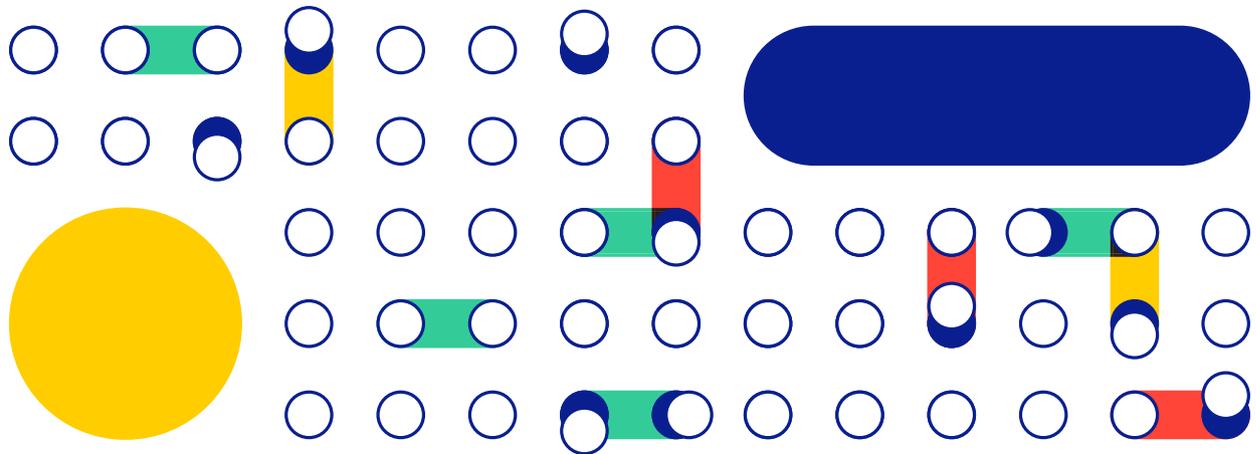


Strategic Development of
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MEDICINE IN HONG KONG



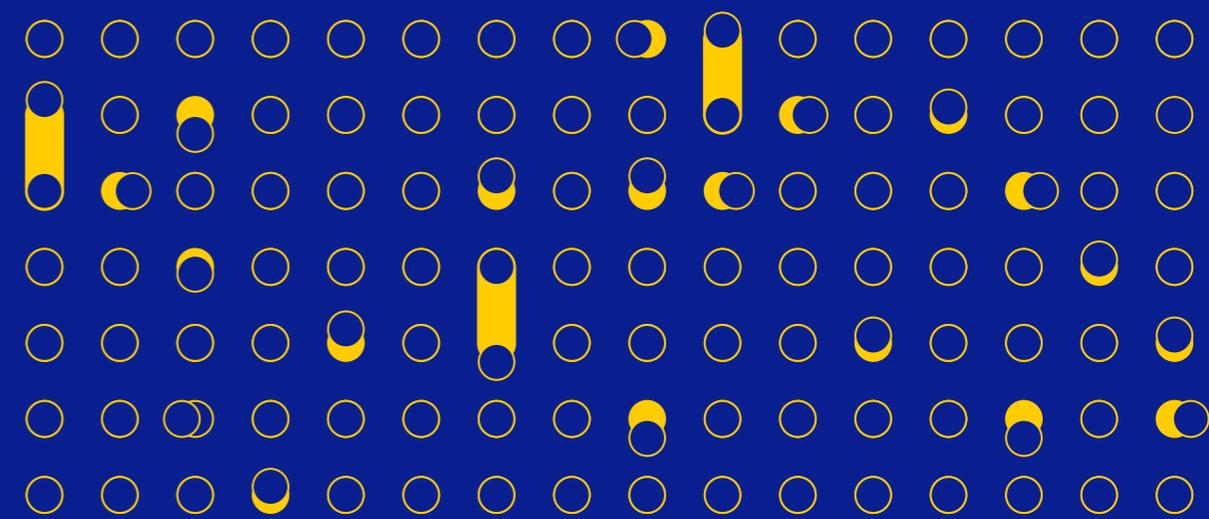


Strategic Development of
genomic
MEDICINE IN HONG KONG

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MESSAGE BY THE SECRETARY FOR FOOD AND HEALTH

Everyone's genome is unique. A human genome consists of 3.2 billion base pairs of A-T-C-G which make up our DNA. If printed on paper, they could fill a small library. While 99.9% of our DNA is the same as that of other individuals, the 0.1% variation makes up the differences between you and me – from skin colour, height, to facial features.

