

“We are looking at NIPT, cancer screening tests, and infectious disease surveillance as growth areas”

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BGI Genomics, a pioneering institution that has embarked on a remarkable journey from its involvement in the Human Genome Project to establishing itself as a global frontrunner in the field of precision medicine. Headquartered in Shenzhen, China, it is the one of leading integrated solutions providers of precision medicine, with services covering more than 100 countries and regions, involving more than 2,300 medical institutions. Jeremy Cao, General Manager of BGI Genomics and BGI Group Southeast Asia delves into the organisation's transformative trajectory and its substantial contributions to the world of genomics.

BGI Genomics has a strong foundation in supporting the Human Genome Project. How has the organisation evolved over the years in terms of its research focus and contributions to the field of genomics?

BGI Group, our parent company, was founded in 1999 to participate in the Human Genome Project, often referred to as one of the three major scientific endeavours of the 20th century, alongside the Manhattan Project and the Apollo Moon Landing Programme. As the sole developing country participating in this project, China took on 1 per cent of the task, completing the sequencing of 30 million base pairs on Chromosome 3.

BGI Genomics has evolved into a leading integrated solutions provider of precision medicine. We are committed to enabling and accelerating scientific innovation, strengthening the prevention and control of genetic diseases, and making meaningful contributions to developing precision medicine and diagnoses.

We have contributed to the development of genomics, in particular cancer research. As of November 2022, BGI Genomics has published over 440 SCI (Science Citation Index) papers in cancer research, with a cumulative impact factor of over 4,000 points. Several BGI Genomics papers have been published in *Nature Medicine*, *Cell*, *Nature*, *Science*, *Cell Research*, *Nature Genetics* and other top scientific journals.

Could you highlight some of BGI Genomics' notable contributions to the field of genomics?

Relying on cutting-edge sequencing and bioinformatics technology, we provide our customers with expert and affordable clinical molecular diagnostic solutions and massive parallel sequencing (MPS) research services. Our services cover over 100 countries and regions, involving more than 2,300 medical institutions.

In Southeast Asia, BGI Genomics has a diverse portfolio of projects. This includes but is not limited to collaborations such as the thalassemia "Screening, diagnosis, treatment" closed-loop programme in Thailand and Indonesia, colorectal cancer screening programme in Thailand, rare cancer research at the National Cancer Centre Singapore, and cervical cancer screening cooperation with Brunei's Ministry of Health.

One noteworthy project is Thailand's thalassemia "Screening, diagnosis, treatment" closed-loop programme. Thalassemia is an inherited blood disorder caused by insufficient or nonfunctional haemoglobin that affects over 345 million people worldwide.

This initiative involves cooperation with the Eastern Economic Corridor (EEC) Office and the Thai Ministry of Health in conducting clinical trials for thalassemia gene therapy. Mahidol University's Siriraj Medical School, Chulalongkorn University's Medical School, and the Thai Clinical Research Center are project partners collaborating with BGI Genomics to facilitate thalassemia gene screening and treatment, ensuring the smooth initiation and execution of clinical trial projects.

As part of the EEC's efforts to establish a commercial clinical thalassemia gene therapy centre, EEC provides the facility while jointly constructing specialised hospitals and offering legal and policy support. In addition to this, BGI Genomics is collaborating to introduce cell storage facilities, leveraging the "cell + gene" platform's advantages.

What challenges and opportunities does the genomics and proteomics industry face in terms of regulatory and ethical considerations, data privacy, and public perception?

Firstly, we must stay true to our mission of 'Omics for all' to make genomics technology more accessible, available, and affordable to enhance health outcomes for all.

In line with our guiding principles, we place a lot of emphasis on regulatory and ethical considerations as well as data privacy. We follow the applicable regulations across the countries and regions of our operations. Regarding data management, we follow industry best practices such as purpose limitation, data minimisation, storage limitation, and confidentiality.

Research must pass our Institute of Review Board of Bioethics and Biosafety (BGI-IRB) review and follow the corresponding international regulations such as the WHO Review of the Work Guidelines for Ethical Committees for the Evaluation of Biomedical Research and the Council for International Organizations of Medical Sciences (CIOMS) International Code of Ethics for Human Health-Related Research.

Looking ahead, what are the future plans and goals for BGI Genomics in terms of advancing genomics research and expanding its services globally?

We are committed to the continued development of precision medicine and diagnoses to enhance health outcomes worldwide. Within Southeast Asia, we are looking at three key growth areas: non-invasive prenatal tests (NIPT), cancer screening tests, and infectious disease surveillance.

Take DNA-based NIPT, for example; our NIFTY test offers screening for some of the most common trisomies present at birth, including trisomy 21 (Down Syndrome), trisomy 18 (Edwards Syndrome), and trisomy 13 (Patau Syndrome). Yet, based on my observations, take-up across Southeast Asia is relatively low at less than 10 per cent. Given NIPT involves a simple blood test that can be done in the first trimester of pregnancy, there is more room for further adoption.

There were over 1.9 million new cases of colorectal cancer in 2020, making it the third most common cancer worldwide. Colorectal cancer (CRC) is often diagnosed at a late stage due to the lack of symptoms – which is why it's often referred to as a 'silent killer.' When CRC is detected at an early stage, the 5-year relative survival rate is about 90 per cent. But only about 4 out of 10 CRC cases are found at this early stage. Our COLTECT screening test is non-invasive and has 88 per cent CRC sensitivity, and for early detection, its sensitivity for advanced adenoma is 46 per cent, which is superior to conventional faecal tests.

Infectious disease surveillance is also crucial in the post-COVID-19 new normal. Our PMSeq solution leverages high-throughput sequencing, microbial-specific database comparison, and intelligent algorithm analysis to identify complicated and critical-to-detect infections. This also promotes insights into antimicrobial resistance and susceptibility (AMR) and contributes to solving problems such as tuberculosis drug resistance.

Ayesha Siddiqui