

From: Bob Wildin [mailto:bwildin@earthlink.net]
Sent: Wednesday, June 02, 2010 10:10 PM
To: Genetic Testing Registry (NIH/OD/OSP)
Subject: Notice Number: NOT-OD-10-101 NIH Plan...GTR

Dear GTR,

I'm responding to the above RFI.

As an independent, non-academic Clinical Geneticist who is the sole Clinical Geneticist provider for two large community hospital systems in two western states, I have a unique perspective on the practical use of clinical genetic tests, as well as the processes involved in assessing tests and getting them approved and performed. In addition, I was previously the Medical Director of a Clinical Molecular Diagnostic Laboratory, so I have that perspective as well.

May I begin by saying how incredibly valuable the GeneTests/GeneReviews resource is. Both the GeneReviews and GeneTests sections are absolutely essential for my work. I can't emphasize that enough.

In contrast, I have found myself with some legitimate concerns due to problems with NIH's implementation of these sites (upon transfer from UW), as well as a culture of apparent disregard for the impact of such implementation changes on the clinical end-user's experience. I have also experienced what I would describe as "lip-service" from NIH when my concerns are directly voiced. Finally, I've observed that NIH's primary interest is in serving the research community first (reference those implementation changes), the gene testing industry second, and the clinical community (which includes physicians and patients) last. For example, while NIH has consumer-oriented information websites for genetics patients, there are at least four different sites, in different institutes, with different information formats, and limited logical inter-linking. I can guess how this situation arose, but it's fundamentally an inward-facing solution to an outward-facing problem. These and similar problems make obtaining useful information a real challenge for clinicians, who, unlike researchers, are on a tight and limited schedule.

For these reasons, I'm gravely concerned that the "elimination" of GeneTests and it's replacement by something grown solely at NIH will turn out to be a net loss, and a serious blow, for those of us who need and use an accessible, reliable, resource that is efficiently designed, end user-tested, and un-cluttered by clinically irrelevant links. NIH must decide who the target beneficiary is: patients and clinicians, or researchers. Trying to please everybody will result in nobody being pleased. As I say, information search must start with "agreement of purpose." Without that, you have Google search. So before you start this project. Please agree on a purpose, not four or five!

I must emphasize that I don't completely understand why GeneTests, an excellent resource, is being eliminated, as opposed to being upgraded. Additionally, I'm really unclear on why NIH believes the testing providers will actually voluntarily provide the extensive (and often changing) additional information you list.

Having said that, the issue of documenting validity and usefulness of the tests is one that is yet unsolved. I and my assistants spend a significant portion of our time (non-billable!!!!) dealing with public and private payers who reject or deny claims because they won't themselves take the time to understand how these tests can actually save them money and improve the health of the patient. It seems clear that they have created arbitrary blanket policies that deny standard-of-care testing in order to avoid having to learn about this area, to the detriment of their patients and themselves. I have some doubt that having a resource such as the GTR can change that, as it is more a problem of (false) cost-containment priority, lack of appropriately trained personnel, and

attitude than it is a lack of data. However, having a truly authoritative site will make my job a bit easier as I can use that as an “educational tool” for my interactions with payers, without having to resort to PubMed and articles in journals I don’t have access to.

The GTR information I’ve seen is not clear on how the “authoritativeness” of the information will be established. Is all the supporting data self-reported by the testing laboratories, or will there be an FDA-like body that will pass judgment based on published or submitted clinical data? Will every lab upload their own information regarding usefulness or will that data be distilled so that the test itself, rather than each lab’s implementation, is supported in a central location? If labs are the source of the information, there will be bias. Your goal will be undermined by a tendency to provide a false sense of authoritativeness for testing.

I’m glad to see you’ve included cost in your list of desired data. I now fairly routinely “shop around” to find the most cost-effective, qualified lab to perform these often expensive tests. I often save my patients (or their insurers, or the hospital that ends up paying for it) many hundreds of dollars, so I think it’s worth my time. Making that easier would be a real boon.

You may be aware that some labs refuse to post some or all of their prices on the web. Some have complex tiered pricing systems for different payer situations that they will only reveal after a lengthy telephone call. That’s unacceptable. In the interest of fostering legitimate competition, I believe the test price, turnaround time, and an exact description of the test’s coverage and methods, should be REQUIRED (and binding). A generic link to the lab’s web page is UNACCEPTABLE, although an external data link that retrieves real time price from the lab’s pricing database would be acceptable. It ought to be as easy to compare gene test prices, products, and methods, as it is to shop for airplane flights at TripAdvisor.

