Reply to Dr. Strom regarding "A call for mutations"

To the Editor:

In our recent letter,[1] we made the following suggestions:

- A suggestion that mutations were being lost in commercial molecular diagnostic laboratories and others.
- A suggestion that submission of identified DNA variants to mutation databases could be part of accreditation for molecular diagnostic laboratories.
- A suggestion on how the activities of the Human Genome Variation Society (HGVS) could help in the collection and dissemination of these variants.

In his response to our letter,[2] Dr. Strom made several statements that we must respond to.

We made the suggestion that DNA variants were lost in commercial diagnostic laboratories' records based on an unpublished survey of three major molecular diagnostic laboratories who said they had no time to publish or submit to databases. (We have since heard of another major laboratory who does submit to the relevant database.) We have not made statements that "commercial laboratories are not interested in advancing knowledge," or "making no effort to systematically collect data nor report data." Our point was that molecular diagnostic laboratories are busy enough and that there needs to be a system that does not engender much additional work for the publishing of these variants. As a society dedicated to the collection and dissemination of DNA variation, we want to facilitate the availability of this valuable resource for other laboratories.

There was no intention to continue "commercial laboratory bashing," which Dr. Strom claims he has been subjected to. We, as well as clinicians, patients, and researchers, will be delighted to hear of this company's effort to put data into the public arena.

We admit that there needs to be a systematic query of all types of diagnostic laboratories to measure how much data remain unsubmitted to the public system.

Second, we believe that diagnostic laboratories, researchers, and clinicians would benefit from obligatory submission of these variants, because a complete set of mutations for a given gene would be extremely valuable to health care professionals, as well as the research community. We make no apology for making one suggestion of how to have mutations migrate to the public sector. Discussion, logic, and need will decide how best to achieve this goal.

Third, the HGVS does not "own" any databases. Perhaps Dr. Strom is unfamiliar with the purpose of the HGVS; as stated in the bylaws, "The purposes of the Organization shall be, but are not limited to, the following:

- To promote interdisciplinary research on the effects of variations in the DNA sequence including mutations and polymorphisms;
- To encourage the exchange and dissemination of information regarding all aspects of DNA sequence variation, including but not limited to the production of a database for the dissemination of this information;
- To encourage and facilitate collaboration between investigators interested in the broad topic of DNA variation;
- To foster a journal which has as its principal object the publication of manuscripts reporting on mutation research as means to disseminate new information and to promote collaboration;
- To organize a meeting of the membership of the Organization at least once a year.
- To provide consultation to the public on relevant issues of national and international importance."

Thus, submitted information cannot be "used by HGVS for their own ends" except to promote mutation collection to allow better and more efficient genetic health care.

We believe there was nothing in our letter that "insult(ed) the integrity of commercial laboratories." Clearly the activities of Quest Laboratories are exemplary in the area and should be congratulated. It is unfortunate Dr. Strom misunderstood our intention.

Richard Cotton, PhD, DSc
Arleen D. Auerbach, PhD
William S. Oetting, MS, PhD
for the Human Genome Variation Society

References

1. Cotton RGH, Auerbach AD, Oetting WS. A call for mutations. *Genet Med* 2005;7:370.
2. Strom C. In defense of commercial laboratories. *Genet Med* 2005;7:590.