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DEPARTMENT OF COMMERCE

National Institute of Standards and Technology

Genome in a Bottle Consortium – Progress and Planning 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 14 and 15, 2014. The Genome in a Bottle Consortium is developing 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 update participants about progress of the consortium work, continue to get broad input from individual stakeholders to update or refine the consortium work plan, continue to broadly solicit consortium membership from interested stakeholders, and invite members to participate in work plan implementation. Topics of discussion at this meeting will include examples of laboratories using the pilot candidate NIST Reference Material, progress on the next set of NIST Reference Materials, structural variants, and potential Reference Materials for cancer genomics.

DATES: The Genome in a Bottle Consortium meeting will be held on Thursday, August 14, 2014 from 9:00 AM to 5:30 PM Eastern Time and Friday, August 15, 2014 from 9:00 AM to 12:45 PM Eastern Time. Attendees must register by 5:00 PM Eastern Time on Monday, August 11, 2014.

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 (650) 350-2338. To register, go to: https://www-s.nist.gov/CRS/conf_disclosure.cfm?conf_id=7372

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 (www.genomeinabottle.org). On August 16-17, 2012, NIST hosted the first large public meeting of the Genome in a Bottle Consortium, with about 100 participants from government, academic, and industry. This meeting was announced in the Federal Register (77 FR 43237) on July 24, 2012. A principal motivation for this consortium is to enable science-based regulatory oversight of clinical sequencing.

At the August 2012 meeting, the consortium established work plans for four technical working groups with the following responsibilities:

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

(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 technical 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. The consortium meets in workshops two times per year, in January at Stanford University in Palo Alto, CA, and in August at the National Institute of Standards and Technology in Gaithersburg, MD. At these workshops, including the last meeting at NIST in August 2013, participants in the consortium have discussed progress developing well-characterized genomes for NIST Reference Materials and planned future experiments and analysis of these genomes (see https://federalregister.gov/a/2012-18064 and https://federalregister.gov/a/2013-18934 for past workshops at NIST). The August 2013 meeting, which included meetings of each of the four working groups, was announced in the Federal Register (78 FR 47674) on August 6, 2013, and the meeting is summarized at http://genomeinabottle.org/blog-entry/giab-workshop-summary-august-15-16-2013.

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 present appropriate government-issued photo ID to gain entry to NIST. Anyone wishing to attend this meeting must pre-register at https://www-s.nist.gov/CRS/conf_disclosure.cfm?conf_id=7372 by 5:00 PM Eastern Time on Monday, August 11, 2014, in order to attend.

Dated: August 5, 2014.

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