

The Development and Use of the Genetic Testing Electronic Quality Information Portal, GeT-EQUIP

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Background

In 2005 the American College of Medical Genetics (ACMG) recommended a national uniform newborn screening (NBS) panel¹. Many states have adopted these recommendations, resulting in an increased need by healthcare professionals and the public for information regarding diagnosis, intervention, and management of the disorders. However, the desired information may not always be easily

accessible from public information resources. Based on recommendations from the CDC-hosted "Quality, Access, and Sustainability of Biochemical Genetic Testing Working Meeting" in October 2006, we assessed the current information resources for NBS diseases and related genetic testing and explored ways to develop a common information portal for general practitioners, specialists, laboratories, and the general public.

Table 1. Websites reviewed for development of the GeT-EQUIP, highlighted websites were included in the information portal

Website	Domain
ACTion Sheets	http://www.acmg.net/resources/policies/ACT/condition-analyte-links.htm
American Academy of Pediatrics	www.aap.org
American College of Medical Genetics	www.acmg.net
American College of Obstetricians and Gynecologists	www.acog.org
American Society of Human Genetics	www.ashg.org
EuroGene	www.eurogene.org
Genes and Disease	http://www.ncbi.nlm.nih.gov/books/bv.fcgi?rid=gnd
GeneTests	www.genetests.org
Genetic Alliance	www.geneticalliance.org
Genetics Home Reference	www.ghr.nlm.nih.gov
National Human Genome Research Institute	http://genome.gov/
National Coalition for Health Professional Education in Genetics	http://www.nchpeg.org/
National Institutes of Health, Office of Rare Diseases	http://rarediseases.info.nih.gov/asp/diseases/diseases.asp
National Laboratory Network for Rare Disease Genetic Testing	www.rarediseasetesting.org
National Newborn Screening and Genetics Resource Center	http://genes-r-us.uthscsa.edu/
National Organization for Rare Disorders	www.rarediseases.org
Online Mendelian Inheritance in Man	http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?db=OMIM
OrphaNet	www.orpha.net
WebMD	www.webmd.com
Wrong Diagnosis	www.wrongdiagnosis.com

1. Watson, et al. Newborn screening: toward a uniform screening panel and system. *Genetics in Medicine*. 2006; 8(5) 12S-252S.

Methods

Twenty commonly used websites (Table 1.) were assessed for availability of basic information, genetic testing information, laboratories, testing algorithm, sensitivity/ specificity of genetic tests, interpretation of test results, and

disease management for the 84 diseases on the ACMG NBS panel. Websites selected for the information portal contained "stand alone" information, or did not provide information from outside sources.

Results

Thirteen websites were included in the information portal. Two of these websites contained restricted-access content areas. Seven websites were excluded because they provided information from external

sources and/or lacked relevant information. Many websites lacked information on test algorithms, sensitivity/specificity, interpretation, and disease management.

Conclusions

Availability and accessibility of NBS information is an increasing public health need. Using the information compiled the Genetic Testing Electronic Quality Information Portal, or GeT-EQUIP was developed to provide a vehicle for

streamlined, user-friendly information search. This website is available through the CDC's website at : <http://wwwn.cdc.gov/dls/genetics/getequip/portal.aspx> (figures 1-3).

