CUSTOMER SPOTLIGHT
REGINA MARIA PRIVATE HEALTH NETWORK

Expand the scope of genomic studies by bringing NGS into your laboratory

NGS is now more accessible than ever with user-friendly solutions, proven performance, and comprehensive expert support from Illumina

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Introduction

Next-generation sequencing (NGS) technology is revolutionizing scientific research with its remarkable precision, versatility, and ability to enable studies with large data sets across multiple sample and assay types. Lower sequencing costs, coupled with user-friendly benchtop sequencing systems and simple workflows, make NGS accessible for labs of all sizes. As a result, many researchers are choosing to bring NGS into their own labs, empowering them to generate high-quality, reproducible data that lead to valuable biological insights.

The Regina Maria Genetic Laboratory, based in Bucharest, Romania, offers a full suite of genomic assays and services. Their team recently switched from outsourcing NGS assays to bringing Illumina NGS solutions in house, enabling them to gain more control over their operations. Even though the team had minimal experience with NGS, they were able to quickly and easily adopt Illumina solutions and develop streamlined workflows to meet their customers' needs. We spoke to Dr Irina Iordanescu, the coordinator of the Regina Maria Genetic Laboratory, about how bringing NGS into their lab has helped optimize their laboratory operations, efficiently prioritize their work, and improve turnaround times while maintaining a high level of performance.

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Q: Can you provide an overview of the Regina Maria Genetic Laboratory? What services does your laboratory provide?

Irina Iordanescu (II): The Regina Maria Genetic Laboratory is a nationally recognized facility for genetic medicine in Romania[1] consisting of two laboratories, one in Bucharest and another in Cluj. It brings together experts in clinical and molecular genetics, with MDs specializing in clinical genetics as well as biologists and biochemists with expertise in molecular genetics. We provide a comprehensive menu of in-house genetic tests and assays, supporting 16 key areas, including obstetrics-gynecology, oncology, pediatrics, cardiology, endocrinology, hematology. We use state-of-the-art technology with an emphasis on the quality and accuracy of genetic testing. The majority of our NGS assays, including whole-exome sequencing (WES) and targeted sequencing panels, are currently for pediatric genetic disease testing and oncology.

Q: What was your previous experience with NGS?

II: Initially, when we did not have large sample volumes to test in house, we would send our samples out to other NGS providers to run the assays. Our team would then interpret the results. Though we were familiar with the technology and its applications, we did not routinely work on NGS assays so we had limited hands-on experience with library preparation and sequencing.

Q: What challenges did you face when outsourcing sequencing? Why did you choose to bring NGS into your laboratory?

II: The biggest challenge that we faced was turnaround time. First, we had to collect enough samples to send out then we had to wait until they were processed by the NGS lab. This meant that we depended on their timelines to get our results. We also could not prioritize samples based on our customers' needs so it was challenging to meet tight deadlines. As our sample volumes began to go up, we started considering bringing NGS in house so we could have greater control over the process, provide results faster, and potentially reduce costs to our customers.

Q: Why did you choose Illumina for your NGS needs?

II: Our team was excited to bring NGS in house. We didn't consider any other company for our NGS needs because we were set on Illumina. We knew and trusted Illumina, so it was our first choice.

Q: How did you implement NGS in your laboratory?

II: Our team had already been doing interpretation and our next step was to start the wet lab methods, such as sample processing and library preparation. After we gained some hands-on experience and our sample volumes began to increase, we decided to invest in an Illumina sequencing system. We decided on the NextSeq™ 2000 System because it would allow us to do a range of NGS assays, from WES to smaller targeted panels, so we could serve all our customers' needs. Once everything was set up, we ran some samples in our lab and sent the same samples to our NGS service providers to compare the results. This gave us the confidence that our processes were working well.

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Q: What challenges did you face while bringing NGS in house? How did you overcome these?

II: Setting up our NGS workflows was quite straightforward. I believe we were the first laboratory in Romania to set up the NextSeq 2000 System. We did not face any major challenges. We were happy with the support, service, and training we received. Our local distributor, ELTA 90, and Illumina were very supportive during the setup and implementation. They were with us throughout the process and answered our requests whenever we called so it was a good experience.

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Q: Did you require any additional support while developing your NGS workflows?

II: We required training on the workflow because we currently use the manual workflow which can be time and labor intensive. However, we are in the process of setting up an automated pipetting system to streamline and speed up the process.

Q: What is your overall experience using Illumina workflows?

II: We are very excited to be able to run our NGS assays in the lab. The whole process of setting up and implementation has been positive. We have had a very good experience with Illumina and our local contact ELTA 90 who have helped us whenever we needed support. Any time we had any issues, they were prompt in helping us solve them.

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Q: How has implementing WES in house impacted your operations? What improvements have you seen compared to sending samples out to a services lab?

II: The biggest improvement that we have seen using Illumina NGS and the Illumina DNA Prep with Exome 2.5 Enrichment is in the turnaround time. We are now able to provide results faster and within the promised timeframe without missing deadlines. We also have much greater control over prioritization and can accommodate urgent samples, which was not always possible when we were outsourcing NGS. By processing, sequencing, and analyzing samples within our own laboratory, we are also able to drop prices for our customers.

Q: Do you have any advice for other laboratories who might be considering insourcing NGS?

"Illumina was our go-to option...we would absolutely recommend Illumina to colleagues who are thinking of starting NGS in their own labs."

II: We think it is great to be able to perform NGS in house. It may seem challenging at first but, overall, it is very rewarding to be able to run your own tests and not depend on other labs. With the right team and training, NGS is now quite accessible. Illumina was our go-to option for germline testing and we would absolutely recommend Illumina to colleagues who are thinking of starting NGS in their own labs.

Q: What are your thoughts on the future of genetic medicine at the Regina Maria Genetic Laboratory?

II: Here in Romania, we are still relatively new to NGS compared to Western Europe, so there is a lot of potential for genomic technology to expand. We hope to see large-scale studies using whole-genome sequencing (WGS) and epigenetics being undertaken. In our laboratory, we would like to offer NGS for many more types of assays than we currently do, including prenatal testing, preimplantation testing for embryos, and WES or WGS trio studies. It is certainly an exciting time for genetic medicine.

Learn more

NGS for beginners

NextSeq 1000 and NextSeq 2000 Systems

Illumina DNA Prep with Exome 2.5 Enrichment

Illumina genomic services

Reference

1. Regina Maria Private Health Network. About the Genetic Center Bucharest Laboratory. https://www.reginamaria.ro/laborator-genetic-center-bucuresti. Accessed November 19, 2024.

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