Some testing labs offer tests where the gene or the test are patented. That information should be disclosed on the website, e.g., “gene/process patent holder” or “gene/process patent licensed”, and link to the USPTO patent record.

Because testing labs often offer many tests which they document and track in their own database systems, you will need to offer a “bulk upload” format, in addition to a web browser manual entry format for laboratory data entry. This MUST BE simple, thoughtful, and designed for efficient use by the end user, unlike NIH’s entrez SOAP implementation.

A significant differentiator for laboratories, from the end-user perspective, is the information contained in the test result report. Some provide basic, stripped down reports designed for the primary care clinician that are inadequate for Clinical Genetics professionals to adequately judge what was actually done. Others are compiled largely of lengthy stock text written by legal, rather than medical experts. Others contain misleading, self-serving, or inaccurate facts or recommendations. The usability of both formats is, in my opinion, seriously compromised. I think the laboratories should be required to submit two sample reports (one “normal” and one “abnormal”) that the GTR user can view (e.g., as PDF’s), to judge whether the report they can expect to see is aimed appropriately for their uses. This is not a trivial issue. I’ve stopped using well known, sophisticated, well-respected labs because they don’t document on the report how they came to the conclusion that a complex set of tests was “normal.” But I had to experience the pain of dealing with those reports before I could avoid them.

A huge burden in ordering gene tests is the filling out of a forest of forms. Each laboratory has “their” forms which must be “fished” out of their websites. Some are easier than others to find. It would be great if a) there were an at least partially standardized form (think the standardized NIH Grant/Biosketch forms), and b) the test’s specific form (with the test in question already

checkmarked/defined) was immediately accessible via a link in the test record in GTR. NO FISHING! The same principle applies to “clinical data” form and “consent” form required by many testing laboratories. The filling of these forms takes an enormous amount of non-billable time, and the technology exists to make this go much faster. Please think how you can facilitate this.

The other aspect of obtaining a test on a patient is obtaining payer pre-authorization. This is often an arduous process. The testing lab must supply – again, in a standardized format directly in the GTR record – the information required by insurers/payers for the preauthorization request (cpt codes etc.). Ideally, that information would be supplied with a complete description of the test, and information that supports its validity and usefulness, in a printable format that can be submitted to insurers. NO CUSTOM LETTERS BY ME!

I’m sure I have many other suggestions, but dinner is on the table. Please contact me for clarifications, or (fee-based – I’m an independent businessman) further assistance.

Best wishes,

Bob Wildin, M.D.
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Portland, OR, and Boise, ID
Also: Epintell, LLC (www.epintell.com)

From: Bob Wildin [mailto:bwildin@earthlink.net]
Sent: Thursday, June 03, 2010 11:24 AM
To: Genetic Testing Registry (NIH/OD/OSP)
Subject: GTR RFI

Dear GTR,
I wanted to add one more suggestion for the GTR. The submitters of validity and usefulness information must provide a conflict-of-interest disclosure. If end-users are expected to find this information credible, the other interests of the authoritative parties that could compromise credibility must be transparent.
Thanks,
-- Bob

Bob Wildin, M.D.
ABMG Clinical Geneticist

From: Wildin, Bob [mailto:ROBERT.WILDIN@providence.org]
Sent: Tuesday, June 15, 2010 2:36 PM
To: Genetic Testing Registry (NIH/OD/OSP)
Cc: 'bwildin@earthlink.net'
Subject: GTR features

Dear GTR,
I provide clinical genetics services to multiple organizations and I’ve already provided comments for the RFI using my direct e-mail address: bwildin@earthlink.net.
An additional issue has been raised at the institution below, but one that affects us everywhere. Labs should be required to supply the minimal sample requirements for Adults AND PEDIATRIC AND NEONATAL PATIENTS. Due to the layering of laboratory services, it often

takes 3 to 5 phone calls to find out that a test documented as requiring 10 ml of blood can actually use 2 ml. This is unacceptable. This is a huge difference for neonates, especially premature and low birth weight infants.

Please require labs to specify their minimum sample volume requirements for infants, so that that information can be used directly without a lot of additional human intervention.

-- *Bob*

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