In Defense of Commercial Laboratories

To the Editor:

In a letter entitled "A call for mutations",[1] the authors writing for the Human Genome Variation Society (HGVS) make repeated unsubstantiated allegations regarding commercial laboratories performing molecular testing. They assert, without documentation, that commercial laboratories are not interested in advancing knowledge and are making no effort to systematically collect data nor report data in peer reviewed literature and publicly available databases. Recently, at the business meeting of the American College of Medical Genetics, the discussion turned into a commercial laboratory bashing event. We are all physicians and scientists, and I feel that this Journal should require a modicum of proof before authors are allowed to criticize commercial laboratories based on supposition and rumor.

As the director of a commercial laboratory, I feel I can no longer sit idly by while my laboratory and those of my colleagues are repeatedly criticized without any facts being offered into evidence. For the record, our laboratory has published several articles in this Journal and others reporting allele frequencies,[2–5] rare mutations,[6] and collaborated with academic groups to describe novel diseases.[7] Whenever appropriate we submit new and rare mutations to the appropriate databases and are always willing to share our data with clinical and scientific colleagues in order to provide better care. We have already submitted 8 novel mutations to the Cystic Fibrosis Database;[8] del promoter and exon 1, del promoter and ex 1, ex 2, del ex 2, 3, del ex 4, 5, 6a, del ex 17a, 17b, del ex 17a, 17b, 18, del ex 22, 23, del ex 22, 23, 24 and have manuscripts under consideration containing several novel mutations in beta globin, MECP2, in addition to CFTR. We are completely committed to the advancement of knowledge.

The suggestion that we must be coerced into participating in the authors' programs by regulatory agencies is particularly odious. The authors suggest, "it would be even more desirable if such submission was part of activities required by accreditation." Ironically, if such a policy were to be put in place by the College of American Pathology or the Clinical Laboratory Improvement Act, academic laboratories would face the same coercions. I must wonder how and why this particular organization, HGVS, has never contacted our laboratory to inform us of their databases and to request our participation, but rather resort to an open letter with a condescending and accusatory tone. And why should HGVS, as opposed to any number of other public databases, be the beneficiary of this coercion? In addition, the authors neglect to specify what mechanisms are in place to ensure compliance with the Health Insurance Portability and Accounting Act, and what provision is made to ensure that the submitted information is not used by HGVS for their own ends, such as publication and/or grant acquisition.

Contrary to popular opinion, physicians and scientists who have chosen to work in commercial laboratories have neither sold their souls to the devil nor abandoned their ethical and moral principles to provide the best possible care to patients. Because of economies of scale and automation we are able to perform high throughput testing at a lower cost while maintaining the highest possible quality standards.[9] We provide an important resource to the clinical, pharmaceutical, and academic communities.

Commercial laboratories often must pick up the slack when an academic investigator declines to offer clinical or prenatal testing for patients whose mutations they discovered. Any investigator or clinician who has made the effort to contact me or anyone in my laboratory will find a willing collaborator who will make every effort to provide information that will further knowledge and/or benefit an individual patient.

The adage; *one collects more flies with honey than with flypaper*, applies here. If the authors' goal was to encourage submission of data into their program, why did they find it necessary to insult the integrity of commercial laboratories while making their plea? I can only assume that they had prejudged us in the commercial sector as being unwilling or unable to participate. So I say to the authors and the rest of the "academic community"; rather than railing against the rain and assuming the worst, why don't you try working with us in the commercial sector as colleagues and collaborators rather than instruments of the devil? I guarantee you will be pleasantly surprised.

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