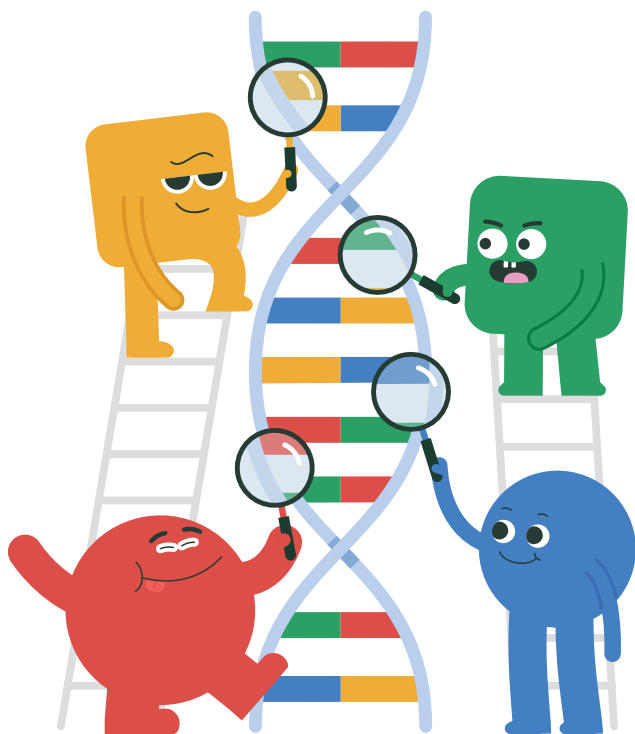


SEE THE UNSEEN  
看見病因



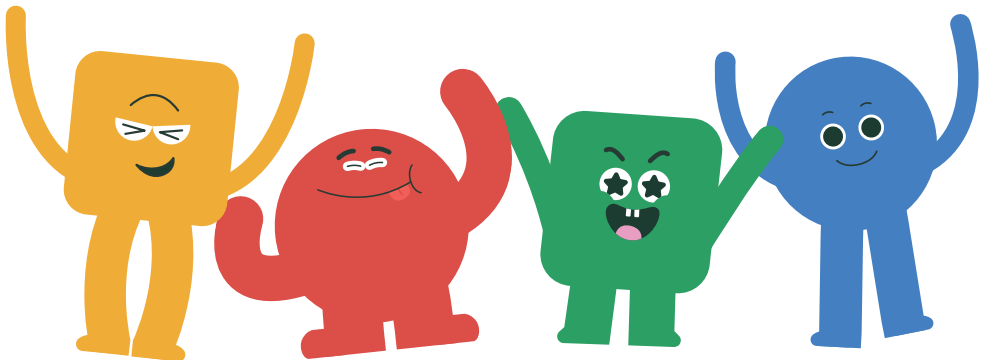
**Hong Kong Genome Project**

## 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 Genome Institute (HKGI) was established by the Food and Health Bureau in 2020 to implement the Hong Kong Genome Project (HKGP) so that patients could benefit from more precise diagnoses and more effective treatment. HKGP also aims to nurture talents, promote the development of genomic medicine in Hong Kong and advance the health of the general population in the long run.

Themed “See the Unseen”, HKGP is the first large-scale genome sequencing project in Hong Kong. By collaborating with the Department of Health, the Hospital Authority and local universities, the Project recruits eligible patients and their family members on a voluntary basis.

In total, HKGP plans to conduct Whole Genome Sequencing for 20,000 cases in two phases, the pilot phase and the main phase. Data collected will be used for clinical application. 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:



食物及衛生局  
Food and Health Bureau



Hong Kong  
Genome Institute  
香港基因組中心

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



衛生署  
Department of Health



醫院管理局  
HOSPITAL  
AUTHORITY



香港中文大學醫學院  
Faculty of Medicine  
The Chinese University of Hong Kong



HKU  
Med

For the pilot phase, HKGI has set up Partnering Centres in three hospitals\* under the Hospital Authority to recruit eligible patients and their family members to join the Project.

香港兒童醫院  
Hong Kong Children's Hospital



威爾斯親王醫院  
PRINCE OF WALES HOSPITAL



瑪麗醫院  
QUEEN MARY HOSPITAL

## 3 Eligibility

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

### Pilot Phase (2,000 cases):

- Patients with undiagnosed disorders and their family members; and
- Patients with hereditary cancer and their family members.

