SVS vs Automated Workflows: Choosing the Right Solution for Research and Clinical Analysis

Just a short while ago, the case of Kellie Carey made it to the front page of the Wall Street Journal. Initially, her prognosis in 2010 was very dire. Three months. Lung Cancer.

Ms. Carey is still alive because they were able to prescribe a drug based on the results of sequencing her tumor. It turned out that Ms. Carey has one of at least 15 lung cancer variations, which were discovered in the last decade through next-generation sequencing of tumors. Based on this knowledge, some major cancer centers are beginning to rethink their approach to treating the disease, and drug companies have begun the laborious process of creating drugs to specifically target one type of cancer.

Here at Golden Helix, we are seeing this shift in our daily work as the latest research is now used more and more to diagnose diseases and find the best possible treatment for a particular patient. Clinicians and researchers are working hand in hand in a way that wasn't previously possible. This trend is particularly evident in major cancer centers as well as children's hospitals.

The clinical applicability of DNA and RNA analytical tools is quickly becoming a reality. As a company, this means our tools must serve both worlds. We have empowered researchers for over 10 years to conduct complex analytical work, and we are committed to doing the same for clinicians.

Research vs Clinical

Research

While researchers may have different goals and areas of focus, common considerations come to light when evaluating software. It must be:

- **Scalable:** Our flagship product, SNP & Variation Suite, can handle everything from a single gene panel to a whole genome sequence. In fact, some of our clients are conducting research on thousands of whole genomes. And it can be run on a desktop computer.

- **Visual:** We've received a lot of kudos for our stand-alone genome browser, GenomeBrowse, which gives researchers an interactive experience of viewing their NGS data in a streamlined and intuitive way. In addition to the stand-alone browser SVS now integrates GenomeBrowse to share the same, amazing visualization experience with our SVS clients.

- **Transparent:** Researchers are very sensitive to any

software package that is a "black box." Understanding what algorithms and methods have been implemented is critical so that results can be externally validated. To that end, we document all of the algorithms in SVS and the science behind them and publish it all online. We have yet to find a competitor who does that.

Clinical

On the clinical side, there are a few other requirements that we have to keep in mind. Software must also be:

- **Simple:** Clinicians are required to spend a lot of time with patients. Any tool they implement must therefore be intuitive and easy-to-use to make the most of their office time. Utilizing a pipeline and workflow created by their counterparts on the research side, clinicians don't need to explore. They need results fast, in an easy-to-read format.

- **Compliant:** Clinical applications are surrounded by a regulated set of processes and procedures because the results need to be repeatable. They become part of the patient record.

Hands-On Time Savings

To date, Golden Helix has been able to contribute to the explosion of knowledge by making researchers more efficient. One of our customers told us that a few years ago, conducting a trio analysis with conventional tools took him 14 days. With Golden Helix software, he is now able to analyze 100 trios within two hours.

Clinicians

- Running well-defined workflow on additional samples
- Minimum user-interface knowledge
- Small learning curve
- Limited hands-on time

Researchers

- Building and testing workflows
- More complex but intuitive interface
- Larger learning curve
- Power to investigate and manipulate data

By further automating a workflow we will be able to simplify the usage of SVS as well as the touch time needed by non-expert users down to the minute range.

Analyzing Data Off a Secondary Analysis Pipeline

Using Excel

At first, a scientist without access to a genetic analysis software package often uses Excel to analyze his data. This is understandable as data is often delivered from the sequencing core in an Excel-compatible format.

While this strategy works, it's often time consuming and tedious. The process usually involves comparing several columns and sorting them in order to pick out or remove variants that fit certain criteria.

Analyzing one trio may take weeks to complete while a software designed for genetic data, such as SVS, makes this type of analysis more intuitive and the complete workflow can be accomplished in a fraction of the time.

Using SVS

Unlike Excel, SVS is designed to handle large genetic data. The powerful toolbox built into the software can seem complex, but it strives to strike a balance between power and intuitive design. Automated Workflows in SVS are far easier to learn, but less flexible by the fact that the well-defined workflows have specific pre-set thresholds, with the intention of limiting user-interaction.

If you are looking for a tool to build a workflow or investigate a variety of different genetic file types, while tweaking the options of the available methods, using SVS independently is a more obvious choice than an automated workflow.

Automated Workflow

However, if you are a clinician that needs proven methods in repeatable processes that can be run on new sample sets, an automated workflow makes sense and will limit hands-on time, thus minimizing the possibility of user-error.

We see SVS as a powerful tool that a research scientist can use to build a new workflow. Once that workflow is well-defined and ready for repeatable use, an automated workflow is an easy-to-learn tool that works within SVS that clinicians can implement without needing the in-depth technical and statistical background.

With Automated workflows in SVS, the user creates a new project and selects the samples, then clicks "Start Workflow". All of the steps that are outlined in the initial dialog are then automatically performed in the order listed. The final workflow report is provided in an easy to read and customizable format.

With transparency in mind, all of the intermediate results

populated at each step in the workflow are available in the SVS project and can be examined for verification at any point upon completion.

Our Approach

As with all offered services, our highly trained staff of bioinformaticists, computer scientists, and statistical geneticists work collaboratively with you, involving you in as much detail as desired. We are also professional: we listen carefully to your requirements, work with you to create accurate specifications, develop and test workflows the same way we do our commercial products, and collaboratively test the product with you to make sure it works seamlessly in your environment.
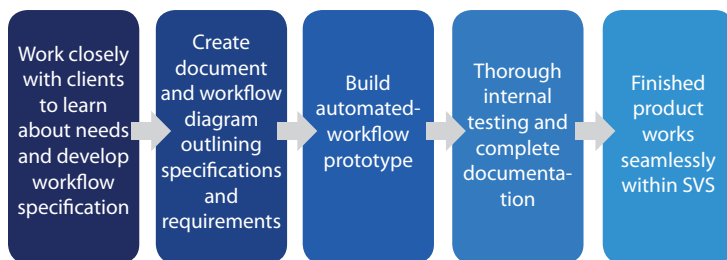
1. Our approach for developing a custom automated workflow begins with working closely with you in order to learn about your needs and develop a workflow specification.

2. Next, we will create a document and workflow diagram outlining the specifications and requirements for the custom automated workflow.

3. Then we will build a workflow prototype.

4. This prototype will undergo the same thorough internal testing that we apply to our commercial products and comes with complete documentation.

5. Finally, the finished product will be tested with you to make sure it works seamlessly in your environment.

At the end of the process, you will own a professionally engineered, "plug-and-play" software tool that can be run on any computer from within SVS using a single, easy-to-understand dialog. The automated function will return all pertinent results, and automatically generate any required visualizations or reports you specify. With transparency in mind, all intermediate results populated at each step in the workflow are available in the SVS project and can be examined for verification at any point upon completion.

You will also receive technical support and standard updates to the workflow for one year.