"SMALLER POPULATION GROUPS WHICH ARE NOT IN THE MIX WITH OTHER GROUPS OFTEN REMAIN ISOLATED FOR CENTURIES OF YEARS."

After launching in 2016, the project caused a stir confirming that it was the breakthrough that the Asian market had been waiting for. The consortium hit headlines when seeking founding partners, and it's no surprise that institutions came flocking. Genomics-driven research and diagnostics company MedGenome, and Macrogen were soon recruited, and with their help and contributions, 2,000 genomes have successfully been sequenced to date.

For Macrogen in particular, getting involved with the organisation was a no brainer; they were already keen to discover and learn more about the Asian genome.

From 2003-2009 they had carried out their own genome project of isolated tribes in Mongolia under a Korean government fund.

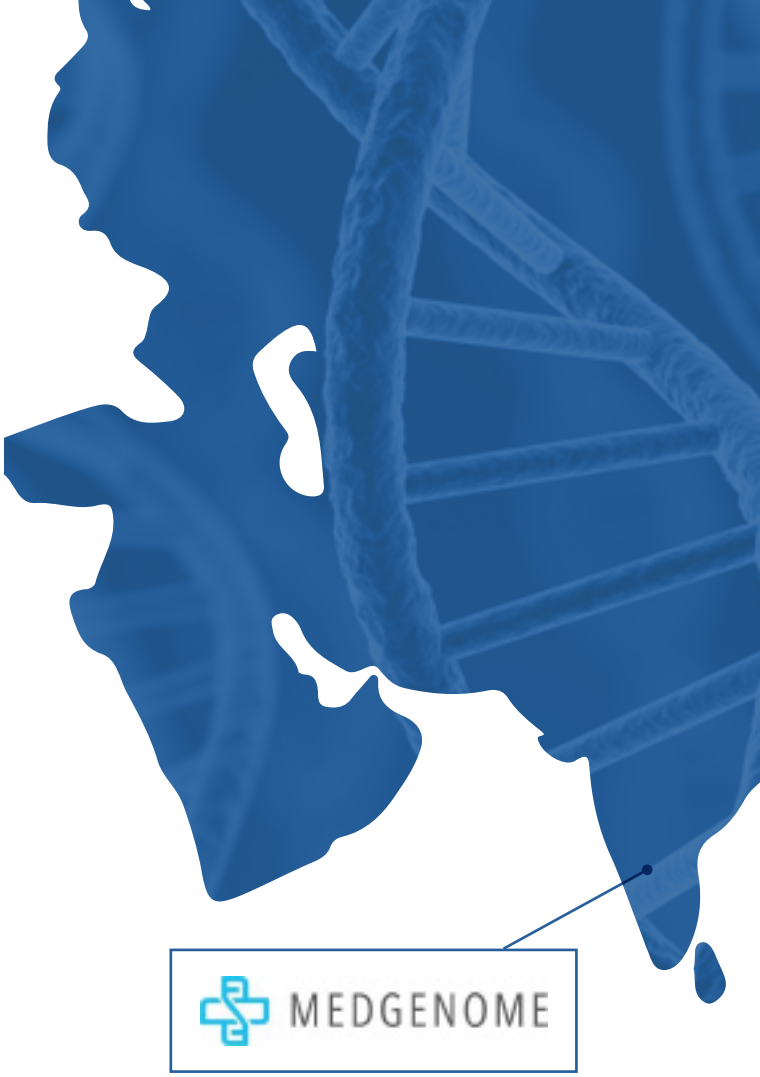
Chairman of Macrogen, Dr Jeong-Sun Seo, explains, "We had already sequenced 1,000 genomes with our own North East Asian genome project from 2014 to 2016 and we wanted to expand that. During this time Dr Schuster informed us of what they were doing and we very quickly accepted".

"Dr Schuster had a very good team in India and South East Asia; we wanted to collaborate with them. The Asian market is very important; we want to make good genomic content for precision medicine. If you combined the genomic information of Asia, then the area would be covered effectively."

Headquartered in Seoul, Korea, Macrogen are already paving the way for genomics in Asia, acting as the world leading genomics service provider spinoff from the Genomic Medicine Institute in Seoul National University. Based on its 20 years of DNA sequencing experience and research in collaboration with GMI-SNU, Macrogen envisions its contributions to society through the realisation of precision medicine.