### Main Phase^ (18,000 cases):

- Patients with diseases covered in the pilot phase and their family members; and
- Patients with other diseases and their family members who might benefit from Whole Genome Sequencing.

\*Names are listed in alphabetical order

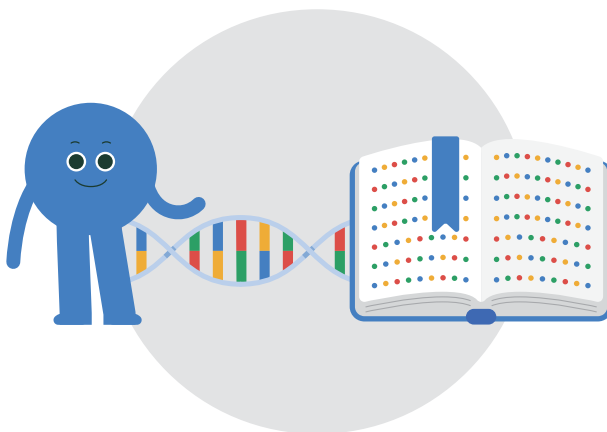
^Details of the main phase will be announced in due course

## 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.

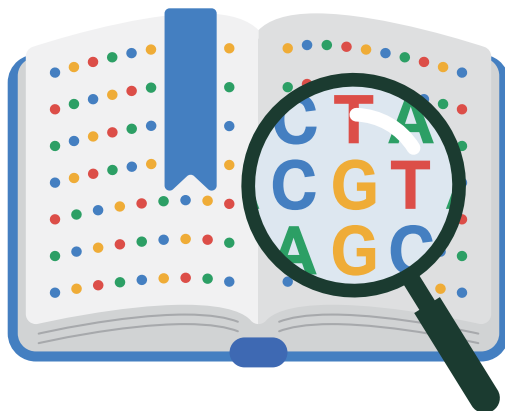
HKGP is set up to help patients and their families look for the disease-causing genetic variants among the millions of differences in their DNA sequences, that is, to see the unseen. This intricate and complex mission will be carried out through Whole Genome Sequencing analysis.



## 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.



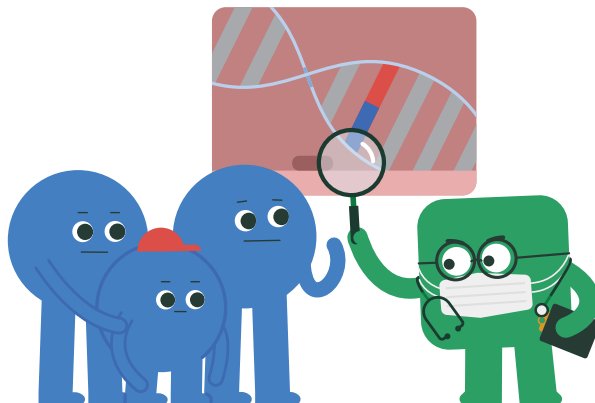
## 6 How could patients and their families benefit from Whole Genome Sequencing?

### 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.

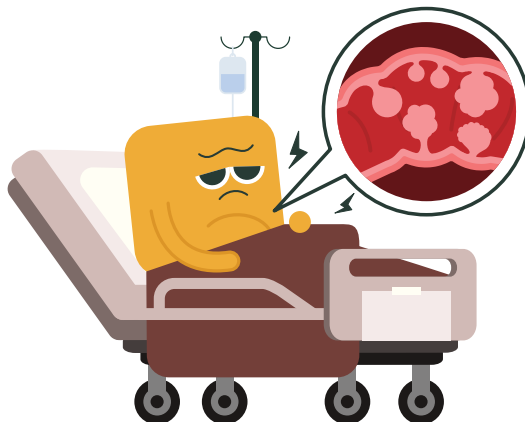


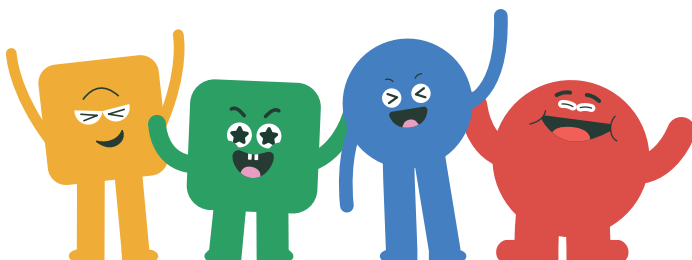
## Case Study (2) – Provide More Precise Treatment for Patients

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.hkgrp.org](http://www.hkgrp.org)**

