

**From:** Dr. Bob Wildin [mailto:bwildin@earthlink.net]  
**Sent:** Wednesday, July 27, 2011 6:08 PM  
**To:** Genetic Testing Registry (NIH/OD/OSP)  
**Subject:** Re: Request for Comments/Genetic Testing Registry

Hi, Dr. Patterson,

Here are my comments on first readthrough.

1) The federal register entry states: "...however, there is no centralized public resource that provides information about the availability and scientific basis of these tests." This is misleading. While GeneTests does not provide the scientific basis for the tests, it is a "centralized public resource that provides information about the availability...of these tests." I'm sad to see such a misleading statement in the Federal Register.

2) I'm glad to see some of my prior suggestions were incorporated.

3) No mention is made of the bulk upload option except in passing in the Federal Register document. I don't think respondents can accurately judge the manual web-based input form without understanding the alternative approach, which they could probably integrate and semi-automate with their own Laboratory Information System. This is a crucial issue, as a poorly designed bulk upload system will change the equation for all users. As such, respondents must judge only the manual system, which, while fairly well designed, is definitely burdensome. The extra work required to manually enter the information must be paid for in higher test prices, while we're trying to limit the costs of advanced health care. Unlike NIH where there is usually a foreign fellow to do the tedious work, companies don't have that much extra staff.

4) the multiplier of 1.2 for the volume of tests expected to be entered into the new system is woefully inadequate. As the result of NIH-funded research (Yeah!) the genetic causes of many more disorders, rare and common, are being identified, and we are about to enter an even more abruptly exponential phase in expansion of clinical testing availability. Furthermore, complex testing, e.g. bundling test approaches, are becoming increasingly common (and often appropriately so), and so these will add to the menu of available tests although they don't actually test for more disorders. I would say a multiplier of 3, at minimum, is easily expected.

5) the difference between "URL for ordering test" and "URL for the Test" right under it is not explained, and is not intuitively obvious.

6) I found it difficult to hone in on the required fields (search for "\*" didn't work in my hands). A separate listing of those required and non-required fields might be helpful for those trying to evaluate this data entry path.

7) unless I missed it, the information does not have information that allows the clinician to:

a. compare test offerings - e.g. bundled gene panels for a specific disease; i.e. does the entered data clearly spell out which genes and which gene regions are examined (for sequencing tests)?

b. the cost of the test.

Both of these are routine elements of the process clinicians go through to identify a laboratory to do a properly targeted and cost-efficient service.

I don't at this time represent any laboratory, but I work in the clinical realm and am a substantial customer for these tests. I hope my comments will help you build a practical service that improves on what currently exists in GeneTests, while not scaring away laboratories. Based on my discussions with colleagues at the March clinical genetics meetings, I think that is a significant risk. If that happens, and if GeneTests goes away, I'll be very sad as it will negatively impact my ability to take care of my patients (who, by the way, are largely on publicly-paid healthcare programs).

Most sincerely,  
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