The genome determines our physical characteristics. Besides, it provides information why we are prone to certain diseases and what treatment is more effective to us. As a result of the rapid technological advancement in recent years, scientists have gained a deeper understanding of the structure and function of our genes and genome. Deciphering the relationship between genomic variations and diseases would lay the foundation for personalised or precision medicine, and hence improves diagnosis, prognostication and treatment of various diseases.

The Government is determined to promote local development of genomic medicine, so that our patients could benefit from more precise diagnosis and more effective treatment. In order to achieve this goal, we need to adopt a multi-pronged approach, which includes enhancement of clinical service, expediting translation of research findings to clinical application, manpower training, strengthening regulatory measures on related privacy and safety issues, as well as enhancing public education.

To study the above issues and propose recommendations is by no means an easy task. I would like to extend my heartfelt gratitude to all members of the Steering Committee on Genomic Medicine and its Working Groups for their invaluable contribution to the discussion. I would particularly like to thank Professor Raymond LIANG, Chairman of the Steering Committee, for his vision and leadership guiding the work of the Steering Committee in the past two years.

I am sure you would find the report informative and useful. The recommendations therein would form the backbone for the concerted efforts of all parties to advance genomic medicine in Hong Kong. We hope that before long, genomic medicine will be integrated into our healthcare system, so that our community could benefit from the scientific advancement and achieve better population health.

Professor Sophia CHAN Siu-chee, JP

Secretary for Food and Health



PREFACE

Genomic information is getting more and more useful for clinical care, including diagnosis and making therapeutic decisions. Genome technology, such as rapid DNA sequencing and data analysis, has made great advances in recent years. This has allowed us to integrate effectively genomic data into formulation of disease surveillance, screening, prevention of diseases, and treatment plan.

Significant progress has been made in the DNA sequencing time, coverage and throughput. Coupled with the rapid drop in the cost of DNA sequencing, the application of genomic information in research and clinical practice is rapidly expanding, including the diagnosis of previously unknown genetic diseases and the identification of driver gene abnormalities which predispose one to cancers. The information may also help discover new treatments. Therefore, genomic medicine is playing an increasingly important role in our medical care and providing potentially great clinical benefits to our patients. Timely and precise diagnosis can be life-saving.

Groups of passionate clinicians and academic researchers in Hong Kong have been making major contributions to genomic medicine and they have gained international recognition. I am glad that the Government has decided to provide full support to these efforts. A Steering Committee was set up gathering local experts to discuss and formulate coordinated strategies to support the development of genomic medicine in Hong Kong. It has been my great pleasure to chair the Steering Committee discussion in the past two years.

I would like to take this opportunity to thank all members of the Steering Committee for their valuable contributions during the period. I believe this report will provide further insights to those who are promoting genomic medicine in Hong Kong and hopefully also interest the new comers.

With the enthusiastic support of all stakeholders, including the Government, the Hospital Authority, academia, professional bodies and the private sector, I am sure we will continue to work together towards making significant contribution to the knowledge and applications of genomic medicine, benefitting our patients more and building a better future for our community.

Professor Raymond LIANG Hin-suen, SBS, JP

Chairman, Steering Committee on Genomic Medicine





EXECUTIVE SUMMARY

The rapid advancement in genomic medicine has presented huge potential in accurate diagnosis, personalised treatment and efficient surveillance of diseases. 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.

The Steering Committee recognised that with the dedicated work of passionate clinicians and researchers in the past few decades, high quality clinical services and impressive research outcomes on genomic medicine have been developed organically benefiting many patients in Hong Kong. Meanwhile, the opportunities presented by recent scientific breakthrough require a clear policy to steer and coordinate the efforts of various institutions, notably the Department of Health (DH), the Hospital Authority (HA), local universities, professional bodies and the private sector, in order to bring the development of clinical application and research in genomic medicine in Hong Kong to the next level.

In particular, the Steering Committee identified the following key issues which ought to be addressed through a comprehensive strategy –

- (a) standardised clinical service provision;
- (b) more efficient laboratory services and translation of new technology to clinical use;
- (c) more healthcare professionals specialised in genomic medicine and enhancement of genomic literacy among general healthcare professionals; and
- (d) ethical, legal and social implications.

With valuable contributions from Members of the Steering Committee at a total of 17 meetings, eight recommendations were proposed in this report.

Recommendation 1 : Launching the Hong Kong Genome Project

In order to significantly advance the development of genomic medicine in Hong Kong, the Steering Committee recommended that a large-scale genome sequencing project should be introduced to serve as a catalyst and anchor for showcasing the clinical benefits, piloting related new policy measures, building up talent pool and testing clinical protocols. With reference to international experiences, such project can also provide the necessary data for researchers to conduct genomic researches focusing on local population, bringing benefits to patients of our community.

