mehnert, 2002].

About 80% of U.S. Internet users have searched the Web for health information [Fox, 2005], including information about genetic disorders [Guttmacher, 2001; Taylor et al., 2001]. Consumers, however, assert that web sites with genetic information are often hard to understand and difficult to navigate [Bernhardt et al., 2002; Mitchell et al., 2003]. Patient education web sites that are user-friendly and provide lay-language materials often focus on a narrow range of disorders or charge a fee for these materials. With the growing public use of the Internet, NLM recognized that consumers wanted a broad spectrum of health information in easy-to-understand language. Beginning in 1998, NLM introduced new resources designed to help the general public and professionals find a wide range of health information that is accurate and up to date.

NLM WEB-BASED RESOURCES FOR NEWBORN SCREENING EDUCATION

NLM has several web-based resources that provide information about newborn screening and rare disorders detected through screening efforts. These resources include MedlinePlus, interactive health tutorials, Genetics Home Reference (GHR), ClinicalTrails.gov, and PubMed. They provide wide ranging

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levels of information for a continuum of audiences from low-literacy consumers to health professionals.

Table 1. MedlinePlus Quality Guidelines for Selecting Online Resources

Quality, authority and accuracy of content
Sites must provide the names of their advisory board members or be published by a government agency. MedlinePlus staff checks Board members' names in MEDLINE and other databases to ascertain whether they have published on the subjects covered by the web site. If the medical authority of the site is not evident, the MedlinePlus team contacts the site's Webmaster requesting for information or clarification.
Purpose of site
The site's primary purpose must be to provide health information, not to sell a product or service. Intrusive or content-linked advertisements disqualify pages from inclusion.
Most of the content must be free and not require registration.
Maintenance
The site must be consistently available, without broken links, and provide a Webmaster address. Pages must display dates.
Quality, nonredundant content
Because MedlinePlus is selective, not comprehensive, links on Health Topic should have minimal redundancy. Each linked document provides unique information to the consumer using that Health Topic page. Some links bring users a clear summary of an entire disease or condition, while others bring unique features, such as different reading levels, clear diagrams, illustrations, or interactive programs.

MedlinePlus

Introduced in 1998, MedlinePlus (http://medlineplus.gov/) is NLM's consumer health web portal [Miller et al., 2000]. MedlinePlus brings together authoritative information from NLM, NIH, and other government agencies and health related organizations. MedlinePlus also has extensive information about drugs, an illustrated medical encyclopedia, interactive patient tutorials, and the latest health news.

Health Topic pages are the core of MedlinePlus. These topic pages provide highly selective collections of links to Web documents, not comprehensive lists of everything on the Web. They point consumers to the best Web resources and minimize redundant listings. As of August 2006, 738 health topics were available on MedlinePlus. Many of these topics are also available in Spanish. MedlinePlus updates the Health Topic pages daily with press announcements from government organizations and with news stories from Reuters and HealthDay.

