CASE STUDY

N_xClinical Software Significantly Increases Efficiency at Ambry Genetics

Background

Ambry Genetics is a privately-held healthcare company with the most comprehensive suite of genetic testing solutions for inherited and non-inherited diseases. Since 1999, Ambry has tested approximately half a million patient samples benefiting 90% of all U.S. patients covered by public and private insurers. Ambry Genetics is dedicated to open scientific exchange to work together to understand all human disease faster and save millions of lives.

Quick Glance

N_xClinical software significantly decreases the time for case analysis and review at Ambry Genetics and provides the laboratory with a fast, multi-user system accessible from anywhere for genomic data analysis and interpretation.

Over 5x more cases processed in the same time with NxClinical

Additional Benefits:

- Improved lab workflow and organization
- A centralized database system to leverage past cases
- Simultaneous and remote access to software
- Audit-trail for traceability and transparency
- First-class Technical Support

Since 2001, Ambry has performed over a million genetic tests. Handling such high sample volumes, and with their commitment to quality, Ambry recently developed a new highly automated "Super Lab" with N_xClinical as a critical component to increase efficiency and consistency while reducing potential human error. The lab allows Ambry to provide cutting-edge, accurate genetic testing in numerous clinical specialties including oncology, cardiology, general genetics, pulmonology, and neurology. True to their commitment of working together, Ambry has developed AmbryShare, a compilation of exome sequencing data from more than 11,000 hereditary breast and ovarian cancer patients available to anyone with an interest in studying it(1).

Challenge

As a quickly expanding lab, Ambry required a growing number of reviewers to view and interpret cases in a multi-user environment. There was a need to improve transparency in array data interpretation from the Clinical laboratory to the variant assessment team and through to the Directors.

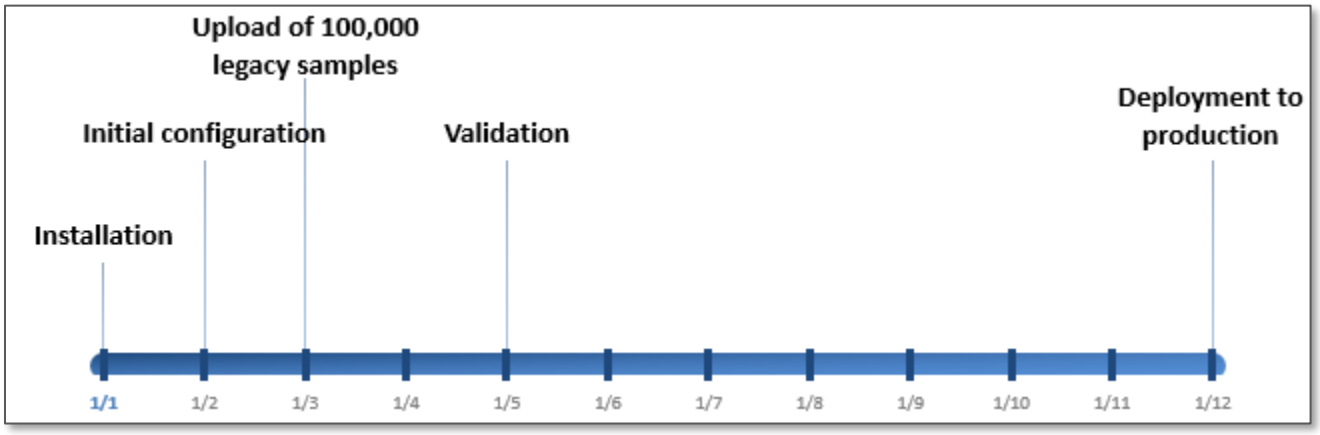
Prior to N_xClinical, Ambry was using BioDiscovery's Nexus Copy Number software for secondary and tertiary analysis. The software used well-accepted algorithms for analysis and interpretation but lacked several features such as a centralized multi-user database, traceability, accountability, and Administrator controlled workflows. Ambry needed a centralized platform whereby all data could be batch uploaded and automatically processed through one of the many preconfigured and clinically validated pipelines. Processed data could then be easily disseminated across the growing enterprise to be accessed by concurrent users, while the system keeps extensive audit trails to record any user modifications.

Solution and Implementation

After extensive review of alternative solutions and evaluation, Ambry selected N_xClinical as their solution for meeting the significant data management challenges posed by their state-of-the-art Super Lab requiring highly automated and quality control processes to keep up with the 24/7 nature of the lab. N_xClinical offered key benefits, such as:

- Administrative control to ensure standardized, validated protocols
- Extensive automation to increase efficiency and minimize chance of human error
- Transparency at all levels with a centralized database of over 100,000 samples accessible by lab technicians, genetic counselors, and clinical directors
- Extensive audit trail to ensure accountability and quality control

Implementation of N_xClinical took just over two weeks and included system installation, setup, validation, and loading of legacy data. As Ambry handles a large volume and offers many tests utilizing a range of different platforms, initial configuration took about one day. Loading of legacy data took some time as Ambry had a large number of legacy samples (several thousand cases). A time-consuming task was ensuring that all samples analyzed in Nexus Copy Number belonging to a single workflow had the same attributes. It took about 15-20 hours of upload of all legacy samples. "The process was a bit tedious, due to the volume of samples we had, but well worth it." This allowed the users to have immediate access to a very large database of previously processed and reported cases as they started working in the new system.



