

From: Myriad Genetics [mailto:[noreply@myriad.com](mailto:noreply@myriad.com)]

Sent: Thursday, June 12, 2014 8:01 AM

To:

Subject: Public Variant Databases: Important Considerations



Dear Colleague,

Variant classification methodology, VUS rates, and variant classification data sharing have been prominent topics in hereditary cancer testing over the past year as the marketplace for testing has expanded. There are a spectrum of opinions and practices among laboratories and providers related to variant classification and data sharing, and we are writing to provide you with Myriad's perspective on public variant databases.

Myriad believes that variant classification databases used for clinical purposes should be subjected to quality assurance oversight. Medical management decisions made on the basis of variant interpretations are crucial to the safety and well-being of patients, and we believe the use of unregulated databases poses risks to patients. At this time, there are no well-regulated public variant databases that meet the accuracy standards necessary to provide good patient care.

The following short video by one of Myriad's Laboratory Directors, Dr. Karla Bowles, highlights some of the concerns regarding use of public databases for clinical care:



Please watch the video and consider the following:

- Myriad shared data with the Breast Information Core (BIC) database until 2007. Myriad decided to stop contributing to the BIC based upon instances of reclassified variants not being updated in the database and the knowledge that the lack of timely updates was having a negative impact on patient care.
- Current public variant databases were designed for research purposes and are not maintained or regulated in a way that provides appropriate patient care. They lack proper quality control to ensure that the information is entered accurately and that any changes in classification are updated in a timely fashion. There is currently no mechanism to alert providers or patients when a reclassification occurs so patients may be over or undermanaged unnecessarily.
- Both the NIH and ACMG have statements regarding the inaccuracies and potential harms of public databases.
- Myriad recently evaluated and compared classifications of over 2,000 variants in BRCA1 and BRCA2 among 5 public databases with the Myriad database. Results show a wide disparity in classification among and within databases that can lead to patient mismanagement if public databases are relied upon for clinical care. Myriad will be sharing this information in an upcoming publication.
- The ultimate consequence of inaccurate and outdated information in public variant databases is negative outcomes for patient care and safety related to false negative, false positive, and misclassified VUS results.

Myriad is dedicated to furthering genetic science and has engaged in hundreds of scientific collaborations with many top institutions to help advance medical and scientific knowledge. For more information please visit: <https://www.myriad.com/responsibility/research-collaborations/>.

Myriad recently announced its participation in a research collaboration with leading cancer institutes to advance the understanding of the risks associated with multiple cancer genes that are being analyzed through next generation sequencing technology. Myriad will be contributing thousands of mutations to the

Prospective Registry of Multiplex Testing (PROMPT) collaboration in addition to facilitating additional studies to characterize the significance of mutations and providing scientific expertise. For more information on Myriad's participation in the PROMPT study please view the press release [here](#).

For additional information on Myriad and resources for your practice, please visit: [www.MyriadPro.com](http://www.MyriadPro.com).

---

Myriad Genetics 320 Wakara Way Salt Lake City, UT 84108 United States

You received this email because you are subscribed to Marketing Information from Myriad Genetics.

Update your [email preferences](#) to choose the types of emails you receive.

[Unsubscribe from all future emails](#)

Powered by [HubSpot](#)