Confused by genetic tests? NIH's new online tool may help

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An online tool launched today by the National Institutes of Health will make it easier to navigate the rapidly changing landscape of genetic tests. The free resource, called the Genetic Testing Registry (GTR), is available at www.ncbi.nlm.nih.gov/gtr/ .

"I'm delighted that NIH has created this powerful, new tool. It is a tremendous resource for all who are struggling to make sense of the complex world of genetic testing," said NIH Director Francis S. Collins, M.D., Ph.D., who unveiled GTR at NIH's observance of international Rare Disease Day. "This registry will help a lot of people — from health care professionals looking for answers to their patients' diseases to researchers seeking to identify gaps in scientific knowledge."

Genetic tests currently exist for about 2,500 diseases, and the field continues to grow at an astonishing rate. To keep pace, GTR will be updated frequently, using data voluntarily submitted by genetic test providers. Such information will include the purpose of each genetic test and its limitations; the name and location of the test provider; whether it is a clinical or research test; what methods are used; and what is measured. GTR will contain no confidential information about people who receive genetic tests or individual test results.

Genetic tests that the Food and Drug Administration has cleared or approved as safe and effective are identified in the GTR. However, most laboratory developed tests currently do not require FDA premarket review. Genetic test providers will be solely responsible for the content

and quality of the data they submit to GTR. NIH will not verify the content, but will require submitters to agree to a code of conduct that stipulates that the information they provide is accurate and updated on an annual basis. If submitters do not adhere to this code, NIH can take action, including requiring submitters to correct any inaccuracies or to remove such information from GTR.

In addition to basic facts, GTR will offer detailed information on analytic validity, which assesses how accurately and reliably the test measures the genetic target; clinical validity, which assesses how consistently and accurately the test detects or predicts the outcome of interest; and information relating to the test's clinical utility, or how likely the test is to improve patient outcomes.

"Our new registry features a versatile search interface that allows users to search by tests, conditions, genes, genetic mutations and laboratories," said Wendy Rubinstein, M.D., Ph.D., director of GTR. "What's more, we designed this tool to serve as a portal to other medical genetics information, with context-specific links to practice guidelines and a variety of genetic, scientific and literature resources available through the National Library of Medicine at NIH."

GTR is built upon data pulled from the laboratory directory of GeneTests, a pioneering NIH-funded resource that will be phased out over the coming year. GTR is designed to contain more detailed information than its predecessor, as well as to encompass a much broader range of testing approaches, such as complex tests for genetic variations associated with common diseases and with differing responses to drugs. GeneReviews, which is the section of GeneTests that contains peer-reviewed, clinical descriptions of more than 500 conditions, is also now available through GTR.

The GTR database was developed by the National Center for

Biotechnology Information (NCBI), part of NIH's National Library of Medicine, under the oversight of the NIH Office of the Director and with extensive input from researchers, testing labs, health care providers, patients and other stakeholders. To view video tutorials on how to use GTR, go to www.youtube.com/playlist?list=PL1C4A2AFF811F6F0B .

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