The Chief Executive announced in the 2018 Policy Address that the Government would launch the Hong Kong Genome Project (HKGP), which aims to sequence around 40 000 to 50 000 genomes in the next six years with a budget of \$1.2 billion allocated in the 2019-20 Budget. The Food and Health Bureau (FHB) plans to establish the Hong Kong Genome Institute (HKGI) to take forward the HKGP within 2020.

Recommendation 2: Enhancing clinical services in genetics and genomics

The Steering Committee supported HA's Strategic Service Framework for Genetic and Genomic Services announced in October 2019, which aimed to provide structured and coordinated clinical services in genetics and genomics in HA.

In particular, the Steering Committee recommended that designated posts for clinical geneticists, pathologists, genetic counsellors and bioinformaticians should be created in HA to establish clear career paths for these professions. In addition, the Hong Kong Children's Hospital, a tertiary referral centre for complex paediatric cases, should play a key role in enhancing service collaboration and research translation in genomic medicine among HA, the Clinical Genetic Service of DH and universities, and in serving as a major training ground for genetics and genomics. The genetic and genomic test formulary should be enhanced with clinical guidelines to ensure the standardised use of genetic and genomic tests, while the mechanism for evaluating the introduction of new genetic and genomic tests in clinical service should be as efficient and flexible as possible.

Recommendation 3: Nurturing talents in genomic medicine

The Steering Committee recommended that the Government should estimate the manpower requirement for professionals in genomic medicine to facilitate the planning of universities, and attract prospective students and overseas talents. Given the wide coverage of genomic medicine, the Hong Kong Academy of Medicine (HKAM) considered that genetics and genomics should be positioned as a subspecialty in some colleges such as paediatrics and pathology, while relevant training could be provided at certification or diploma level in some other colleges such as family medicine. The HKAM has been invited to coordinate with relevant colleges on enhancing training in genetics and genomics of clinicians, including issuing relevant ethical guidelines.

The Steering Committee recognised the important roles of genetic counsellors and bioinformaticians in genomic medicine, and recommended that universities and academic institutes should be encouraged to offer relevant postgraduate-level programmes which benchmark international qualifications.

The Steering Committee considered that clear career paths are the key to building up a talent pool for genomic medicine in Hong Kong. In this regard, HA and DH should actively explore the creation of dedicated posts for genetic and genomic services with clear promotional prospect to attract talents.

The Steering Committee recognised that the genomic literacy of clinicians, nurses and allied health professionals in general should be enhanced. The proposed HKGP will play a catalytic role in enhancing the genetic and genomic knowledge of healthcare professionals and attracting both local and international talents.

**Recommendation 4:
Enhancing public engagement in genomic medicine**

The Steering Committee considered that there is a need to enhance public engagement in the potential and limitations of genomics on healthcare decisions, the proper use of genetic and genomic tests, as well as the relevant ethical implications. FHB, together with DH, HKGI and other stakeholders, should tap on the opportunity of the HKGP to promote public understanding on the above issues.

**Recommendation 5:
Enhancing the laboratory network with effective referral mechanism and centralisation of advanced genetic and genomic tests**

The Steering Committee recommended that a referral mechanism on genetic and genomic tests should be established among laboratories in HA, DH, universities and the private sector, and the provision of advanced genetic and genomic tests supported by public funding should be centralised where appropriate. The proposal could synergise expertise, unify laboratory standards, optimise the use of public resources and allow patients to have more equitable access to appropriate genetic and genomic tests.

**Recommendation 6:
Facilitating the establishment of a biobank network for genomic research**

The Steering Committee considered that instead of establishing a centralised biobank, the Government should explore measures to facilitate the establishment of a network of biobanks across institutions, in order to enable the sharing of de-identified biospecimens and associated data for future genomic research. To this end, there is a need to develop a standard protocol on consent-taking, protection of privacy, etc. which will be piloted in the HKGP. In order to provide incentives, the Steering Committee recommended that the Government should consider making participation in the biobank network as one of the conditions for future genomic research projects to receive government funding (e.g. the Health and Medical Research Fund).