Despite making initial progress, a lot still needs to be done to achieve the ultimate goal. "The genomes of Asian individuals are different from that of European individuals," notes Dr. Seo. "We have the most contiguous, high-quality Asian reference genome on our hands. We would like to use this reference and generate more population-specific references in Asia. These then could be used to identify Asian-specific variants and their allele frequency. While some of them have no effect on Asian people, some will have medical importance," he adds.

Dr. Seo states, "Macrogen continues to try reducing the cost of sequencing by optimising every sequencer and technology. By using our skills and experience, we can help other Asian countries through this genome project." He adds that, eventually, "We would like to connect the genomic data with clinical data, i.e. standardised electronic medical records, in order to revolutionise the healthcare system in Asia, a phenomenon that is already happening in the US through USA precision medicine initiative."



MedGenome on the other hand, agreed with these principles especially after most genetic research globally included only the Caucasian population. After playing witness to the genetic diversity apparent in India, they soon noticed that other Asian countries were displaying similar characteristics, and that small groups didn't seem to be mixing. With a lack of resources, the company soon set out to seek a new mode of discovering more.

Founder, Sam Santhosh, comments, "Smaller population groups which are not in the mix with other groups often remain isolated for centuries of years; they are usually a good platform to understand due to the neutral changes, and genetic differences. However, they are not seen as important from a medical point of view," he reinforces.

"We saw that there was no baseline data, and our diagnostics were not growing fast because we didn't have data to compare to if we noticed a new genetic mutation," recalls Santhosh.

"Baseline data is vital if you want to provide precision medicine, as without it you aren't able to figure out if a genetic mutation is good or bad. Some may be causing major disease; we need to find out which ones are important."

With very little data covering this part of the world, MedGenome yearned for a larger project and this is exactly what they got. With two lines of business, including genetic tests for patients and commercial research, they home one of the biggest plants in India for genetic sequencing.

Professor Jeong-Sun Seo
Chairman of Macrogen and Director
Genomic Medicine Institute (GMI)

Professor Stephan Schuster
Full professor and founding member of SCELSE (RCE)
Nanyang Technical University (NTU), Singapore

Sam Santhosh
Founder
MedGenome

MedGenome, Macrogen and Schuster are all Genome Asia 100k Collaborators, whereas Novogene is helping the region aside from the project

"THE ASIAN MARKET IS VERY IMPORTANT; WE WANT TO MAKE GOOD GENOMIC CONTENT FOR PRECISION MEDICINE."

As a result of joining forces with the project, they were offered a space to open a new laboratory in Singapore. Now able to sequence humans genomes close to a thousand dollars a genome, Singapore acts as a vital hub, all made possible because of brand new technologies.

"Disease in a population group affects the chances of discovering new genes, and therefore mutations are much higher. It becomes much more difficult for researchers to pick out genetically important differences. The technologies can be used within isolated technologies," adds Santhosh.

The initial results from the first stage of sequencing, focusing on the 2,000 sequenced genomes will be available later this year to the public in a published paper. However, Professor Schuster tells Front Line Genomics, "The first phase is being written up as we speak. We can share that the results were quite surprising and the number of variants we have discovered is high. Releasing this data will make a significant contribution to the already existing variant database.

"It will make absolutely clear what we stated during our starting hypothesis, we need to include Asian genetic diversity in order to address important medical questions in terms of whether something is an ethnic markers or a European marker, because in many instances they might be based on largely European frequencies."

There is a real sense that releasing the publication to the public will make a significant contribution to the already existing variant database. The project is now looking to the future, and believes that they will achieve their ultimate goal of sequencing 100,000 genomes in the next three to five years.

"The goal is of course is that people are so interested in the progress of our project, and would rather be a part of it than compete. We believe that the first 10,000 genomes might take longer, but the rate will speed up and be really possible to accomplish," comments Professor Schuster.

The apparent momentum received by the project is both comforting and reassuring, as it reaffirms that the industry are all working towards the same goal.

Santhosh concludes, "The data available will be useful as there is little work being done that actually contributes to the science and base data that we already have, this will be able to help in testing, daily diagnostics and drug discovery. I am optimistic that this will be very, very useful." ■