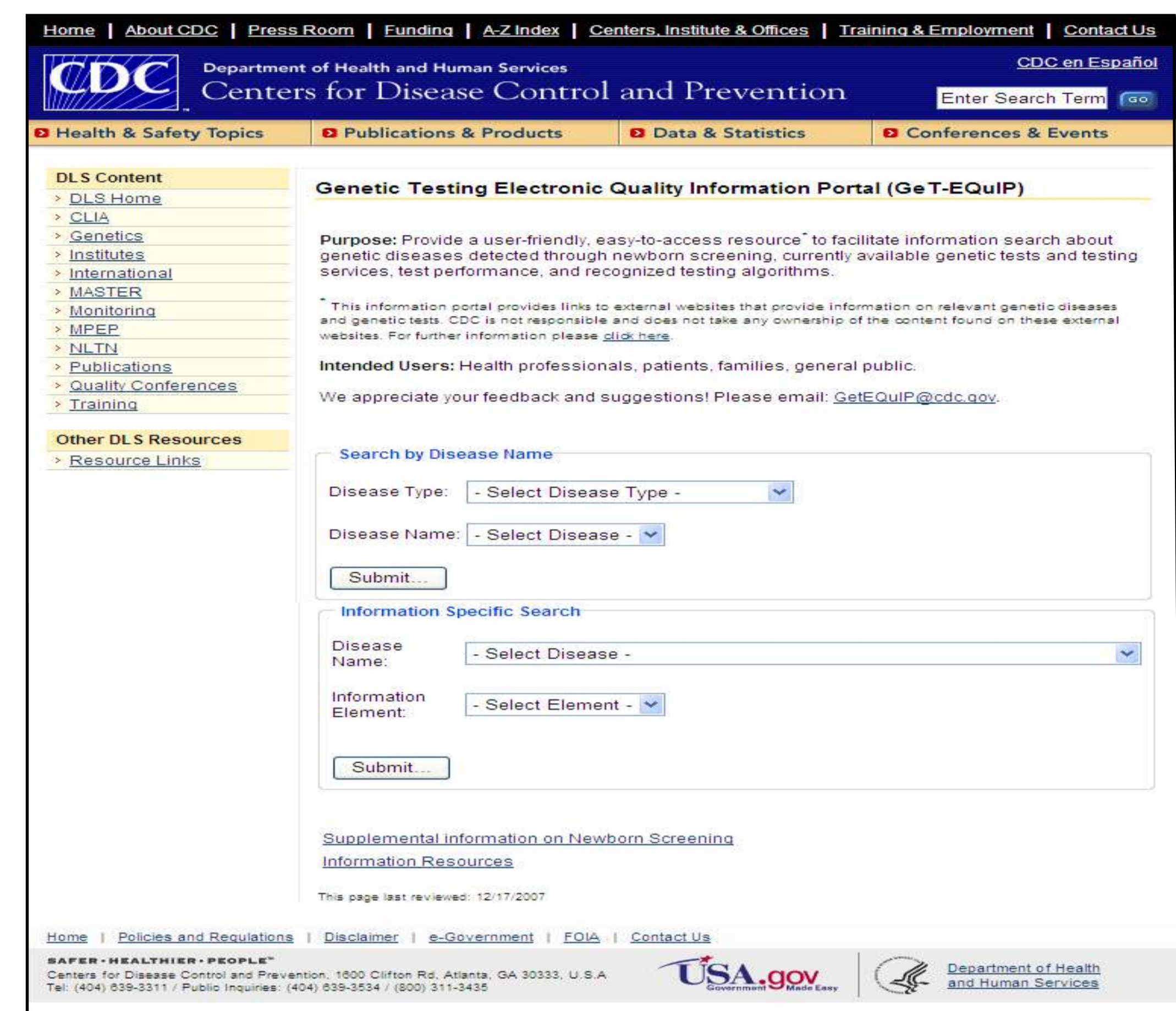


Figure 1. The GeT-EQUIP homepage

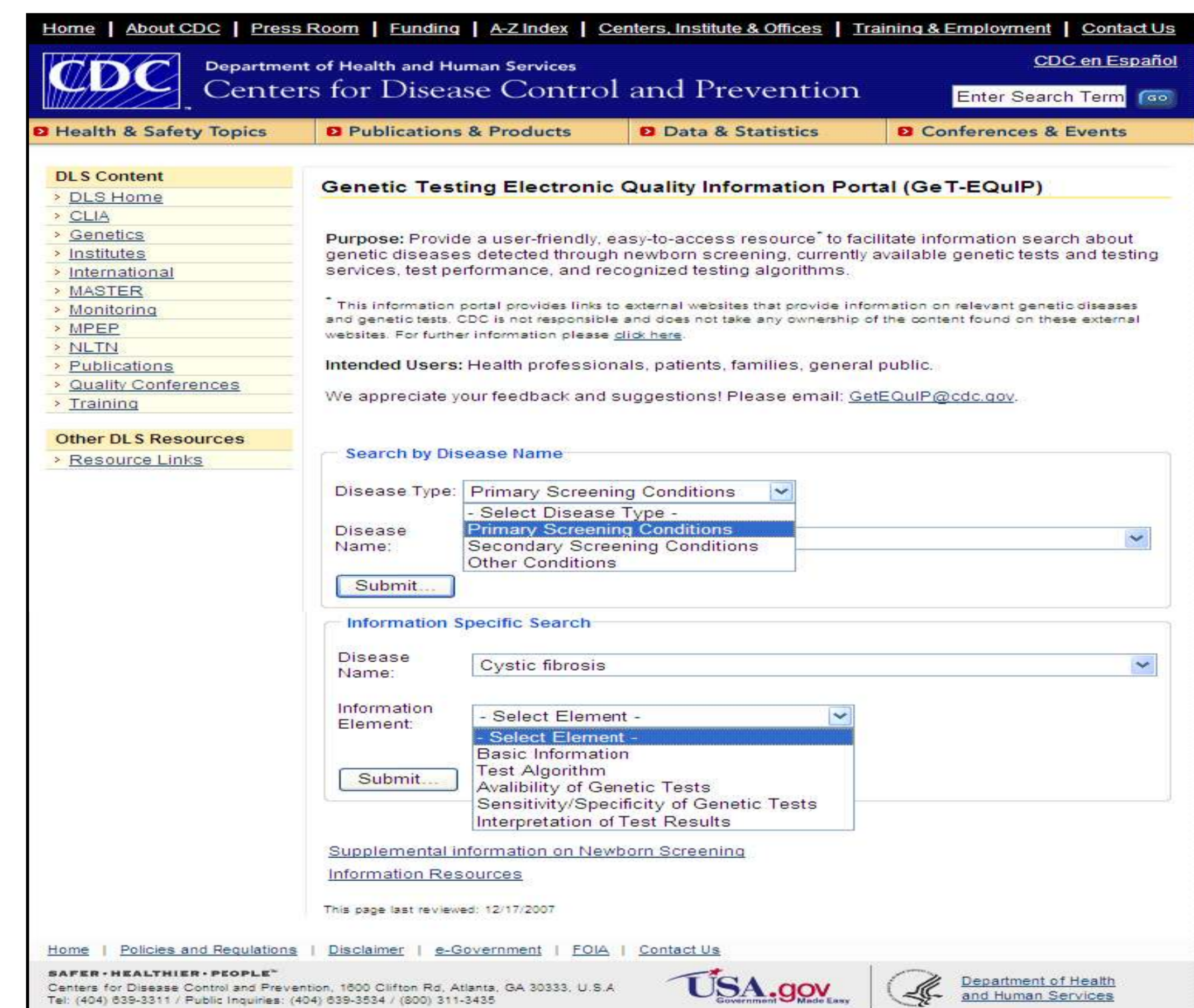


Figure 2. A search can be