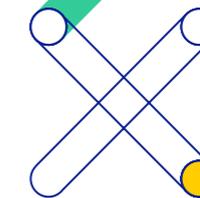
**Recommendation 7:
Enhancing the regulation on use of genetic data for insurance and employment purposes**

The Steering Committee noted that in addition to the Disability Discrimination Ordinance and the Personal Data (Privacy) Ordinance, more specific regulatory measures on genetic discrimination in the context of insurance should be explored. In view of the rapid development of genomic medicine, it would be more pragmatic to first explore the updating of the relevant Code of Practice issued by the Hong Kong Federation of Insurers (HKFI). In updating the Code, HKFI should be invited to take into account the mainstream international practices, with due regard to the local circumstances. The Steering Committee recommended that the Government should continue to review the situation and consider introducing legislative measures as and when necessary. Meanwhile, public education should be enhanced on anti-genetic discrimination for employers and employees.

**Recommendation 8:
Promoting the proper use of genetic and genomic tests**

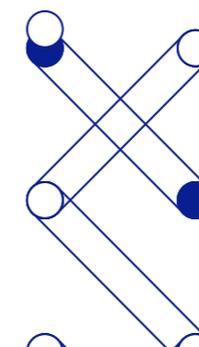
The Steering Committee considered that as direct-to-consumer genetic tests (DTCGTs) have become popular outside the clinical setting, there is a need to introduce suitable measures to address related healthcare and ethical issues. Since many of the DTCGTs could be obtained overseas through online purchase, the most pragmatic approach is to enhance public education on making informed decisions. In particular, consumers should be advised to consult healthcare professional, fully understand the clinical validity and utility of the tests, the privacy and ethical implications, as well as the limitations of the genetic test results, before taking the tests.

For health-related genetic tests, the Steering Committee considered that additional regulatory measures should be introduced, as a test result without proper counselling and professional advice may cause anxiety or false reassurance and would in turn lead to unnecessary investigations or delayed health care. The Steering Committee noted that the Government has been working on a statutory control framework for medical devices, which would cover human genetic tests in vitro that meet the definition of medical devices. Review on the professional qualifications for conducting genetic and genomic tests should also be conducted.

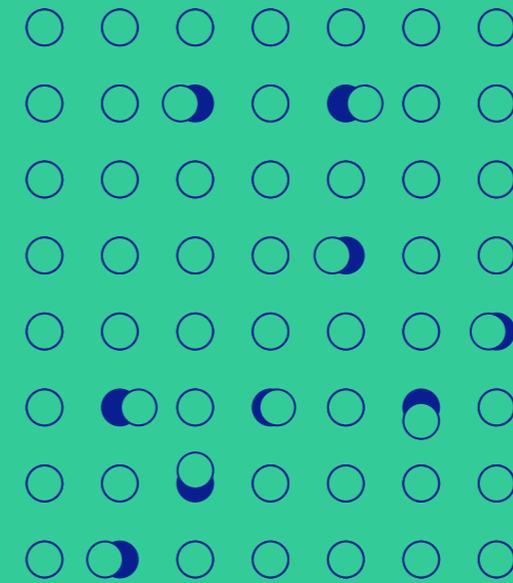


Conclusion and Way Forward

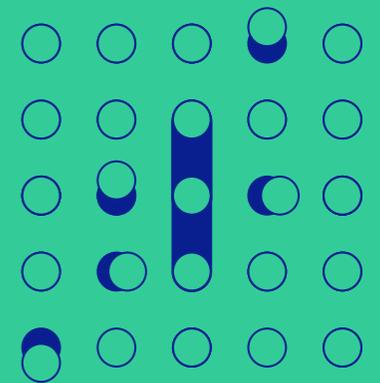
Genomic medicine is shaping the future medical development around the world rapidly. Meanwhile, it also brings about unprecedented challenges in the realms of ethics and privacy. The Steering Committee urged the Government and relevant stakeholders to take forward the eight recommendations with reference to international standards and developments, while taking into account the unique local context. In particular, the Steering Committee looked forward to the introduction of the HKGP, which would serve as a catalyst to pilot and implement the recommendations and pave way for maximising the benefits of genomic medicine in Hong Kong.



CHAPTER 1



CURRENT LANDSCAPE OF GENETICS AND GENOMICS IN HONG KONG



BACKGROUND

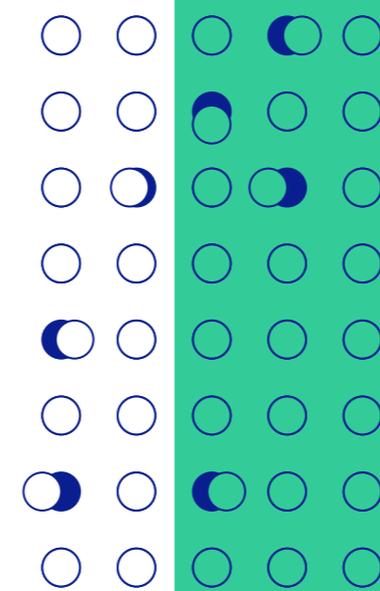
The Chief Executive announced in her 2017 Policy Address the setting up of a steering committee to lead the study on strategies for developing genomic medicine in Hong Kong. Subsequently, the Secretary for Food and Health appointed the Steering Committee on Genomic Medicine (Steering Committee) in December 2017. It is chaired by Professor Raymond LIANG Hin-suen, with members drawing from the academia, professional bodies and experts in different aspects of genomic medicine. The Membership and Terms of Reference of the Steering Committee are at **Annex A**.