As Ambry was moving from BioDiscovery's Nexus Copy Number into N_xClinical (which uses the same algorithms), software validation was a painless process taking about two weeks. After selecting a sample size of ~50 cases, data was loaded and processed in N_xClinical and the call profiles were compared to those from Nexus Copy Number. Ambry made sure that probes flagged in the positive control were reproduced in N_xClinical and that all control samples had to reflect an absence of reportable calls. In addition, they had to make sure that certain software features such as creating array types, sample types and processing types had to be functional.

Results

Since the deployment of N_xClinical, "our site is now able to support rapidly growing sample volume and equally growing staffing where the analysis software is no longer a bottleneck. The traceability provided by

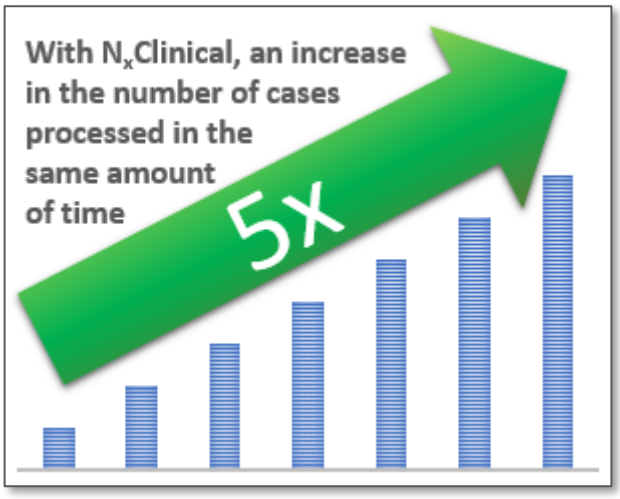
the software during our review stages makes accountability a non-issue," reported Sharon Mexal, Senior Director of Clinical Operations, Ambry.

Improved lab workflow for sample processing

The time-consuming process of organizing the data for upload and queuing it up for Director review has drastically decreased. Previously samples had to be painstakingly organized in batches by the array type/date/time into hundreds of individual folders and subfolders. After data were loaded and processed, Directors needed to know where to find the new cases to review; the meticulous tracking of folder structures was very time-consuming. Now with designated workflows in N_xClinical for each sample type, samples can be uploaded and processed as they come in without prior preparation and the database querying mechanism as well as the built-in sample Stage feature allows Directors to easily locate cases they need to review.

Time savings with real-time sample access

N_xClinical has allowed Ambry to review **upwards of five times more cases in the same amount of time**. Previously, cases from different test workflows were grouped together into different projects. Multiple projects had to be opened and closed repeatedly to compare the case under review with past cases. With all samples from different workflows stored together in one database in N_xClinical, querying the entire database is a breeze. In addition, the interactive browser displays past similar cases and classification alongside the event under review making it quick and easy to confirm interpretation of a new case.



Simultaneous and remote access to software

With a multi-user networked system and unlimited number of users, each user at Ambry can access the software at any time. Users no longer need to wait for an available license to review a case as was the practice with previous software. Audit trailing also ensured changes made by any user are recorded and visible to all users. With remote access from anywhere, all Ambry users, including off-site employees across the nation, can easily access the system.

Centrally managed system access and controls

N_xClinical provides user roles and access privileges with a designated N_xClinical Administrator managing the entire system. This ensures a consistent environment without having to manually create multiple checks and balances. Now the Administrator sets the processing protocols and only users with access to load and process samples can do so without inadvertently changing the settings. Any software updates are designated by the Administrator and performed automatically across software clients ensuring all users are using the same version of the software.

Looking Forward

N_xClinical has greatly increased the efficiency in case review and reporting at Ambry with the company now able to handle five times the case load without needing to acquire additional resources. A designated

N_xClinical Administrator creates the processing protocols and workflows for the different types of arrays ensuring consistency across the lab. The server based system with unlimited users and remote access removed bottlenecks in case review as Directors and other users can access cases from any location without waiting. One of the areas Ambry hasn't yet utilized fully is the Variant Interpretation Assistance system (pre-classification via decision tree) which they hope to start using soon.

Ambry has been testing the upcoming release of N_xClinical and anticipates additional time savings once it is deployed to their production workflow. "Query speeds during bulk analysis will significantly decrease after the release of 3.0 which we anticipate will help our analysts focus more on the task at hand devoid of long wait times." said Aaron Campbell, Supervisor, Clinical Laboratory-Chromosomal Microarray, Ambry. In addition, another feature Ambry values is BioDiscovery's support and accessibility. "We have always voiced our concerns with BioDiscovery and they have been met with either a solution or a timeline to implement one. BioDiscovery optimized the sample querying time for us multiple times before we had a viable solution to analyze in bulk," stated Aaron Campbell. "We hope to continue this dynamic as we continue our relationship with BioDiscovery."

1. The Impact of Data Sharing – A conversation with Dr. Aaron Elliott, CEO of Ambry Genetics. http://www.nature.com/advertorials/insideview/pdf/ivambrygeneticsmar2017.pdf

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