

Shorter turnaround times for genetic testing

Ambry Genetics in California is taking advantage of the increased throughput, reliability and reproducibility of laboratory automation, using Freedom EVO® workstations to streamline its genetic testing workflows for cancers, cystic fibrosis and numerous inherited diseases.



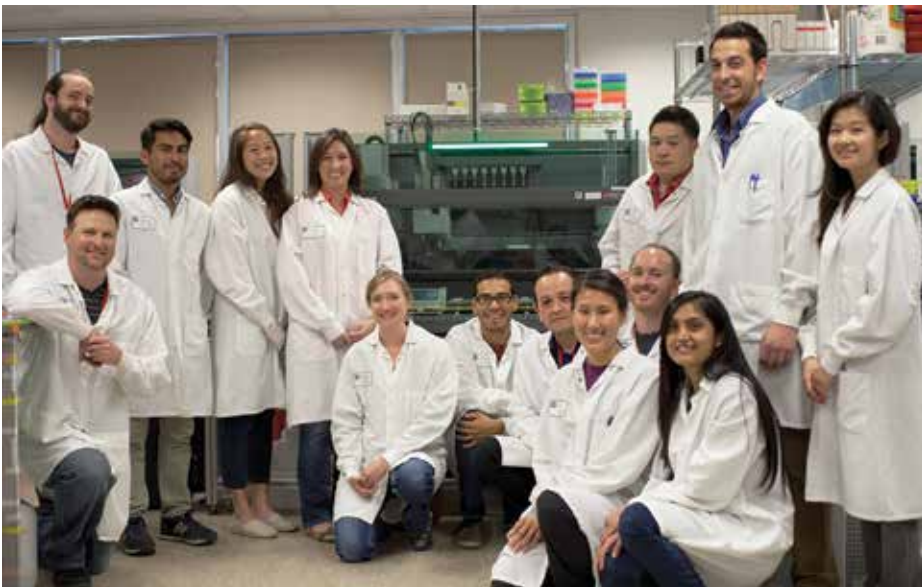
Ambry Genetics, based in Aliso Viejo, California, USA, is a leading clinical diagnostics company at the forefront of genetic testing. It uses a wide range of technologies – including next generation sequencing (NGS), Sanger sequencing, microarrays and multiplex ligation-dependent probe amplification (MPLA) – to offer a portfolio of over 300 highly specialized tests for genetic diseases, including cystic fibrosis and hereditary cancers.

The Company prides itself on its state-of-the-art research and CLIA-approved, CAP-accredited laboratory, and continuously invests in cutting-edge technologies to provide a faster, more efficient service for its customers. This approach means that Ambry's Research and Development Department needs the flexibility to run and assess a broad spectrum of technologies and assay protocols, as Joy Radecki, Associate Scientist, explained: "In R&D, we are always trying to find the best and most efficient technology on the market. We are moving more and more towards NGS,

as is most of the genetic testing environment, however there are many assays which are not suitable for inclusion on NGS panels. As a result, we still do a lot of Sanger sequencing, microarrays and MPLAs, creating multiple workflows in the laboratory."

Automation is vital to meet Ambry's throughput needs and reduce turnaround times and, in 2011, the Company needed to upgrade its existing automated platforms to meet projected growth targets and increase capacity. Joy continued: "We attended demonstrations by eight laboratory automation companies, and Tecan's offering was by far the best choice in terms of flexibility and software programming. The Tecan instruments could clearly satisfy all our automation needs, growing and changing with our workflow, and we could program the systems in house. We initially purchased Freedom EVO 150 workstations with MultiChannel Arm™ (MCA) 96s, Robotic Manipulator Arms and eight-channel Liquid Handling Arms. Since then, we have added multiple Freedom EVO 100 platforms with MCAs or eight-channel Air LiHa Arms. The platforms are extremely reliable and reproducible, and transferring scripts such as liquid classes between machines is also very helpful for process development. All the workstations have the capability to perform DNA extraction, microarray set-up and sample preparation for Sanger sequencing and NGS, but we have found that it is more efficient to optimize each system to a specific workflow, while still retaining the flexibility to perform other tasks as required."

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The Ambry Genetics team

"Our Tecan representative was invaluable in terms of the initial support he provided in choosing the correct instrument configuration, helping us to select the right arms and deck layout. We went through some of our standard operating procedures,

and worked together to build up the Tecan platforms. It was also recommended that we implement the Freedom EVOware® Normalization Wizard, as each NGS assay includes several normalization steps and needs samples to be normalized to specific concentrations. This application alone has been a lifesaver for us, saving several hours every day. By allowing us to simply specify the final target concentration for each well, the platform can quickly and easily prepare 96 samples for NGS, achieving in around 20 minutes what previously took two technicians an hour and a half. It is easy to modify; it only takes two minutes to go into the Wizard script to change the target concentration, which is very helpful for assessing new assays, each of which seems to need a different sample concentration.”

“We also have integrated PosID™ scanners on our workstations running DNA isolation protocols. This allows the technicians to simply load the samples onto the workdeck, and the system scans the barcodes and assigns an ID to each sample, creating an output file that can be easily converted and loaded onto our large DNA isolation instruments. This



Intuitive touchscreen simplifies operation

eliminates the need for a second technician to check the sample labels, freeing up additional staff time and reducing a four-hour isolation process for 96 samples to just 90 minutes.”

“The Tecan platforms are extremely easy to set up and program, and the new Freedom EVO 100s also have the TouchTools™ graphical user interface. This has really helped with staff training, as I can program the

instrument to walk them through the deck set-up with photos and audio guides. The service from Tecan has always been extremely honest and helpful and, whenever we have encountered technical issues or challenges with day-to-day scripting, the Tecan team gets back to me with a solution within a day. I have also attended several of Tecan’s training courses; these were very useful for understanding the capabilities of the systems and, as a result of what I learned, we are hoping to move some of our scripts into Freedom EVOware Plus to help increase our capacity in the future.”

“The Freedom EVO platforms are perfect for our continuously changing protocols, because of their high flexibility and the numerous options available. They have had a huge impact on our throughput and turnaround times, ensuring reliable and reproducible results, which are crucial in a clinical laboratory,” Joy concluded.

To find out about Tecan's genomics solutions, visit www.tecan.com/genomics

For more information on Ambry Genetics, visit www.ambrygen.com



TouchTools guides the user through instrument set-up