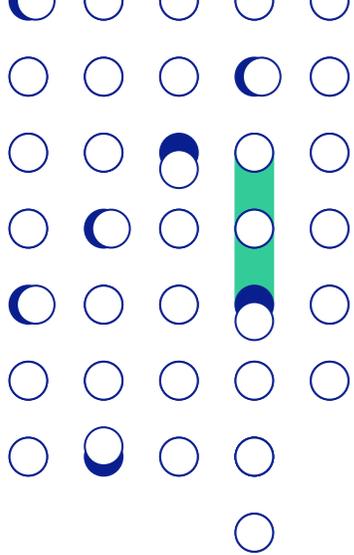
In addition to discussion at Steering Committee level, to allow for more focused study on specific topics, the following three working groups were established –

- (a) Working Group on Biobank
- (b) Working Group on Laboratory Network
- (c) Working Group on Hong Kong Genome Project.

Membership of the Working Groups is at **Annex A**.

The Steering Committee and its three working groups held a total of 17 meetings from January 2018 to November 2019. Detailed discussion and recommendations are set out in this report.





Chapter 1

CURRENT LANDSCAPE OF GENETICS AND GENOMICS IN HONG KONG

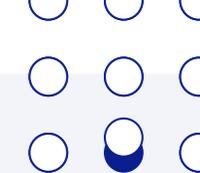
- 1.1** Clinicians at the Department of Health (DH), the Hospital Authority (HA), universities and private hospitals have been providing high quality clinical genetics service to the general public in Hong Kong in the past decades. These services include diagnosis of genetic disorders, genetic screening, genetic counselling and genetic testing in relation to disease management. At the same time, local academic researchers have attained internationally recognised achievements in genomic research. This Chapter provides an overview of the current landscape of services and researches in genetics and genomics in Hong Kong.

DH

- 1.2** The Clinical Genetic Service (CGS) of DH, led by a Consultant Clinical Geneticist, has been the main provider of public genetic and genomic services on a territory-wide basis in Hong Kong since 1981. There are four types of genetic and genomic services provided by the CGS, namely genetic counselling service, genetic screening service, genetic laboratory service and genetic health promotion. As the CGS does not offer treatment services, patients with treatment needs will be referred to respective specialties at HA for follow-up management. CGS handles about 1 800 new cases every year, with a cumulative caseload involving 38 000 families since its establishment.
- 1.3** The genetic counselling service of CGS is delivered through three genetic counselling clinics where the patients or families are assessed and provided with appropriate genetic testing to substantiate the genetic diagnosis. Genetic counselling aims to help patients or families with genetic diseases or rare syndromal disorders to understand the hereditary nature, the mode of inheritance, the recurrence risk of the disease / disorder within the family, and any preventive measures in order to make informed decisions. If clinically indicated, expectant mothers would be referred to prenatal diagnostic clinics of public hospitals for prenatal diagnostic procedures. From 2014 to 2018, an annual average of about 5 100 patients / families attended the genetic counselling clinics.
- 1.4** On genetic screening service, CGS has been providing free-of-charge neonatal screening service for Congenital Hypothyroidism (CHT) and Glucose-6-Phosphate Dehydrogenase (G6PD) Deficiency since 1984, and is responsible for the overall planning, coordination and evaluation of the Newborn Screening

Programme for Inborn Errors of Metabolism (IEM), which was regularised in 2017 and is being extended to all public hospitals in phases.

- 1.5** The genetic laboratory in CGS is the first genetic laboratory accredited under the Hong Kong Laboratory Accreditation Scheme (HOKLAS) in Hong Kong. It provides a variety of genetic tests, both cytogenetic and molecular, to cater for the needs of the genetic counselling team for disease confirmation or prediction. CGS is also one of the two accredited training centres for doctors to become specialised in Genetics & Genomics (Paediatrics).
- 1.6** CGS conducts public education on genetic diseases and genetic health through lectures and talks, educational pamphlets and media interviews. It is the Government's health advisor on the aspect of genetic and genomic medicine.
- 1.7** While CGS has had frequent collaborations with universities to bring the most advanced genetic technologies like chromosomal microarray, whole exome sequencing and bioinformatics platform into clinical application, there is also room for further collaboration with universities to facilitate transfer of high quality research into clinical practice.
- 1.8** In December 2019, the CGS moved to the premises of the Hong Kong Children's Hospital (HKCH). More collaboration is expected with HA's future genetic and genomic services. There is also potential for further collaboration with various specialties of HA in order to benefit more patients.