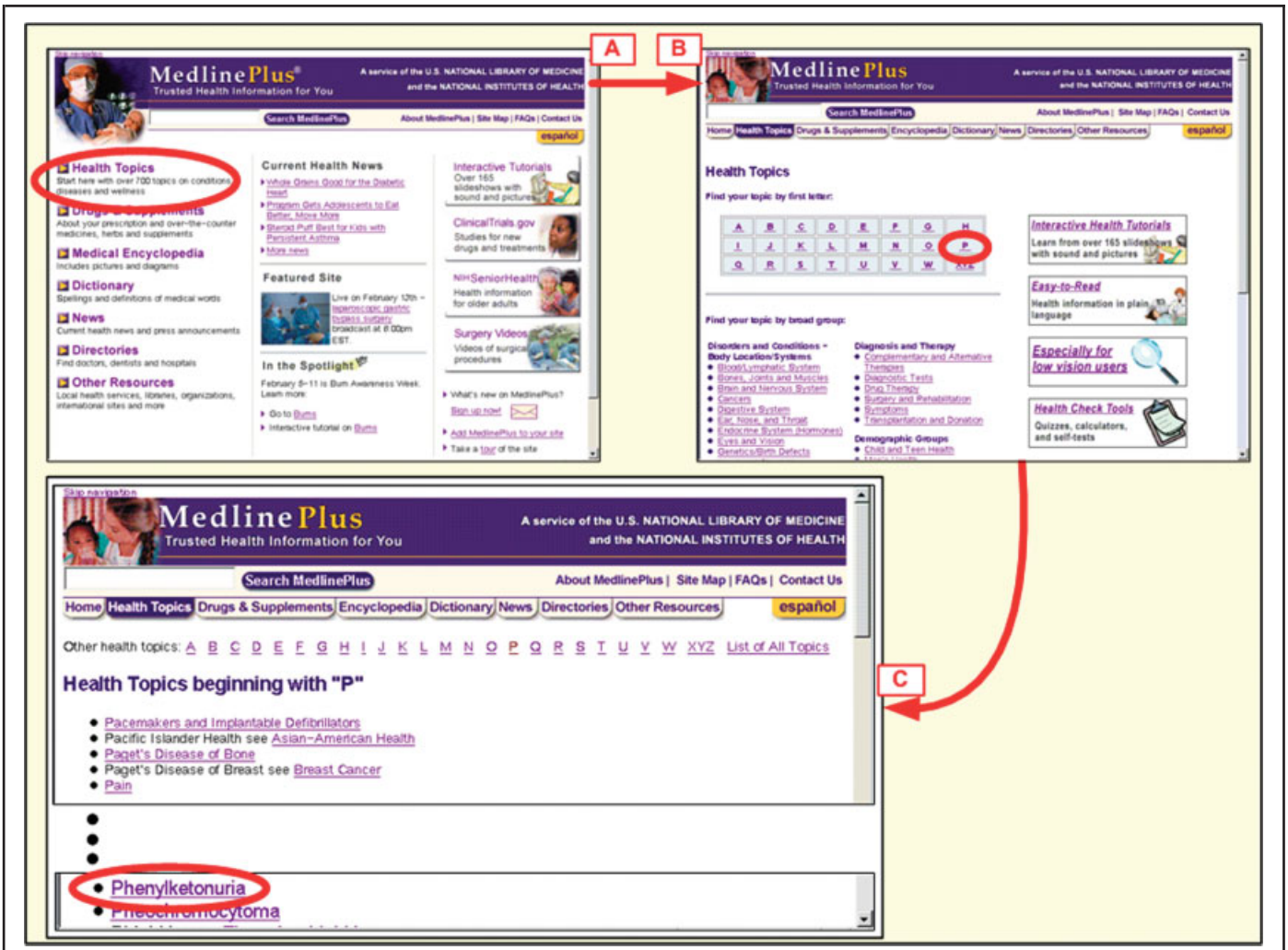

Fig. 1. Images A–C illustrate how to find the MedlinePlus health topic for phenylketonuria. [Color figure can be viewed in the online issue, which is available at www.interscience.wiley.com.]

The experienced biomedical librarians who create and maintain Health Topic pages organize them into categories such as overview, diagnosis, treatment, and prevention. They also include the source of online resources and identify easy-to read-materials and other special features such as pictures, diagrams, and flowcharts. The Health Topic pages function like a table of contents to chapters in an Internet book. This arrangement helps consumers easily scan the pages and provides them with a refuge from the overwhelming amounts or varying quality of Internet health information.

NLM uses established guidelines (Table 1) to identify and select high-quality information produced by other NIH Institutes, government organizations, and nongovernmental health information providers. MedlinePlus publishes these guidelines (http://www.nlm.nih.gov/medlineplus/criteria.html) so that organizations can evaluate themselves against the quality standards. These guidelines assure users that the information they find on Health Topic pages is reliable and up to date. MedlinePlus does not use cookies and does not request any personal information.

NLM has several web-based resources that provide information about newborn screening and rare disorders detected through screening efforts. These resources include MedlinePlus, interactive health tutorials, Genetics Home Reference, ClinicalTrails.gov, and PubMed.

Licensed content supplements the Health Topic pages in areas where authoritative Web content is not available. MedlinePlus licenses drug information, herbal and dietary supplement information, an illustrated medical encyclopedia, health news, a medical dictionary, and low-literacy interactive health tutorials.

