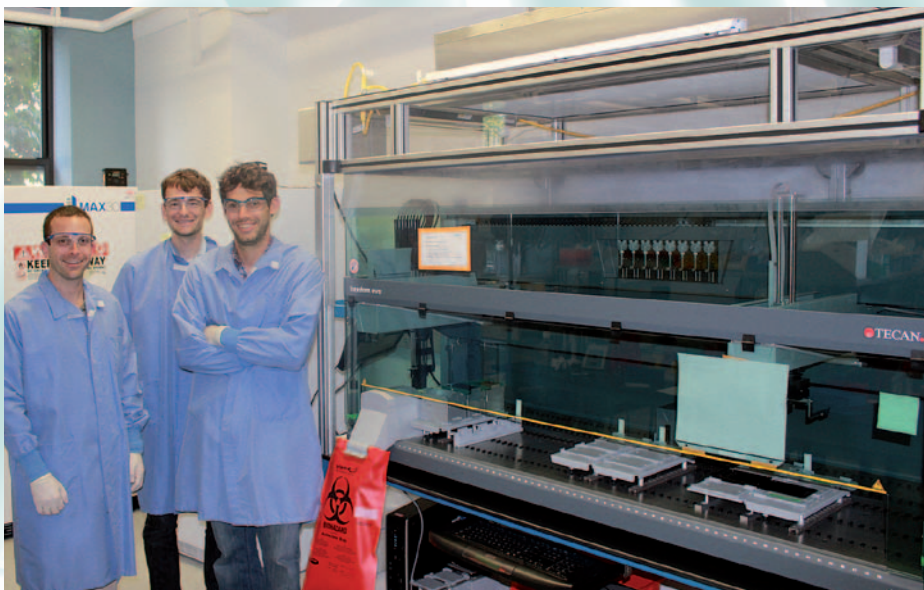


Automated screening for recessive disorders elevates sensitivity to new heights

Carrier screening based on next generation DNA sequencing has been completely automated at Good Start Genetics® Inc., with proprietary chemistry and customized protocols on six Freedom EVO® platforms giving significantly better turnaround times and increased reliability and reproducibility.



Greg Porreca, Pat Saunders and Mark Umbarger with the Freedom EVO

“We chose our first Tecan system because it was one of the more customizable and readily adaptable systems available... we still feel the same today.”

Good Start Genetics, Inc., Cambridge, Massachusetts, is an innovative molecular diagnostics company that offers carrier screening for recessive genetic disorders, launching its pre-pregnancy screening service across the USA in 2011. Dr Mark Umbarger, Associate Director of Technology at Good Start, explained: “We use next generation DNA sequencing to assess whether patients referred to us by *in vitro* fertilization (IVF) clinics are carriers of certain recessive genetic disorders. The specific disorders tested are tailored to the patient, as directed by the physician. The panel of pre-pregnancy tests includes all 14 disorders recommended by the American Congress of Obstetricians and Gynecologists and the American College of Medical Genetics. Next generation sequencing (NGS) is allowing us to look deeper within each disease-associated gene and therefore enables us to identify a larger

number of disease-causing mutations. This comprehensive approach allows us to provide a more sensitive screen than is currently on the market. For example, we can detect ~500 cystic fibrosis disease-causing mutations while conventional genotyping-based approaches typically detect ~100 mutations.”

Mark continued: “When it came to automation, we spoke to a number of other companies that were already using Tecan platforms and they recommended these systems.” Pat Saunders, Molecular Biology R&D Engineer at Good Start, added: “We chose our first Tecan system because it was one of the more customizable and readily adaptable systems available and, despite keeping a close eye on the market, we still feel the same today. We now have six Freedom EVO platforms – three Freedom EVO 200s and three

Freedom EVO 150s – purchased over the last two years. All the platforms have eight-channel Liquid Handling Arms and PosID™ modules, and are adaptable to both 96- and 384-well microplate formats. Some are equipped with a Robotic Manipulator Arm, others with a MultiChannel Arm™ 96 for assay development.”

“All of our assays are automated on the Tecan instruments, starting with DNA extraction from blood through to the genetic assays. There is virtually no hands-on pipetting, with full integration and automation of all steps, including library construction. The platforms in each of our laboratories – development, DNA extraction, pre-PCR and post-PCR – are configured accordingly.”

“Sample preparation, such as that for library construction, molecular barcoding and NGS

on our Illumina® HiSeq™ System are all done using proprietary chemistry developed in house, and fine-tuned, highly customized protocols,” Pat continued. “Some assays are completed in less than eight hours, others take a bit longer. All our tests go through validation on two levels – analytical, for accuracy, and clinical, to assess robustness in the production clinical laboratory. The validation process for our NGS assay on the Tecan platforms took multiple months and comprised simultaneous sequencing of hundreds of samples – target capture, molecular barcoding, sequencing and analysis – using an NGS-based approach, and also by an alternative reference method. We compared the results from hundreds of samples from both methods and found we had a very high specificity and sensitivity.”

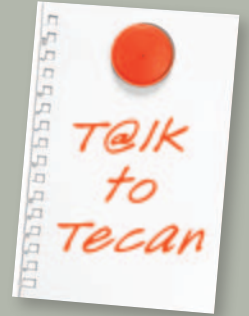
Mark concluded: “For Good Start Genetics, the three major benefits of the Tecan systems are reliable and walkaway automation, high throughput and reproducibility. We are constantly innovating or developing new approaches, improving current methods and coming up with new genetic tests, so the platforms’ flexibility is really important. We have been able to customize each system to our requirements and everyone in R&D and Clinical Operations at Good Start is very happy with them.”

To find out more about Tecan’s genomic solutions, visit www.tecan.com/genomics

To find out more about Good Start Genetics, visit www.goodstartgenetics.com



Nicholas Smith, Head of Product and Strategic Marketing Partnering Business



Leading the debate

Tecan’s Partnering Business has a long history as a skilled and reliable original equipment manufacturer (OEM), bringing many years of expertise to the development of instrumentation for partnering companies. Our direct customers in the OEM business are usually diagnostic companies who approach Tecan with a specific need, looking for a partner to develop a solution for them.

But what makes a company stand out as the OEM partner of choice?

Clearly a system must be developed at the right cost, with the required functionality, and in the specified timeframe, but Tecan stands out from the crowd, offering more than just the basic necessities. Developing an understanding of our customers’ customer – the end-user of the instrument – is the key to that. By being proactive, we add even more value to OEM relationships by understanding our customers’ customers as well as they do, anticipating the future demands of the industry and using our expertise to suggest new approaches and technologies to make our customers more competitive in the market. Activities such as engaging with key opinion leaders, attending relevant conferences and establishing our own focus groups, for example the Tecan Symposium held recently in Boston on the subject of mass spectrometry (see pages 6-7), all contribute to our in-depth knowledge of the field. Our dedicated and experienced team continues to develop a better understanding of end-user applications, enabling Tecan to build superior solutions for its direct partners and ultimately benefitting their own customers – the end-users – as well. It’s a win-win situation.

Email talk@tecan.com to tell us what you think differentiates Tecan from other original equipment manufacturers.