HA

- 1.9** There is no dedicated clinical department providing genetic and genomic services in HA. Instead, HA's genetic and genomic services have mostly developed at the local level and on an independent basis by individual clinicians or hospitals according to the interest and expertise of the respective staff and local needs, and have largely been driven through laboratory initiatives. As a result, there is wide variation in the types and complexity of genetic and genomic services provided across all seven Clusters, and overall the provision in HA is very limited.
- 1.10** There are limited standard protocols or test criteria for genetic and genomic services in HA. Referral practices rely heavily on the informal links between individual medical practitioners or departments, giving rise to inequitable access to the services. In 2015, HA launched the Genetic Test Formulary to provide a directory of genetic tests available in the HA and the DH. Departments such as Pathology and Obstetrics and Gynaecology (O&G) work closely with the clinical teams to provide these genetic tests to HA patients, while the follow-up services for ambiguous or abnormal results are mostly provided by the clinical team of respective specialties.
- 1.11** There are no clinical geneticists in HA. Training programmes on genetics and genomics such as genetic counselling courses are provided for clinicians and other healthcare professionals under the existing training framework. There is currently no evaluation mechanism for translating research-based services into HA services, thus hindering the transfer of well-evidenced services or tests from the academic sector to HA's mainstream clinical practice in a timely manner. To meet the service gaps of genetics and genomics as described in the above paragraphs, HA has issued the Strategic Service Framework for Genetic and Genomic Services (SSF GGS) on 14 October 2019, as detailed in Chapter 4.

UNIVERSITIES

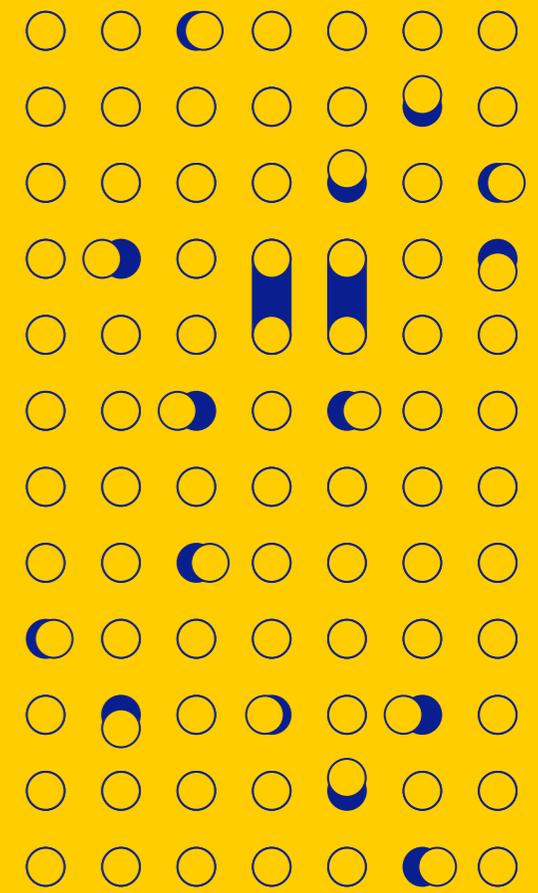
- 1.12** The two medical schools of the University of Hong Kong (HKU) and the Chinese University of Hong Kong (CUHK) have been playing an indispensable role in providing clinical services and advancing research in genetics and genomics. They have also been training relevant healthcare professionals to support genetics and genomics.
- 1.13** At present, HKU partners with the Hong Kong West Cluster of HA to provide clinical genetic services to patients under a combined model of patient care, teaching and research. For instance, the Hereditary Gastrointestinal Cancer Genetic Diagnosis Laboratory provides charitable genetic diagnosis service and maintenance of family cancer registry, receiving territory-wide referrals. At the same time, the Hong

Kong Hereditary Breast Cancer Family Registry supports genetic testing and counselling for individuals with higher risk of developing breast, ovarian and other cancers. Other services include pre-implantation genetic testing, clinical genetics with focus on paediatrics diseases and immunodeficiency, cancer target gene panel testing, etc. The Centre for Genomic Services in HKU also provides a wide range of DNA sequencing services to support genomics and proteomics research.

