

## Enabling new discoveries with cutting edge technologies

In 2018, the United Kingdom completed its initiative of sequencing 100,000 genomes with the hopes that their findings would lead to better, earlier diagnosis and personalized healthcare for cancer, rare diseases, and infectious diseases. The closing of this study is reflective of how far the sequencing industry has come since the Human Genome Project (HGP) was declared complete in 2003. The development of new genomic and next generation sequencing (NGS) technologies has not only led to advancements in biopharma but in many different markets such as agriculture, consumer genomics, population-scale genomics, and synthetic biology. Companies like Thermo Fisher Scientific, Inc. are on the forefront of leading the market for the creation of new technologies, products, and services to support the growing field of genomics and next generation sequencing.

The global market for sequencing in 2018 was \$10.7 billion and is forecast to increase at a compound annual growth rate (CAGR) of 18.0% to reach \$24.4 billion in 2023.<sup>1</sup> The sequencing services market is also seeing continuous growth and is projected to reach a forecast value of \$14.0 billion by 2023.<sup>2</sup> Scientific innovations, medical initiatives, and market conditions are responsible for the continued growth in sequencing, including precision medicine, large-scale genomics, population sequencing initiatives, long-read sequencing platforms, large volume applications for clinical diagnostics, and the Asia-Pacific market.

While genomic studies and sequencing initiatives are providing a means for groundbreaking discoveries they have also led to an increase in the amount of overhead that organizations are needing to manage.

Laboratory information management systems (LIMS) are being widely used to aid organizations in managing the overhead that comes with performing sequencing and conducting genomic studies. Due to this, the laboratory informatics market has also seen continuous growth.

### **A snapshot at new ways NGS technologies are being used**

Next generation sequencing is providing a mechanism for researchers and physicians to make deeper connections between the biology and successful treatment of cancer. Tissue biopsies have historically been the standard of care for the formulation of a treatment plan, however, tissue biopsies are invasive and not always reliable due to the location of a tumor or metastasis. More recently, liquid biopsies are being used to detect the DNA that is shed by tumors, circulating tumor DNA (ctDNA). ctDNA represents a very small fraction, (<0.5%) of cell free DNA (cfDNA) in the blood.<sup>3</sup> NGS is performed on ctDNA using instrumentation such as the Ion Torrent™ Ion Personal Genome System™, to detect genetic alterations that may be present across diverse cancer subtypes. Performing sequencing on ctDNA refines the molecular stratification of tumors and can be correlated with optimal treatment and prognosis for a patient. There are several mutations in DNA that can be responsible for driving tumorigenesis and NGS enables researchers and physicians to better understand how to diagnose and treat those mutations. The use of liquid biopsies and NGS provides clinicians with the information that they need to better treat patients, and biopharmaceutical companies with the information that they need to develop new therapies.

Genomics and NGS are also playing a large role in the agricultural market, technologies are being used for plant and animal gene expression analysis, genome sequencing, genotyping, genetic engineering, and genetically modified organism (GMO) testing and detection. According to Thermo Fisher, targeted genotyping by sequencing (GBS) is one of the latest innovations in genomics- assisted breeding programs. It enables researchers to interrogate hundreds to thousands of markers across hundreds to thousands of samples simultaneously. Molecular breeding reduces the amount of time and the cost associated with delivering plant and animal species for agricultural use. There are commercially available amplicon resequencing workflows, such as the Applied Biosystems™ Agriseq™ GBS solution that can be used with Ion S5™ sequencing to provide optimal genotyping across several different species, with up to 2.6 million genotypes, all within a day.<sup>4</sup> The use of robust technologies in the agricultural market enables researchers to achieve more accurate marker-assisted breeding, monitor traits throughout a lineage, and advance plant and animal breeding programs.

The relatively low cost to perform sequencing has led to an increase in sequencing for personal genomics, or direct to consumer genomics. The data collected from personal genomics is not intended to be used to drive clinical care decisions, but for genealogy/ancestry investigations or to understand if there is a genetic variation present that is associated with a disease. Companies that provide personal genomic platforms can typically offer three different types of analyses that range in costs; SNP genotyping, targeted sequencing, and whole genome sequencing. The availability to have ones own genomic data generated at a relatively low cost exemplifies how far science and technology has come since the first human genome was sequenced.



