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June News and Product Briefs By Aaron Kroll

News Briefs

The **Global Alliance for Genomics and Health**(GA4GH), an international coalition dedicated to maximizing the potential of genomic medicine, marked its second anniversary this month. GA4GH now includes over 320 organizations across 32 countries, while more than 700 individuals around the globe are actively developing dozens of tools, methods, and approaches to facilitate effective, responsible data sharing. GA4GH Working Groups have developed a regulatory framework to guide the responsible sharing of genomic and health-related data; a GA4GH Genomics API to enable the interoperable exchange of data in DNA sequence reads; and a security infrastructure that recommends policy and technology options for the ecosystem, among other achievements. [Press release](#)

The **National Cancer Institute** will enroll up to 1,000 patients in a new national oncology clinical trial called NCI-MATCH (Molecular Analysis for Therapy Choice), using genetic sequence information obtained from **Thermo Fisher Scientific**'s targeted next-generation sequencing (NGS) technology. As many as 3,000 tumor samples will be sequenced for this program across multiple academic centers. The targeted sequencing assay will include 143 genes selected using the OncoPrint Knowledgebase, the world's largest collection of oncology data. The panel design also enables simultaneous sequencing of a wide range of genetic alterations, including single nucleotide variants (SNV), small insertions and deletions (indels), copy number changes, and chromosomal translocations. [Press release](#)

DNAexus is supporting the 3000 Rice Genomes Project (3K RGP) using its cloud-based genome informatics and data management platform. 3K RGP, a partnership between the Chinese Academy of Agricultural Sciences (CAAS), the International Rice Research Institute (IRRI), and BGI in China, has analyzed 3,000 rice genomes, generating more than 100 TB of data that was processed on 37,000 CPU cores concurrently, using DNAexus to complete the mapping and variant calling. Discoveries from this project will support efforts to breed new rice varieties that have improved yield, higher nutritional value, or greater tolerance to pests, diseases, and drought. [Press release](#)

OmniSeq has begun its spin out from Roswell Park Cancer Institute (RPCI) and will immediately begin commercializing its suite of products to support a physician-driven, collaborative approach to genomic diagnostics, matching cancer patients to therapies tailored specifically to them. The

OmniSeq Precision Medicine Technology platform provides physicians with information about a patient's cancer through a proprietary genomic sequencing algorithm and a system that continuously ingests and analyzes peer-generated clinical experiences and therapeutic outcomes for patients. [Press release](#)

Merck & Co. is making *The Merck Manuals*, a series of medical reference books, available digitally for the launch of Global Medical Knowledge 2020. Users can access the new multimedia website, MerckManuals.com, at no charge. The content will soon be translated into multiple languages and made available as a mobile app. The move to an electronic platform will allow for regular updates to the material and real-time news-feeds about the latest developments in medicine. The website also features expert commentary, infographics, animations, video clips, health calculators and trackers, quizzes, as well as a drug interaction checker, pill identifier, and guide to pronunciation of medical terms. [Press release](#)

Scientists from the **Icahn School of Medicine at Mount Sinai** have developed a new technique to reveal epigenetic mechanisms that can drive virulence in bacteria. The research team employed the RS II system from Pacific Biosciences, which can collect data on epigenetic base modifications simultaneously as it collects DNA sequence data. The researchers studied seven bacterial strains, revealing distinct types of epigenetic heterogeneity. For *Helicobacter pylori*, a pathogenic bacterium that colonizes over 40% of the world population and is associated with gastric cancer, the team discovered that epigenetic heterogeneity can quickly emerge as a single cell divides, and different subpopulations with distinct methylation patterns have distinct gene expressions patterns. [Press release](#)

Researchers at **EMBL-EBI** have developed a new approach to studying the effect of multiple genetic variations on different traits. The new algorithm makes it possible to perform genetic analysis of up to 500,000 individuals and many traits at the same time. The researchers tested their algorithm on data from two studies from public repositories, and compared the results with existing tools. Their study of four lipid-related traits (LDL and HDL cholesterol levels, C-reactive protein, triglycerides) showed that the new method is substantially faster, and can explain a larger proportion of these traits in terms of the genetics that drive them. [EMBL story](#)

Qubole announced that **Station X** is using Presto on Qubole's cloud-based big data platform to power GenePool, a software-as-a-service solution for real-time analytics of genomic and medical information. GenePool allows researchers to manage, visualize and analyze processed genomic data from one to thousands of patients and securely share this data with colleagues. Qubole's self-service platform for big data analytics runs on Amazon AWS, Google Compute Engine and Microsoft Azure. [Press release](#)

Tute Genomics closed a Series A1 round of venture capital funding. The company raised over \$3.9 million from investors including Intermountain Healthcare, Healthbox, and Tencent. [Press release](#)

New Products

QIAGEN and **Inova Translational Medicine Institute** (ITMI) announced the launch of Inova Genomes, a compendium of ethnically, phenotypically, and ancestrally diverse whole-genome



sequencing data for researchers to use in cohort analysis programs or to improve success rates in diagnostic odyssey cases. QIAGEN will serve as the exclusive distributor of the database, which is accessible via the company's Ingenuity Variant Analysis and CLC Biomedical Genomics Workbench platforms. The Inova Genomes dataset was built by ITMI from a collection of more than 7,000 whole genomes derived from over 2,800 families, and has been annotated with electronic health record information. QIAGEN also announced the launch of its CLC Microbial Genomics Module to visually explore and analyze microbiomes. [Press release 1, 2](#)

10X Genomics announced the full commercial release and availability of the GemCode Platform, with instrument shipments and installations underway. The GemCode Platform is a molecular barcoding and analysis platform that consists of instrumentation, reagents and software, which delivers long-range genomic information, including haplotype phasing and structural variation. (For more on GemCode, see "[10X Genomics at AGBT.](#)") [Press release](#)

Quanterix Corporation announced commercial availability of its multiplex panels, providing users with the ability to develop their own multiplex assays using the company's Simoa technology. This allows users to achieve similar sensitivity with each marker in a multiplex panel as they would if they were to test individual assays. Quanterix has also developed a multiplex homebrew kit, allowing customers to use their own antibody pairs to develop custom multiplex Simoa panels. The first commercially available multiplex kits are 3-plex panels of cytokines, for measuring inflammatory response. (For more on the Simoa, see "[Quanterix Aims for Early Clinical Adoption with High-Throughput Protein Assays.](#)") [Press release](#)

Core Informatics announced the launch of the Platform for Science Marketplace, a catalog of pre-configured scientific applications. The apps are built on Core Informatics' Platform for Science by an ecosystem of partners and customers. Existing apps include configurable solution sets for scientists involved in Drug Discovery (small molecule and biologics), Next Generation Sequencing (NGS) and Biobanking. New applications are also being created for Bioprocessing, Crop Science, CPG and more. [Press release](#)