Fig. 2. The health topic page links to other NLM resources, such as Interactive tutorials, Clinical trails.gov, and PubMed. The left hand side of the topic page serves as a table of contents for phenylalanine-specific resources and related topics. [Color figure can be viewed in the online issue, which is available at www.interscience.wiley.com.]

Interactive Health Tutorials

In partnership with the Patient Education Institute, NLM provides users with interactive health tutorials (http://www.nlm.nih.gov/medlineplus/tutorial.html) for nearly 200 topics. Using Flash technology, audio narration, and animated graphics, each tutorial explains a procedure or condition in easy-to-read language. A printed summary is available for each tutorial. The tutorials also include quizzes to reinforce major points of the presentation. All tutorials are available in both English and Spanish and are one of MedlinePlus's most popular features. They are an invaluable information source for patients, including those with low vision or low-literacy skills.

ClinicalTrials.gov

In 2000, NIH launched the ClinicalTrials.gov database (http://clinicaltrials.gov/) to provide consumers with access to medical research studies that seek to evaluate the safety and effectiveness of new drugs, medical procedures, or other means of treating, diagnosing, or preventing diseases. The database is maintained by NLM's Lister Hill National Center for Biomedical Communications [McCray and Ide, 2000]. For a wide range of diseases and conditions, ClinicalTrials.gov offers up-to-date information for locating federally and privately supported clinical trials in the United States and more than 120 countries. In 2005, more than a dozen major medical journals began requiring registry in ClinicalTrails.gov prior to publication of clinical studies, and the number of journals requiring registry is expected to grow. As of August 2006, the ClinicalTrials.gov database contained about 31,700 clinical studies.

For each study in the database, ClinicalTrials.gov provides a title, description, and design of the study; requirements for participation; locations where the study is available; and contact information. In addition, ClinicalTrials.gov indicates whether the study is ac-

Fig. 3. The interactive health tutorial provides a multimedia presentation on several facets of phenylketonuria (reproduced with permission from the Patient Education Institute (x-plain.com)). [Color figure can be viewed in the online issue, which is available at www.interscience.wiley.com.]

tively recruiting patients, not yet recruiting, no longer recruiting, or the study is completed. Learning aids are also available and include a glossary of terms used in clinical trials and a series of frequently asked questions that provide introductory information about clinical trials.

Genetics Home Reference

Launched in 2003, Genetics Home Reference (GHR) is an NLM web site (http://ghr.nlm.nih.gov) that provides consumer information about genetic conditions and the related genes and chromosomes [Mitchell et al., 2004]. Using a question-and-answer format, GHR provides short summaries of genetic disorders and the genetic variations responsible for these disorders. The site's content is continually monitored to ensure that it is accurate and up to date. As of August 2006, summaries were available for 205 conditions, 328 genes, and all the human chromosomes. GHR provides information for the 29 conditions included in the core screening panel recommended in the draft report by the Health Resources and Services Administration [2005].

GHR's materials are developed with the recognition that GHR users may have a limited science background. The summaries are written on a high-school level in lay-friendly language. Each condition summary provides a concise description of the disorder and an explanation of its incidence or prevalence, inheritance pattern, and genetic etiology. Summaries of related genes explain the gene's normal functions and the effect of mutations and polymorphisms on these functions. Chromosome summaries describe the basic characteristics of each chromosome, and some summaries include a discussion of related chromosomal disorders.

Each type of summary links to a wide spectrum of online resources to accommodate the varied needs of GHR users. The criteria used in selecting these online resources (http://ghr.nlm.nih.gov/ghr/page/Disclaimers) are similar to the MedlinePlus quality guidelines. GHR also offer learning aids such as glossary and an illustrated tutorial called the Help Me Understand Genetics Handbook.

PubMed

PubMed (http://pubmed.gov) is powerful but easy-to-use search system for MEDLINE. MEDLINE is the NLM's premier bibliographic database covering the fields of medicine, nursing, dentistry, veterinary medicine, the health care system, and the preclinical sciences. MEDLINE contains bibliographic citations and author abstracts from more than 4,800 biomedical journals published in the United States and 70 other countries. The database contains more than 16 million citations dating back to the mid 1950s. Coverage is worldwide, but most records are from English-language sources or have English abstracts. Free,

full-text articles are available from many journals through PubMed Central®, a digital archive of life sciences journal literature.