## Utilization of a LIMS to manage laboratory overhead

Organizations want to focus on performing genomic (including sequencing) studies to make new discoveries, treat patients, and serve their customers. However, there is a large amount of work operationally that goes into managing these complex studies. Key challenges amongst laboratories performing NGS or genomic analyses are:

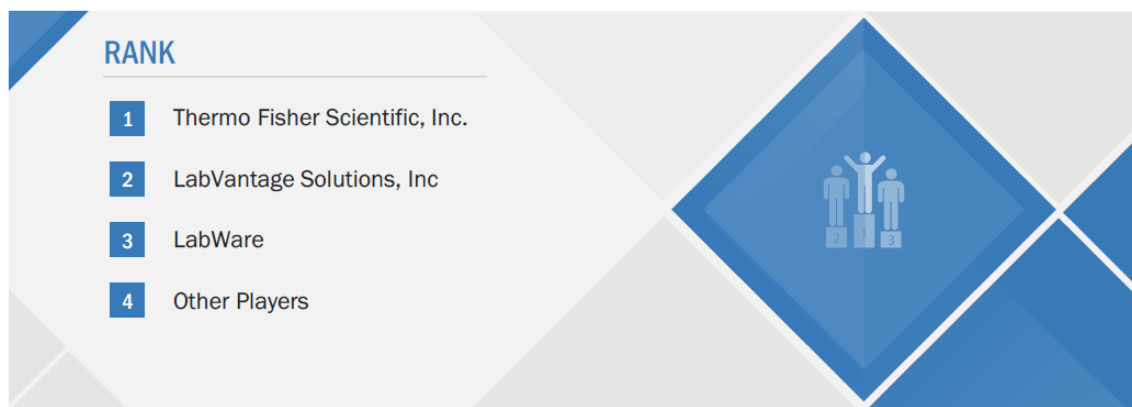
- Handling and storing large amounts of sequencing data
- Maintaining sample chain of custody records
- Managing complex workflows and evolving techniques
- Understanding the current volume of work and assay statuses
- Facilitating communication within an organization and/or with CROs
- Avoiding the creation of data silos
- Ensuring that data is secure

To address these challenges many laboratories are now using LIMS, as the market has seen a return on investment and low total cost of ownership (TCO) for labs that have implemented these systems, more specifically labs that have implemented LIMS in the cloud. LIMS that are hosted in the cloud provide ease of access, improved productivity, outsourced hardware/server maintenance, and increased data security.

LIMS work behind the scenes to ensure that data is secure and is of high integrity. They simplify the data management process while providing many benefits. The benefits of a LIMS can be seen regardless of the type of organization using one (i.e. biopharmaceutical vs agricultural vs core facility). Common benefits amongst these various organizations include streamlined laboratory workflows, enhanced operational efficiencies, ensured quality and integrity in sample results, increased data security and monitoring, and quickened project handoffs.

Thermo Fisher Scientific, Inc. is the global laboratory informatics market leader<sup>5</sup> and offers a variety of LIMS software products.

**FIGURE 29** GLOBAL LABORATORY INFORMATICS MARKET PLAYER RANKING, 2018



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## Conclusion

NGS has evolved into a strategic technology that organizations are using to bring insights from genomics into their business. The use of informatics platforms (LIMS) throughout genomics studies are providing organizations the opportunity to be more agile and efficient in their operations allowing them to focus on science. Continued growth in the field of genomics (including sequencing) is creating many new opportunities across different markets; the use of precision medicine to treat devastating illnesses, the ability to quickly genotype DNA to identify parental lineages, the possibility for a physician to more accurately treat an illness, and farmers the chance to quickly grow more sustainable crops. Creating new technologies and systems to further the field of genomics supports Thermo Fisher Scientific's mission of enabling its customer to make the world healthier, cleaner, and safer.

## References

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