- 1.14** CUHK partners with the New Territories East Cluster of HA to provide genetic testing services for a wide range of cancer diseases such as colorectal cancer, breast cancer, ovarian cancer and brain cancer. It also provides pre-implantation genetic screening and diagnosis, prenatal genetic screening and diagnosis and expanded newborn screening for IEM. The O&G Department of CUHK partners with The Baylor College of Medicine in the United States to provide one-stop diagnosis, consultation, genetic counselling, risk assessment and testing services in the Prince of Wales Hospital. Among other research achievement, CUHK introduced the clinical service for non-invasive prenatal diagnosis of Down syndrome in 2011. In the area of cancer detection, CUHK has pioneered a number of approaches to cancer liquid biopsy, such as the detection of nasopharyngeal carcinoma and genome-wide approaches for screening multiple types of cancer.
- 1.15** Other universities including the Hong Kong University of Science and Technology (HKUST) also participate actively in scientific research on genomic medicine, with recognised achievements in areas such as Alzheimer Disease and brain tumour. The Research Grants Council under the University Grants Committee, the Health and Medical Research Fund and the Innovation and Technology Fund have been supporting local universities to conduct various research projects on genomic medicine.

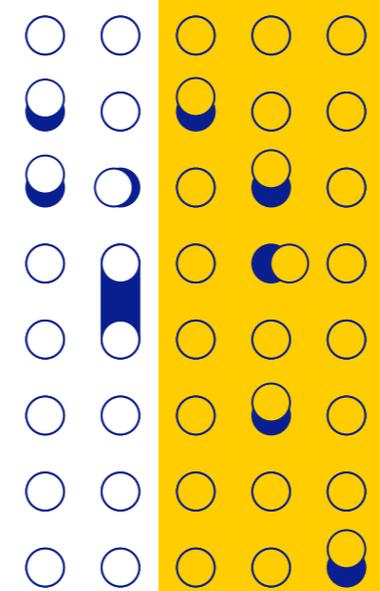
PRIVATE SECTOR

- 1.16** In addition to the public sector and universities, part of the increasing demands for clinical genetic service have been met by the private sector. In 2016, a private hospital in Hong Kong set up its own Clinical Genetics Service, consisting of regular genetic diagnostic and counselling clinics, supported by an in-house laboratory with cytogenetics, molecular genetics, biochemistry and haematology capacity. The service works closely with the Reproductive Centre in the hospital which provides prenatal and pre-implantation component for genetic testing.
- 1.17** At the same time, direct-to-consumer genetic tests (DTCGTs) have become more and more prevalent. Some of the tests can be ordered online from overseas suppliers or obtained from retail outlets without prescription. There has been rising concern on the validity of such tests and the proper handling of the test results.



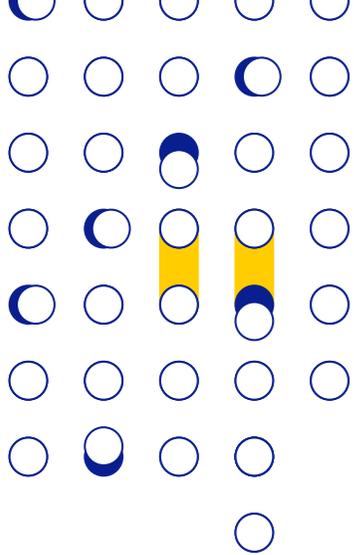
CHAPTER 2

KEY ISSUES TO ADDRESS



SUMMARY

- 1.18** The Steering Committee reviewed the current landscape of genetics and genomics in Hong Kong. It concluded that with the contribution of passionate clinicians and researchers, DH, HA, universities and the private sector have been providing high quality clinical services and producing impressive research outcomes on genomic medicine in the past few decades.
- 1.19** Notwithstanding the achievements of individual services and researchers, the Steering Committee considered that a clear policy to steer and coordinate efforts across different sectors will significantly boost the development of genomic medicine in Hong Kong. As such, there is a need for the Government to formulate a comprehensive strategy on the multi-faceted issues in order to bring the development of genomic medicine to the next stage for the benefit of the community in the long run.



Chapter 2

KEY ISSUES TO ADDRESS

- 2.1 The Steering Committee recognised that in order to promote the future development of genomic medicine in Hong Kong, a number of key issues had to be addressed with concerted efforts of all parties. This Chapter outlines the key issues discussed, which are systemic in nature and require a comprehensive strategy to tackle.