PubMed's LinkOut® feature provides access to a wide variety of relevant web-accessible resources, including full-text publications, biological databases, consumer health information, and research tools. Currently, citations from more than 4,600 journals are linked to the full text on publishers' web sites. Users may have to register, or there may be a fee or subscription required to access the full text. PubMed is an indispensable tool for users who want to read the latest research from professional medical journals.

PKU—A CASE STUDY IN USING NLM RESOURCES

PKU, a genetic disorder included in all newborn screening programs, will serve as an example to illustrate the educational features of NLM resources. The NLM web sites described earlier are crosslinked to one another. In this case study, a search will begin on the MedlinePlus web site and will illustrate connections to the other NLM resources and the authoritative information available to patients and health professionals.

Beginning with the MedlinePlus homepage (http://medlineplus.gov), the user chooses "Health Topics," then the letter "P," and the topic title for PKU or phenylketonuria, as diagrammed in Figure 1. Once on the topic page for phenylketonuria, the user can scan the contents of the page from the box in the left margin (Fig. 2). Each topic page is like a table of contents that allows the user to go quickly to a specific area of interest such as "Nutrition." Additionally, the left margin provides a tailored PubMed search of MEDLINE as well as links to related health topics that may be of interest to the user. For example, the link for Newborn Screening takes the user to a Health Topic page that covers the general aspects of newborn screening and provides resources describing disorders that are detected through screening programs.

The PKU Health Topic page also integrates links to other NLM products and the relevant NIH organization. On the right-hand side of the topic page, there are links to an interactive tutorial, GHR, and the National Institute for Child Health and Human Development—the primary NIH organization for PKU research. Clicking on the link to the interactive tutorial provides users with a multimedia presentation (Fig. 3) that explains the concepts of proteins and amino acids (particularly phenylalanine), PKU symptoms, the genetic cause of this disorder, PKU testing, treatment, and maternal PKU.

