For discussion on
10 September 2021

Legislative Council Panel on Health Services

Implementation of Hong Kong Genome Project

Purpose

This paper updates Members on the implementation of the Hong Kong Genome Project (HKGP).

Background

2. Genomic medicine[1] is an important sphere in contemporary medicine and scientific research. Given its rapid advancement and huge potential in screening, diagnosis and personalised treatment of diseases, many countries have actively carried out large-scale genome sequencing projects in recent years to collect genome data for cutting-edge scientific research and clinical purposes.

3. In view of the importance of genomic medicine to future medical development, the Secretary for Food and Health appointed the Steering Committee on Genomic Medicine (the Steering Committee) in December 2017 to lead the study on strategies for developing genomic medicine in Hong Kong. On the recommendation of the Steering Committee, the Chief Executive announced the launch of the HKGP in the 2018 Policy Address, with $1.2 billion subsequently earmarked in the 2019-20 Budget for related purposes. The Food and Health Bureau (FHB) briefed this Panel on the details of the HKGP in January 2019 and on the overall strategic development of genomic medicine, including the important roles played by the HKGP, in June 2020.

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[1] A genome is the complete set of DNA found within a cell. Genomic medicine uses genome data to support clinical treatment.

Whole Genome Sequencing

4. Whole genome sequencing is a technology widely used in the international medical and research fields to determine all deoxyribonucleic acid (DNA) sequence in one's genome. According to international and local experience[2], the diagnostic yield of uncommon diseases could be raised from around 10% up to around 30% by using whole genome sequencing as compared with common diagnostic tests. Furthermore, whole genome sequencing enables us to understand better on the genomic changes that cause an illness of an individual or a cluster, thereby helping clinicians decide on more personalised treatment for patients according to their conditions. In the long run, a large volume of whole genome sequencing data together with the clinical data of local patients and their families will be of great research value, as they will help researchers gain a better understanding of the causes of various hereditary diseases, thereby leading to the development of more effective screening, diagnosis and treatment methods.

Update on the HKGP

5. The FHB established the Hong Kong Genome Institute (HKGI) in May 2020 to take forward the HKGP in partnership with the Hospital Authority, the Faculty of Medicine of the Chinese University of Hong Kong, the Li Ka Shing Faculty of Medicine of the University of Hong Kong (HKU) and the Department of Health. Three advisory committees comprising university academics, doctors, representatives from patient groups and the industry have been set up to advise the HKGI on the scientific, data and ethical issues respectively. The HKGI is staffed by a multi-disciplinary team of professionals including doctors, scientific officers, bioinformaticians and genetic counsellors, etc.

6. The HKGP will be implemented in two phases. The pilot phase, which covers about 2 000 cases with undiagnosed disorders and

[2] According to the literature review of the Hong Kong Genome Institute, the diagnostic rate of uncommon diseases using whole genome sequencing is in the region of 21% to 30%.

hereditary cancers, has commenced in July 2021. The main phase, which is extended to cover 18 000 cases of diseases linked to hereditary components, is expected to commence in mid-2022. With both patients and their family members recruited, it is estimated that a total of about 50 000 genomes will be sequenced under the HKGP.

Workflow of the HKGP

7. In the pilot phase, eligible participants will be recruited by the partnering centres at the Hong Kong Children's Hospital, Prince of Wales Hospital and Queen Mary Hospital in collaboration with the HKGI. Healthcare professionals of these hospitals will conduct preliminary screening and refer suitable cases to the project teams of the partnering centres. When cases are considered suitable for inclusion in the HKGP after being assessed by the partnering centres, genetic counsellors will explain the project in detail to the patients and their family members, seek their authorisation for the use of their samples and clinical data in the HKGP, and draw their attention to the fact that participation in the HKGP is entirely voluntary and will not affect the patients' regular clinical treatment. Genetic counsellors will ensure that participants have signed the consent form with informed consent before they are admitted to the HKGP.

8. After giving informed consent, participants will have their blood samples taken by healthcare personnel at the hospitals for the purpose of sequencing[3]. The HKGI will retrieve DNA from the samples for whole genome sequencing to obtain the genome data. Clinical data will also be collected from participants and analysed together with the genome data using a bioinformatics platform.

