



Collaboration news: Medical Device Innovation Consortium Launches Initiative to Improve Accuracy of Next Generation Sequencing-Based Cancer Diagnostics

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Initiative to develop, manufacture, and validate publicly available somatic reference samples and create public genomic datasets with the potential to be used by sponsors and regulators

ARLINGTON, Va.--([BUSINESS WIRE](#))--The Medical Device Innovation Consortium ([MDIC](#)) formally launched its Somatic Reference Samples (SRS) Initiative with a pilot project to improve the validation and regulatory review process for cancer diagnostics based on next generation sequencing (NGS).

MDIC will lead a collaboration with the U.S. Food and Drug Administration (FDA), the National Institute of Standards and Technology (NIST), National Institutes of Health (NIH) and industry stakeholders to manufacture, validate, and distribute SRSs to simplify and support validation of NGS-based cancer diagnostics. The initiative also includes the goal to create a publicly available global genomic data resource library of datasets with the potential to be used by sponsors and regulators.

"NGS is a powerful technology enabling breakthroughs in diagnostics and ultimately therapeutics. These diagnostic tests need to be validated for accurate clinical use, and reference samples are essential to the validation process. But well-characterized and widely accepted reference materials do not exist for NGS-based diagnostics, complicating the development and validation process," said Andrew Fish, President and CEO, MDIC. "Through the MDIC SRS Initiative, we are developing reference samples and data sets that can be used globally by test developers and regulators to bring more consistency to NGS-based cancer diagnostic development, increasing the confidence and accuracy of these tests, which will ultimately lead to more accuracy in diagnosis and treatment for patients."

"There is a need for appropriately consented, highly characterized, and broadly available reference materials that may improve the accuracy, reliability, and transparency of NGS-based oncology tests and support the generation of validation data for use in regulatory submissions. The reference samples and datasets being created by the MDIC Somatic Reference Samples Initiative can help fulfill this need," said Wendy Rubinstein, MD, PhD, Director, Personalized Medicine, Center for Devices and Radiological Health, U.S. Food and Drug Administration.

[Horizon Discovery](#), a PerkinElmer company, is conducting the development and manufacture of these reference samples. The goal of this pilot project is to individually engineer 10 gene variants clinically associated with cancer into a highly characterized human cell line – GM24385 (PGP/GIAB) – using Clustered Regularly Interspaced Short Palindromic Repeats (CRISPR) technology resulting in ten edited human cell lines, each containing one gene variant clinically associated with one or more specific cancers. The cell lines, containing confirmed sequences, will be combined and available in FFPE format. The fully characterized reference samples will be commercially available to the end users from Horizon and the characterization data will be accessible through public databases, including [precisionFDA](#), an FDA-sponsored site for data information, sequencing, and bioinformatics.

NIST's involvement in this work is to foster understanding.* "As our collaboration validates the MDIC SRS samples, we will also learn and build best practices to improve the efficiency and sustainability of reference sample validation and dataset generation in general. This project is a major step in the development and optimization of new sequencing technologies and the translation of DNA sequencing to clinical applications for diagnosing and treating cancer," said Justin Zook, PhD, Co-Leader, Biomarker and Genomic Sciences Group, National Institute of Standards and Technology.

MDIC's SRS project began as a working group in 2018, representing more than forty stakeholders with a mission to address the gap in reference material for NGS-based diagnostic tests. The initial output of this working group, [MDIC's SRS Landscape Analysis](#), published in 2019, was a comprehensive catalog of existing clinically relevant cancer variants and available reference samples. Based on the unmet needs discovered through the SRS Landscape Analysis, the SRS Initiative has prioritized a subset of ten variants to be engineered into reference samples for the pilot project. In addition to the utility of these samples in development, validation, and regulatory review, these samples potentially have value in reimbursement decisions, post-market monitoring and informing the path forward for additional SRS.

The work in the SRS Initiative is funded in part by the Gordon and Betty Moore Foundation (Grant GBMF), the National Philanthropic Trust, Illumina, and Quidel.

** Affiliation with a commercial product does not imply recommendation or endorsement by the National Institute of Standards and Technology, nor does it imply that the materials or equipment identified are necessarily the best available for the purpose.*

About MDIC

Founded in 2012, the Medical Device Innovation Consortium (MDIC) is the first public-private partnership created with the sole objective of advancing medical device regulatory science throughout the total product life cycle. MDIC's mission is to transform health care into human care. Collaborating with our partners to advance science, we enable transformational medical technology to shape the world we want to live in and make that world possible by shortening the path from innovation to safety to access. For more information, visit [mdic.org](#).

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