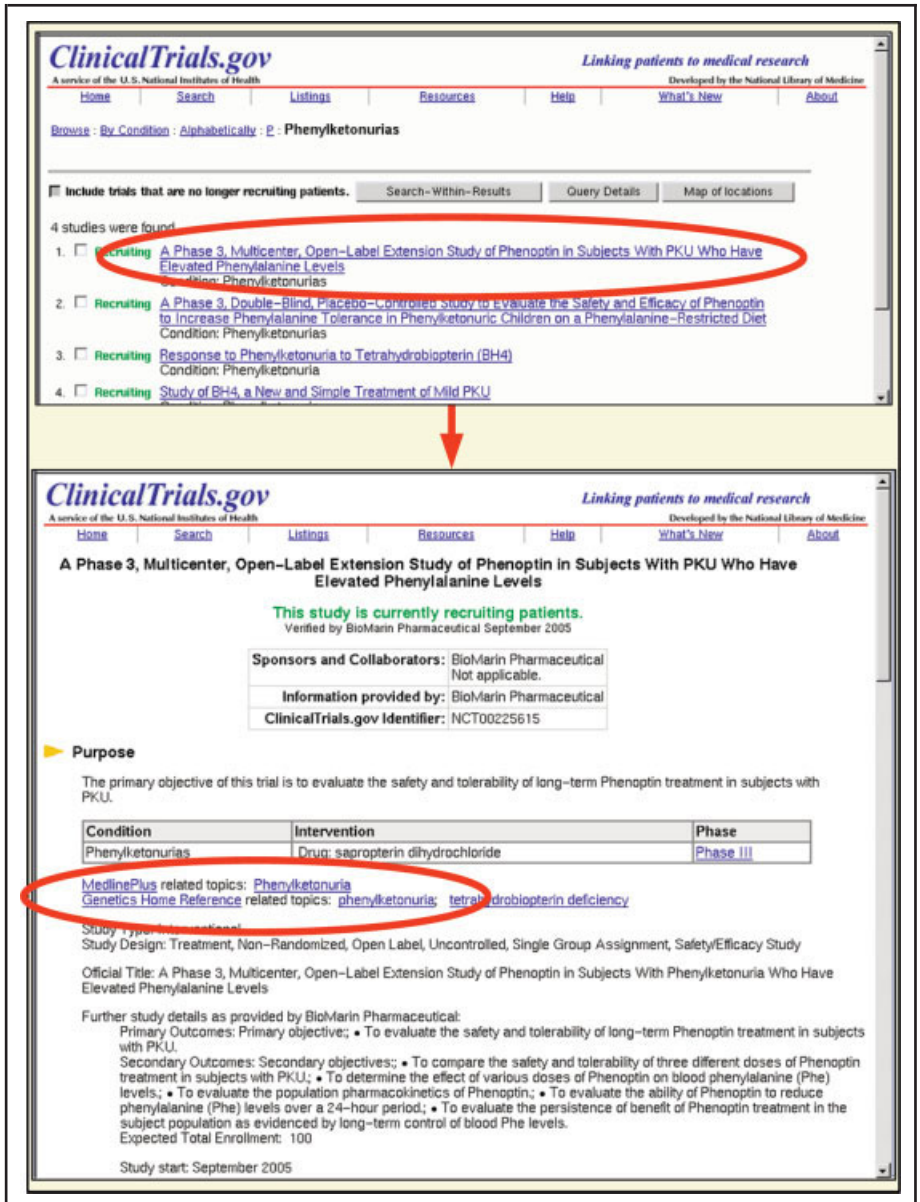

Fig. 4. A search of the ClinicalTrials database reveals several studies related to phenylketonuria. The information page for these trials provides links to MedlinePlus and Genetics Home Reference. [Color figure can be viewed in the online issue, which is available at www.interscience.wiley.com.]

If the user prefers to scroll through the Health Topic page, the first listings are for NIH publications and overviews of PKU from organizations such as the March of Dimes, followed by links to information on nutrition and related issues such as a babysitter's guide to PKU and facts about aspartame. Scrolling further down the page, users find links to ClinicalTrials.gov, genetic resources, support organizations, and laws and policies related to PKU. The link to ClinicalTrials.gov (also available from the left margin) provides the user with a list of PKU trials (Fig. 4) that are recruiting or not yet recruiting. Checking the box labeled "Include trials that are no longer recruiting patients" shows all trials for PKU, including trials no longer recruiting or completed. Clicking on a link to any of these trials will bring the user to an information page about the study. ClinicalTrials.gov information pages include links to MedlinePlus and GHR.

Selecting the link to GHR will bring the user to the GHR condition summary for PKU (Fig. 5). From this summary, users learn that PKU increases blood levels of phenylalanine, an amino acid that is obtained through the diet, and the disorder is detected in one in

