

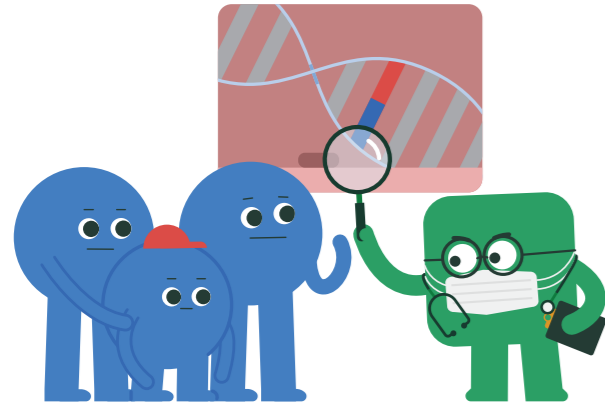
## 6 How could patients and their families benefit from Whole Genome Sequencing?

### Case Study (1) – Enhance Diagnostic Rate

Patient A was a 4-year-old boy with developmental delay and epilepsy. He needed regular clinic visits and medication to manage the condition. After various medical tests, the cause of his disease remained unknown.

With Whole Genome Sequencing, the medical professionals successfully identified the variants in his genome that were related to his condition and were thus able to provide more targeted and effective treatment.

His parents always wanted a second child but were worried that the child might suffer from the same disease. The Whole Genome Sequencing results of both parents revealed that there was no hereditary link regarding the boy's genetic variant. This discovery eased their worry and greatly helped with their family planning.

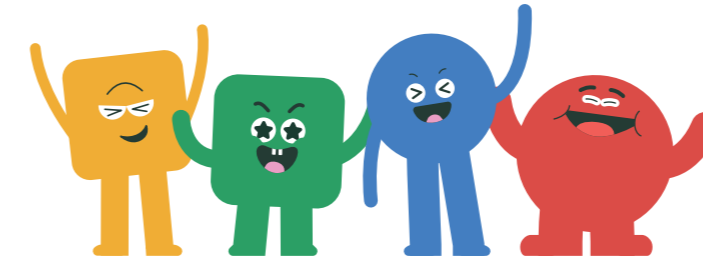
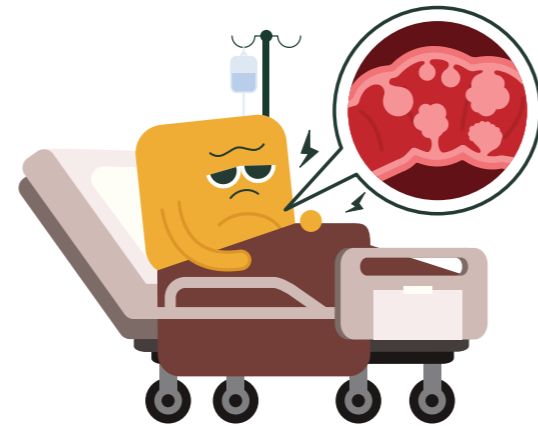


### Case Study (2) – Provide More Precise Treatment for Patients

In a medical check-up, a large number of polyps were found in Patient B's large intestine. The doctor suspected that it was "polyposis syndrome" caused by a specific genetic variant.

For patients suffering from this condition, their large intestines would be filled with thousands of cancer-induced adenoid tumours. To reduce the risk for cancer, in general, patients would be advised to have their entire large intestine removed; yet by doing so, their quality of life would be adversely affected.

The doctor conducted Whole Genome Sequencing for Patient B to look for the cause of the disease. It was found that Patient B's genome contained a variant that caused polyps but not malignant cancer. The doctor concluded that the risk of those polyps turning into bowel cancer was low. As a result, Patient B did not need to have his entire large intestine removed and was free from the adverse effects brought by the surgery.

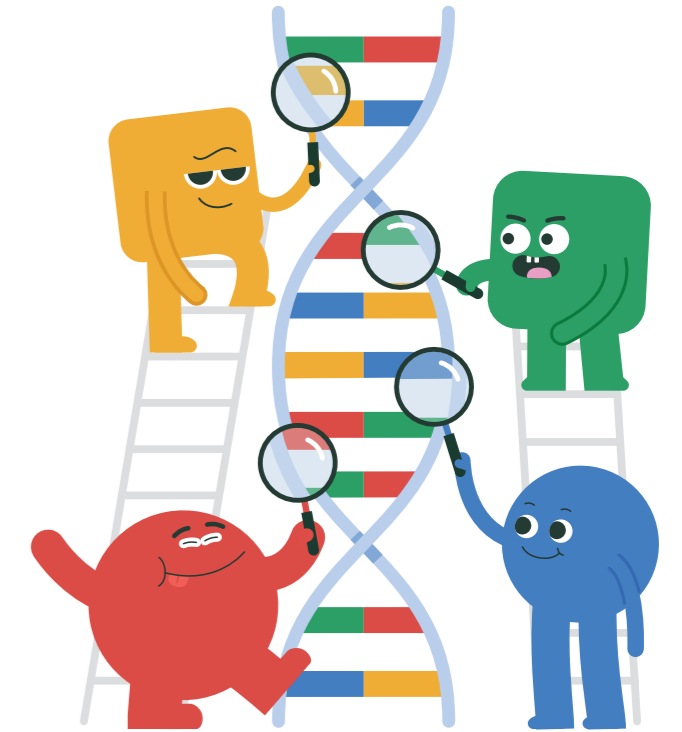


For more information about HKGP,  
please visit  
[www.hkgp.org](http://www.hkgp.org)