9. After analysis, the HKGI will send reports to the partnering centres for follow-up by healthcare personnel. Some of these reports may enable clinicians of the partnering centres to make more precise diagnoses, while some may indicate that no useful clinical information has been

[3] All participants are required to donate blood samples. Some are also required to donate saliva or tissue samples depending on their medical conditions.

derived from the current data analysis. A multi-disciplinary team including the attending clinician of the case will evaluate the report findings in detail and take clinical follow-up actions accordingly, such as conducting a confirmatory test or referring the case to other specialties. Attending clinicians and genetic counsellors of the partnering centres will explain the reports and the follow-up actions to participants. Given the rapid development in genomic medicine, the HKGI will re-analyse participants' data from time to time in light of the latest medical research outcomes, and will notify the partnering centres of new findings for follow-ups.

10. Making reference to similar overseas projects, genomic and clinical data collected under the HKGP will be de-identified and kept in a bioinformatics analysis platform set up by the HKGI for access by approved researchers, with a view to promoting the development of local medical research and international academic exchanges.

Personal Privacy Protection and Data Security

11. Personal privacy and data security are the prime consideration of the HKGP. All personal information will be collected, handled and used in strict accordance with the Personal Data (Privacy) Ordinance and protected with stringent measures. In formulating the HKGP implementation plan, the HKGI consulted the Privacy Commissioner for Personal Data to ensure compliance with relevant privacy protection requirements, and engaged independent third parties to conduct Privacy Impact Assessment and relevant information technology security risk assessment.

12. Whole genome sequencing will be performed by the Centre for PanorOmic Sciences of the HKU and the laboratory of the HKGI in the pilot phase. Genome data and related medical information so obtained will be encrypted and stored in the HKGI database. Only authorised designated HKGI staff and healthcare personnel will have permission to access and analyse participants' medical records and genome data through the bioinformatics analysis platform set up by the HKGI. Designed with

stringent security features, the platform has different levels of access right set according to the clinical duties and operational needs of dedicated staff. All data must be de-identified before they can be used for research purpose. To access and analyse such data through the platform, qualified research institutions and related personnel must obtain prior approval from the Research Ethics Committee.

13. The HKGP database is wholly owned and administered by the HKGI, with security measures employed for data protection in accordance with locally and internationally recognised standards of information technology security. These measures include data encryption, user identification, fine-grained access control, audit log and a password policy. All identifiable personal data will only be kept in a database within Hong Kong.

Promoting Development of Innovation and Technology (I&T)

14. At present, Hong Kong researchers rely mainly on an internationally shared human genome database, which primarily contains Caucasian data, when conducting researches on genomic medicine and biotechnology. As genomic information and disease conditions vary across races, a lack of genome data of our population has impeded the development of such researches in Hong Kong.

15. Along with benefiting patients with more precise diagnoses and treatments, the HKGP will also establish a genomic and clinical database of the local population. This will enable researchers to carry out studies on, for example, cancer genomics, pharmacogenomics and other phenotype-genotype association specific to our population (especially the South China population) to better understand the causes of various hereditary diseases among the local population, thereby developing more effective screening, diagnosis and treatment methods. The large database and analysis platform of the HKGP are also conducive to the development of biomedical research, big data analysis and artificial intelligence technologies.

16. With the laboratory and data analysis centre located in the Hong Kong Science Park, the HKGI will achieve greater synergy with scientific research institutions thereat in such areas as biomedical technology and information and communications technology. The HKGI will strengthen cooperation with local universities and research institutions to nurture talents required for the development of genomic medicine (in particular genetic counsellors and bioinformaticians, who are currently in severe shortage) and provide relevant career opportunities. In future, the HKGI will also collaborate with overseas scientific research organisations on mutual access to de-identified data to enhance research outcome, and to strengthen Hong Kong's international status in such areas as genomic medicine and I&T. Local universities and I&T institutions welcome and have indicated strong support for the HKGP.

Way Forward

17. The pilot phase is expected to be completed in mid-2022. The main phase will commence afterwards based on the advice of the Scientific Advisory Committee. The whole project covering the whole genome sequencing of about 50 000 genomes is expected to be fully completed in 2025. In the long run, the HKGI can leverage on the HKGP to unleash the potential of genomic medicine, on the one hand establishing a genome database of the local population to enhance clinical application of genomic medicine for the benefit of patients and their families, and on the other hand promoting research in genomic medicine and related fields to facilitate future development of innovative scientific research in Hong Kong.

Advice sought

18. Members are invited to note the content of this paper.

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