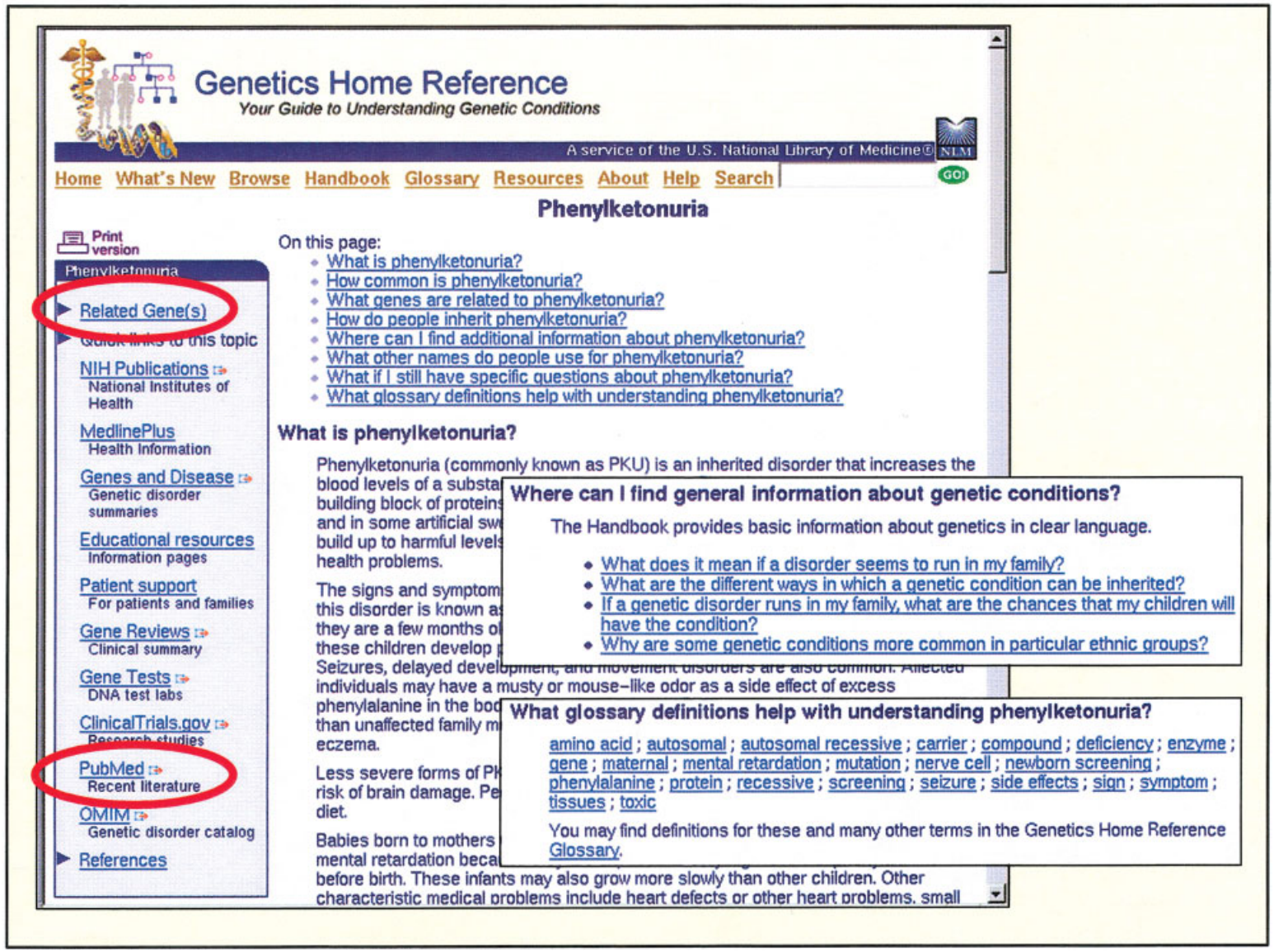

Fig. 5. Genetics Home Reference condition summaries provide learning aids such as relevant information in the tutorial Handbook and links to glossary terms. The left margin of each summary page offers quick links to the related gene(s) and additional resources such as PubMed. [Color figure can be viewed in the online issue, which is available at www.interscience.wiley.com.]

