

# The Illumina Genomics Architecture workflow enables sequencing of 100,000 genomes in Singapore

NovogeneAIT provides sequencing services to create the most comprehensive database to date covering genomes in Asian populations



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Sequencing of the human genome has resulted in many improvements for human health and disease management. However, our knowledge of the human genome is skewed towards populations who have access to these genomic studies and is missing features of underrepresented backgrounds. This bias can lead to regional differences in our genetic information that obscure important variants relevant to human health. Governments and organizations around the world are addressing this lapse with large-scale population genetic studies. One of the organizations supporting a large-scale population health study is [NovogeneAIT](#) Genomics Pte Ltd in Singapore. NovogeneAIT is a joint venture between the Novogene Group and AIT Biotech.

NovogeneAIT is currently supporting the National Precision Medicine (NPM) program. Singapore has appointed NovogeneAIT to sequence 100,000 human genomes as part of the SG100K project. This initiative will capture detailed genetic data for 100,000 consented participants from Singapore, including local ethnic Chinese, Malay, and Indian populations. The project also aims to monitor long-term health outcomes of participants to yield insights into Asian genomic diversity and Asian-specific genetic disease factors. NovogeneAIT is deploying the Illumina Genomic Architecture (IGA) to leverage the efficient workflows, intuitive analysis, and secure data tracking features needed to meet the demands for efficiency and accuracy of this large-scale project. We spoke to Sheanne Soh, who is leading the next-generation sequencing (NGS) team at NovogeneAIT, to learn more about their process.

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#### **How does NovogeneAIT contribute to the National Precision Medicine program and the SG100K project?**

**Sheanne Soh (SS):** NovogeneAIT serves as the sequencing service provider for the NPM initiative in Singapore, a landmark initiative to transform health care by leveraging advancements in genomics. Being the sequencing service provider for this project, we are responsible for sample and library preparation using the IGA workflow, sequencing, and the secured transfer of data to PRECISE\* for their review.

We will be sequencing the genomes of 100,000 Singaporeans. Because Singapore's ethnic diversity captures more than 80% of Asia's overall diversity, this will allow us to establish Asia's leading reference genome database. This data will deepen our understanding of the underrepresented Asian genome and genetic diseases of significance to Asians.

#### **Why did you choose to use the IGA workflow for this project?**

**SS:** NovogeneAIT carried out sequencing of 10,000 human genomes under phase one of the National Precision Medicine Initiative—the SG10k project. We took about three years to complete this project using the Illumina HiSeq™ X platform. To complete sequencing of 100,000 genomes within three years, we knew that we needed to significantly improve our productivity and efficiency. After assessing the various different sequencing workflows on the market, we found that the IGA workflow was best suited to our needs and made the decision to adopt the well-designed workflow for this landmark project.

#### **How has your experience with the IGA workflow been?**

**SS:** The IGA workflow is very intuitive, with lots of prompts to guide my team through each of the steps. This enables us to run more samples with much greater efficiency than other workflows, including the one that was used in SG10K project. Furthermore, the IGA workflow is highly streamlined with limited hands-on time, allowing my team to focus on other administrative work or take lunch breaks between operations.

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\* Precision Health Research, Singapore (PRECISE) is the central entity implementing Singapore's National Precision Medicine (NPM) strategy.

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### How long did it take to get the IGA workflow up and running?

SS: Getting started was relatively quick. The IGA setup, training, and validation only took three months. We currently have five dedicated Illumina NovaSeq™ 6000 systems that we run seven days a week and process up to 840 samples a week. We also had ample training from the Illumina team to get us familiar with the IGA workflow, which took about one month.

### How many samples do you anticipate processing?

SS: We are fully confident that we will be able to sequence > 33,000 samples per year to achieve our goal of 100,000 samples by 2025.

### What do you think of the IGA workflow?

SS: My team members who were previously involved in SG10K and other large projects were very excited about the IGA workflow. They enjoyed the ease of use and minimal hands-on time of the IGA workflow as compared to previous workflows that demanded up to eight hours of hands-on time per day and over one-and-a-half days to manually process 96 samples. Unlike other workflows, the IGA workflow doesn't involve any complicated library preparation steps. It is intuitive and easy to grasp, especially for someone already familiar with Illumina workflows.

With the SG10K project, it took us one to one-and-a-half days just to complete the library preparation step for 192 samples. After which, we would have to wait until the following day to sequence them. The whole preparation process required at least three to four days before we could commence sequencing. With the IGA workflow, we can complete library preparation for 192 samples and start the sequencing run on the same day. The IGA workflow is a lot quicker.

Also, before we began using the IGA workflow, we needed to manually and individually quantify and qualify sample libraries before they could be combined into a sequencing pool for one NovaSeq flow cell. That was a lot of work and effort. The IGA workflow streamlines this process. For samples prepared together in the same plate, you can simply run them on the same flow cell without quantifying them separately.

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## How is the IGA workflow helping you meet your goals?

**SS:** NovogeneAIT is one of the largest and most reliable NGS service providers in this region and a trusted genomics partner to the life science community. Our mission is to advance genomics and improve life by supporting researchers to achieve key scientific goals. This is what matters to us and this efficient and proven workflow will allow us to deliver the best service to our partners. The IGA workflow is a good fit for meeting our sequencing goal of 100,000 genomes and we are very pleased to be part of this national population health initiative.

## Learn more

Illumina Genomics Architecture, <http://support-docs.illumina.com/SHARE/IlluminaGenomicsArchitecture/Content/SHARE/FrontPages/IGA.htm>

SG100K project, <https://www.npm.sg/collaborate/partners/sg100k/>

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