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Dear Colleagues,

The recent American College of Medical Genetics and Genomics Meeting in Charlotte gave me the opportunity to catch up with many old friends and acquaintances while also giving me a chance to make some new ones. For those of you that I have not yet met personally, please allow me to quickly introduce myself. My name is Madhuri Hegde, and I am the Vice President and Chief Scientific Officer at PerkinElmer Genomics. Previous to coming to PerkinElmer and starting this laboratory, I had the opportunity to work with many of you as the Executive Director of the Emory Genetics Laboratory. At Emory Genetics Laboratory, we implemented several NGS based tests, and in most cases were the first laboratory to bring these tests to the clinical arena; all while extensively publishing our results to share with the genetic community as a whole. The laboratory was staffed with ABMG certified board directors and ABGC certified genetic counselors, and we aimed to set the standard for variant interpretation and “free” access to the laboratories’ variant classifications through our revolutionary program “EmVClass.” My goal at PerkinElmer Genomics is to even further enhance this experience while continuing to offer high quality genetic and genomic testing using the infrastructure provided by PerkinElmer.

Now that I have introduced myself, I want to talk a little about PerkinElmer Genomics. During the recent ACMG meeting there was a lot of interest in what we are doing here at PerkinElmer Genomics, but there were also a lot of questions about how can we perform this testing so quickly and at such an affordable cost? Having worked in clinical genomics for over 20 years, I fully appreciate the importance of working with a laboratory that you can trust, and so I wanted to take this opportunity to personally answer some of these questions for you.

“How can you maintain superior quality in your tests while offering them so affordably to patients?”

This is a great question, and in fact, is one of the original factors that attracted me to accept my current role at PerkinElmer. For years PerkinElmer has been known as one of the industry leaders in laboratory automation equipment and technologies, selling equipment into many of the large commercial genetic laboratories around the world. With the resources at PerkinElmer, I knew that I would be able to build a very robust sequencing pipeline (with 10 NovaSeqs), which not only affords high quality low cost sequencing data but also ensures we will not have any downtime due to lack of instrument backup. Additionally, I recognized that PerkinElmer has been a global leader in newborn screening while working very closely with state labs to ensure that every baby born in the US gets timely access to NBS. Thus, after looking at the unique combination of (and access to) technologies, resources, and potential patient populations, I knew that joining PerkinElmer would allow me to build a laboratory with unmatched efficiencies and sustainability.

So no, PerkinElmer Genomics is not sacrificing quality in our assays in order to drive down prices. Instead, we have made the conscious decision to pass on the cost-savings to our clients that we have realized through our unique position within PerkinElmer in order to make testing more affordable than ever before.

“I am always leery to work with a brand new laboratory, why should I trust you?”

I completely understand the question/comment, and I am sure that your patients appreciate the due diligence as well. Although PerkinElmer Genomics is relatively new to the clinical genomics market, the laboratory is actually a well-established entity and has been operating in the newborn screening (NBS) space since 1994. During that time the laboratory has screened over seven million newborns in North America and Europe while also offering 2nd-tier genetic testing, when appropriate. When I joined PerkinElmer 2.5 years ago, it was with the goal of expanding upon this NBS service to also include a full suite of genomic testing from Next-Generation Sequencing (NGS) to Whole Genome Sequencing (WGS). Combined with over 20 years of experience in the clinical genomics space, including the foundation of the Emory Genetics Laboratory, I have assembled a team of experienced professionals from around the genetics industry to ensure that you receive the experience and quality that you are accustomed to.

“How are you able to maintain those prices and TATs without skipping on quality? Are your reports just being sent out through a bioinformatics pipeline?”

I previously touched on the lab efficiencies that PerkinElmer Genomics is able to leverage through the relationship with our parent company PerkinElmer, which allow us to create such an attractive and affordable pricing model. However, we have also implemented other internal reporting efficiencies, such as our Ordered Data Interpretation Network (ODIN) platform, which allows for both rapid and rich data interpretations by our team of FACMG-certified lab directors. Our ODIN platform is a key internal driver that will allow us to consistently meet our TATs. With regards to variant classification, being one of the authors on ACMG’s joint consensus recommendation on the Standard and Guidelines for the Interpretation of Sequence Variants, I fully appreciate the importance of a full and robust variant classification process. At PerkinElmer Genomics, all of the variants are classified according to the ACMG guidelines, and all of our reports are written and reviewed by ACMG-certified geneticists to ensure high-quality result reports. We also openly share and interact with other laboratories to resolve conflicting variant interpretation in open-access databases.

It is my hope that this information gives you some insight into what my team and I are trying to accomplish at PerkinElmer Genomics, which is dually reinforced by our company’s guiding principles:

- Patients and healthcare providers are at the heart of what we do. We believe that genetic testing is a deep-rooted partnership, and are committed to making it accessible and meaningful to the patients and families we serve around the world.
- Leveraging PerkinElmer's position as an industry-leader in Next-Generation Sequencing (NGS) work-flow solutions, we create unmatched efficiency and sustainability within the lab. These capabilities enable us to accelerate our report delivery and increase affordability empowering clinicians to utilize genetic testing earlier in the diagnostic process.
- We understand that generating data is a small part of the genetic testing journey. Our team of experts takes pride in offering families and clinicians insightful and meaningful results. Delivering answers that empower action is our ultimate goal.

If you have any further questions, we would love to hear from you at Genomics@PerkinElmer.com. You can also learn more about us by visiting our website at www.PerkinElmerGenomics.com. Thank you again for your interest in our laboratory, and hope to have the pleasure of working with you in the near future.

Sincerely,



Madhuri Hegde, Ph.D., FACMG
PerkinElmer Genomics Vice President and Chief Scientific Officer