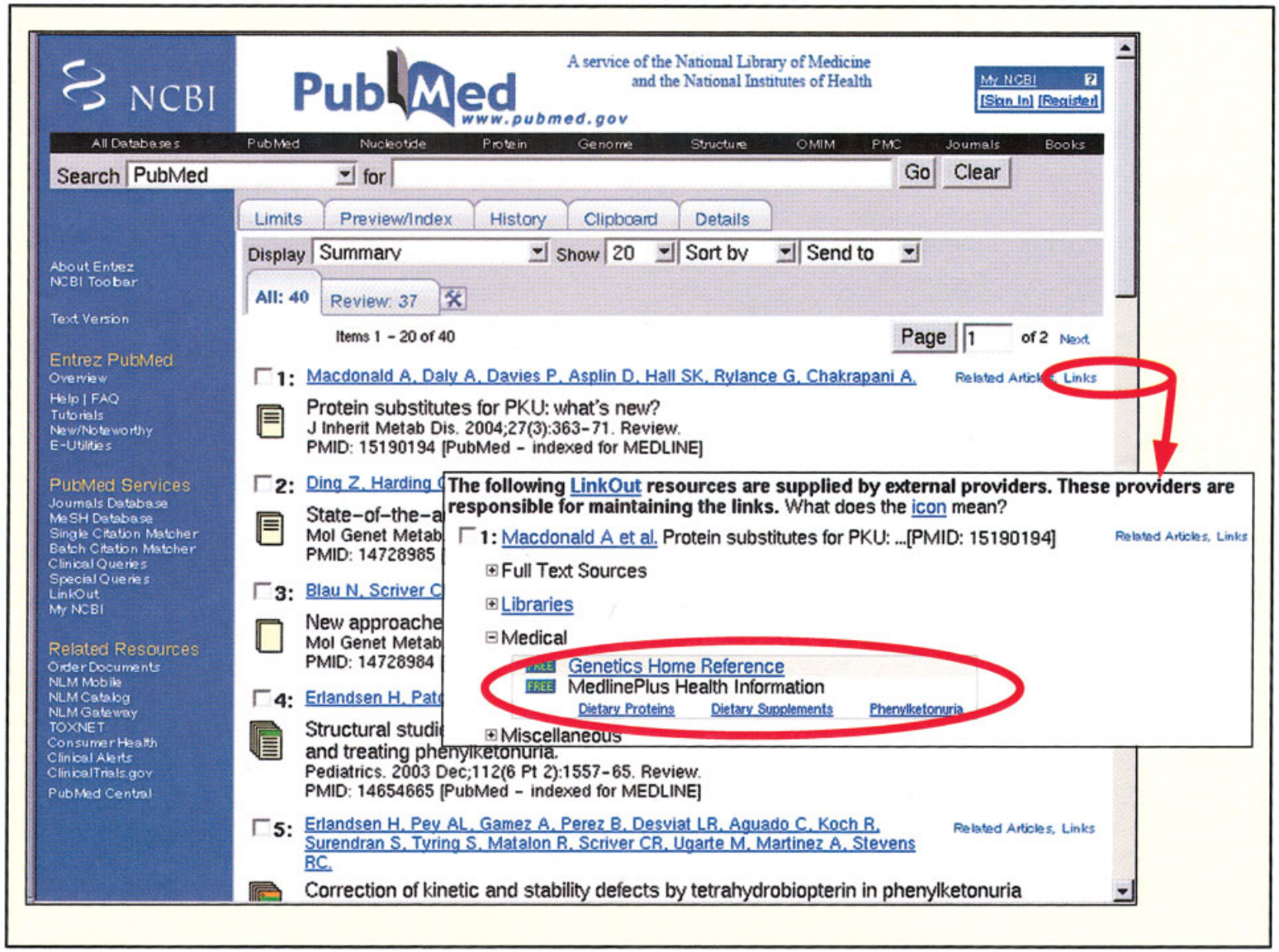

Fig. 6. A search using PubMed finds dozens of articles on phenylketonuria for the past year. The LinkOut feature can be used to find additional resources, such as MedlinePlus and Genetics Home Reference. [Color figure can be viewed in the online issue, which is available at www.interscience.wiley.com.]

10,000–15,000 newborns. The summary describes signs and symptoms and explains the concept of autosomal recessive inheritance. Users can also read a brief explanation of how mutations in the *PAH* gene cause PKU. The condition summary includes links to glossary terms and sections of the Handbook to help users with unfamiliar terminology or concepts. The left margin of the page offers links to the related gene and additional resources from NIH, NLM, and other organizations. These resources range in complexity from lay-level overviews to the genetics database Online Mendelian Inheritance of Man [Hamosh et al., 2005].

From the GHR summary, selecting the link to PubMed brings the user to a tailored search of PubMed/MEDLINE (Fig. 6). The search will find dozens of articles from the primary biomedical literature that report findings on treatment, management, screening, and *PAH* mutations. If an article is of particular interest, users can choose the "Related Articles" link to retrieve a precalculated set of PubMed citations that are closely related to the selected article. Users can also click on "Links," then LinkOut for a list of sources for a full text of the article as well as related resources such as the MedlinePlus Health Topic page for PKU.

This case study highlights the network of NLM resources that provides valuable information for parents and health providers.

SUMMARY

Expanded newborn screening and subsequent detection of rare genetic disorders challenges medical providers and parents to understand the consequences of these disorders. In 1998, NLM began to introduce web-based educational resources that are designed for the general public and are also suitable for healthcare professionals. These resources include MedlinePlus, GHR, ClinicalTrials.gov, and PubMed. These NLM web sites address dozens of disorders detected through newborn screening and provide users with a continuum of medical information ranging from easy-to-understand information about genetic etiology, inheritance, and incidence to available clinical trials and relevant biomedical literature. NLM web sites also direct users to additional online resources that meet quality standards. These government web sites are freely accessible and offer information that is reliable, accurate, and up to date. ■

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