performed by disease category or using the advanced search by information element.

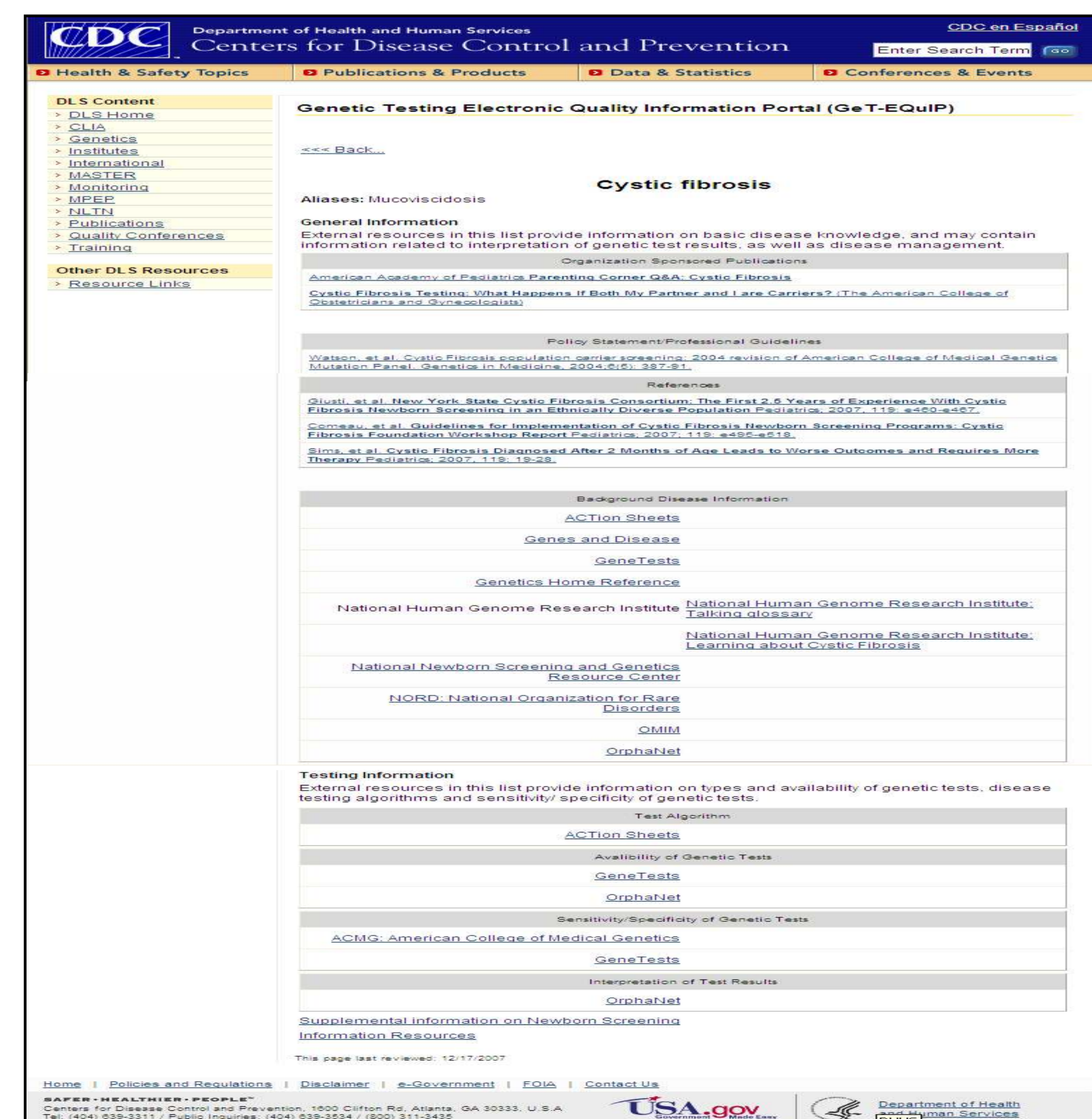


Figure 3. Search results page