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DEPARTMENT OF COMMERCE

National Institute of Standards and Technology

Genome in a Bottle Consortium – Work Plan Review Workshop

AGENCY: National Institute of Standards & Technology (NIST), Commerce.

ACTION: Notice of public workshop.

SUMMARY: NIST announces the Genome in a Bottle Consortium meeting to be held on Thursday and Friday, August 16 and 17, 2012. The Genome in a Bottle Consortium is planning to develop the reference materials, reference methods, and reference data needed to assess confidence in human whole genome variant calls. A principal motivation for this consortium is to enable performance assessment of sequencing and science-based regulatory oversight of clinical sequencing. The purpose of this meeting is to get broad input from stakeholders about the draft consortium work plan, broadly solicit consortium membership from interested stakeholders, and invite members to participate in work plan implementation.

DATES: The Genome in a Bottle Consortium meeting will be held on Thursday and Friday, August 16 and 17, 2012. Attendees must register by 5:00 PM Eastern time on Thursday, August 9, 2012.

ADDRESSES: The meeting will be held at the National Institute of Standards and Technology, 100 Bureau Drive, Gaithersburg, MD 20899 in Room C103-C106, Building 215. Please note admittance instructions under the SUPPLEMENTARY INFORMATION section of this notice.

FOR FURTHER INFORMATION CONTACT: For further information contact Justin Zook by email at jzook@nist.gov or by phone at (301) 975-4133 or Marc Salit by email at salit@nist.gov or by phone at (301) 975-3646. To register, go to: https://www-s.nist.gov/CRS/

SUPPLEMENTARY INFORMATION: Clinical application of ultra high throughput sequencing (UHTS) for hereditary genetic diseases and oncology is rapidly growing. At present, there are no widely accepted genomic standards or quantitative performance metrics for confidence in variant calling. These standards and quantitative performance metrics are needed to achieve the confidence in measurement results expected for sound, reproducible research and regulated applications in the clinic. On April 13, 2012, NIST convened the workshop "Genome in a Bottle" to initiate a consortium to develop the reference materials, reference methods, and reference data needed to assess confidence in human whole genome variant calls. A principal motivation for this consortium is to enable science-based regulatory oversight of clinical sequencing.

At present, we expect the consortium to have four working groups with the following responsibilities:

(1) Reference Material (RM) Selection and Design: select appropriate cell lines for whole genome RMs and design synthetic DNA constructs that could be spiked-in to samples.

(2) Measurements for Reference Material Characterization: design and carry out experiments to characterize the RMs using multiple sequencing methods, other methods, and validation of selected variants using orthogonal technologies.

(3) Bioinformatics, Data Integration, and Data Representation: develop methods to analyze and integrate the data for each RM, as well as select appropriate formats to represent the data.

(4) Performance Metrics and Figures of Merit: develop useful performance metrics and figures of merit that can be obtained through measurement of the RMs.

The products of these working groups will be a set of well-characterized whole genome and synthetic DNA RMs along with the methods (documentary standards) and reference data necessary for use of the RMs. These products will be designed to help enable translation of whole genome sequencing to regulated clinical applications.

There is no cost for participating in the consortium. No proprietary information will be shared as part of the consortium, and all research results will be in the public domain.

All visitors to the NIST site are required to pre-register to be admitted and have appropriate government-issued photo ID to gain entry to NIST. Anyone wishing to attend this meeting must register at https://www-s.nist.gov/CRS/ by 5:00 PM Eastern time on Thursday, August 9, 2012, in order to attend.

Dated: July 18, 2012

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