 Hong Kong  
Genome Institute  
香港基因組中心



SEE THE UNSEEN  
看見病因



## Hong Kong Genome Project

## 1 Project Overview

Genomic medicine is an important sphere in contemporary medicine and scientific research, with huge potential in disease screening, accurate diagnosis and personalised treatment. In view of this, the Hong Kong SAR Government established the Steering Committee on Genomic Medicine in December 2017 to map out the strategies for developing genomic medicine in Hong Kong and subsequently set up the Hong Kong Genome Institute (HKGI) upon the recommendation of the Steering Committee to promote the city's long-term development of genomic medicine.

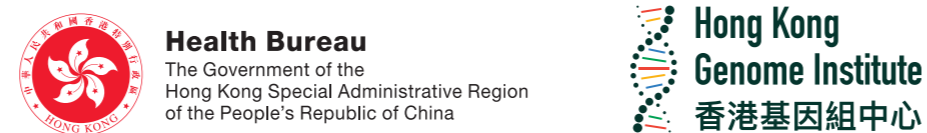
Upholding the vision **"to avail genomic medicine to all for better health and well-being"** and fully supported by the Health Bureau, HKGI works in close collaboration with the Department of Health, Hospital Authority, medical schools of local universities and other stakeholders to accelerate the development of genomic medicine in Hong Kong along four strategic foci: integrate genomics into medicine, advance research, nurture talents and enhance public genomic literacy.

As the first step towards achieving its vision, HKGI launched the Hong Kong Genome Project (HKGP) in 2021. As the city's first large-scale genome sequencing project, HKGP serves as a catalyst to benefit patients and their families with more precise diagnosis and personalised treatment through whole genome sequencing (WGS). It also aims to establish genome database of the local population, testing infrastructure and talent pool to address the healthcare needs of Hong Kong in the long run.

HKGP focuses mainly on diseases and research cohorts that would benefit from WGS. They include undiagnosed diseases, hereditary cancers and cases related to genomics and precision health. Participation of HKGP is voluntary. Depending on clinical and research needs, data collected from HKGP will be used for clinical applications. Anonymised data will be used for approved medical research under stringent scrutiny, enabling clinicians and scientists to better understand the relationship between genomes and different diseases. The knowledge and experience accumulated will play a crucial role in fostering future medical development in Hong Kong.

## 2 Project Implementation

Overall coordination and execution of the Project is carried out by:



HKGI collaborates with various stakeholders\* to implement the Project:



HKGI has set up Partnering Centres in three hospitals and four referring networks\* under the Hospital Authority (HA), and is extending collaboration with other HA hospitals, to recruit eligible participants to join the Project.

Partnering Centres:



Referring Networks:



\*Names are listed in alphabetical order

## 3 Eligibility

Patients of the following diseases/ cases and their family members are covered by HKGP:

- Undiagnosed disorders;
- Hereditary cancers; and
- Cases related to genomics and precision health.

Eligible participants have to be referred by designated Partnering Centres to join the Project.

## 4 What is "Genome"?

"Genome" refers to the entire set of genetic materials in a living thing. For every human being, the genome includes over 20,000 genes. Each of us has our own unique genome, which can be thought of as an operation manual for our body. It provides instructions that determine the physical characteristics of our body such as skin colour, height and potential risks of developing different diseases.

If we compare the genomes of two people who are not related by blood, 99.9% of their genomes will be the same and only 0.1% of them will be different. However small this 0.1% might look like, it is significant enough to represent millions of differences in the two DNA sequences, accounting for each person's unique physical characteristics.

HKGP is set up to help patients and their families look for the disease-causing genetic variants among the millions of differences in their DNA sequences, that is, to see the unseen. This intricate and complex mission will be carried out through Whole Genome Sequencing analysis.



## 5 What is "Whole Genome Sequencing"?

There are four types of bases that serve as the basic unit of DNA: A, T, C and G. The sequence, or order, of these bases determines an individual's genetic code. This code carries the genetic information and instructions for our body's physical features and functions. "Whole Genome Sequencing" is the technology that enables us to read the sequence of all 3 billion bases in the human genome.

With genomic data obtained through this technology, medical professionals and scientists have to analyse and research through a large number of cases over a long period of time in hope of identifying the genetic variants that might be the causes of specific diseases, after which more targeted and effective treatment options could be developed for patients.