CLINICAL SERVICE PROVISION

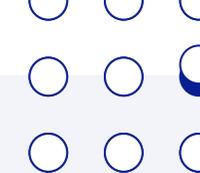
- 2.2 As mentioned in Chapter 1, the development of genomic medicine in Hong Kong has been organic and sporadic, highly dependent on the individual efforts of passionate clinicians and academic researchers. For historical reason, CGS of DH provides the majority of public diagnostic and counselling service to families with inherited genetic disorders, while follow-up treatment is provided by HA. Meanwhile, there is no dedicated genetic and genomic services within HA. The clinical services provided by the two medical schools are research-based and not routine services. There is a need to explore a more standardised and coordinated clinical pathway for genetic and genomic services in Hong Kong, so that patients can access to clinical genetic and genomic services on a fair and equitable basis.

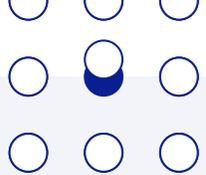
LABORATORY SERVICES AND TRANSLATION OF NEW TECHNOLOGY TO CLINICAL USE

- 2.3 Similar to clinical service provision mentioned in paragraph 2.2, the laboratories of DH, HA and universities have developed different sets of genetic and genomic tests according to their own need, capacity and resource, resulting in duplication and inefficiency in the utilisation of genetic tests. In addition, without a well-established mechanism to evaluate the clinical validity and utility of new genetic and genomic tests developed by different institutions, transfer of new technology into clinical use is severely hindered.

NUMBER OF SPECIALISTS AND LEVEL OF GENOMIC LITERACY AMONG HEALTHCARE PROFESSIONALS

- 2.4 The provision of genetic and genomic services requires the support of healthcare professionals providing diagnosis, counselling, testing and test interpretation. Overall speaking, Hong Kong lacks a sufficient pool of genetic and genomic professionals. As of October 2019, there are only five paediatricians accredited in the subspecialty of Genetics & Genomics (Paediatrics) under the Hong Kong College of Paediatricians, who manage not only children but also adult patients with genetic disorders. The College of Pathologists is in the process of admitting the first fellows in Genetic and Genomic Pathology.
- 2.5 The qualifications and training requirements for genetic counsellors, a well-established profession in advanced economies, are yet to be defined. At the same time, while bioinformaticians play an essential role in the delivery of genomic services, the pool of bioinformaticians is not enough to meet the demand in Hong Kong. The regulatory framework for Medical Laboratory Technologists also needs to be reviewed to take into account the latest development of genetic and genomic tests. More efforts should be devoted to the development of these professions, including the enhancement of training and establishment of clear career paths. The genetic and genomic literacy of healthcare professionals also needs to be enhanced to facilitate the application of genomic medicine in different clinical care settings.



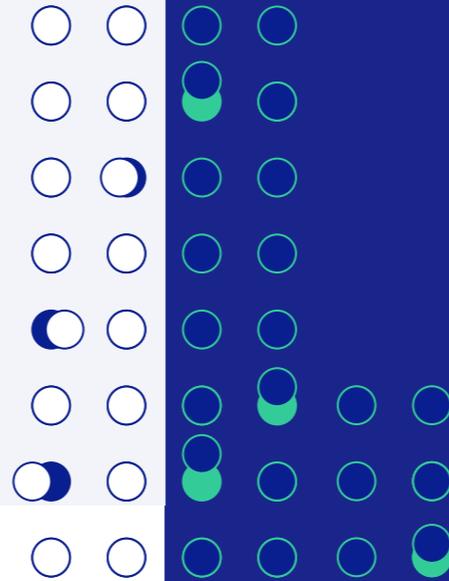


ETHICAL, LEGAL AND SOCIAL IMPLICATIONS

2.6 Genetic information is regarded as highly sensitive because of its potential to reveal an individual's traits and characteristics in addition to disease risks of an individual and his / her family. The diagnosis of a genetic condition and the prevalence of DTCGTs arouse concerns from ethical, legal and social perspectives. These concerns range from the analytical and clinical validity of a genetic test, to potential discrimination by insurers and employers based on genetic information. Proper regulatory and educational measures should be explored to address these issues to facilitate the development of both clinical services and researches on genomic medicine.

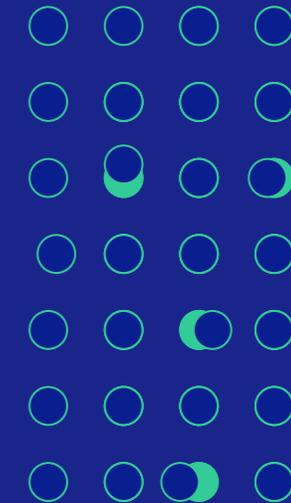
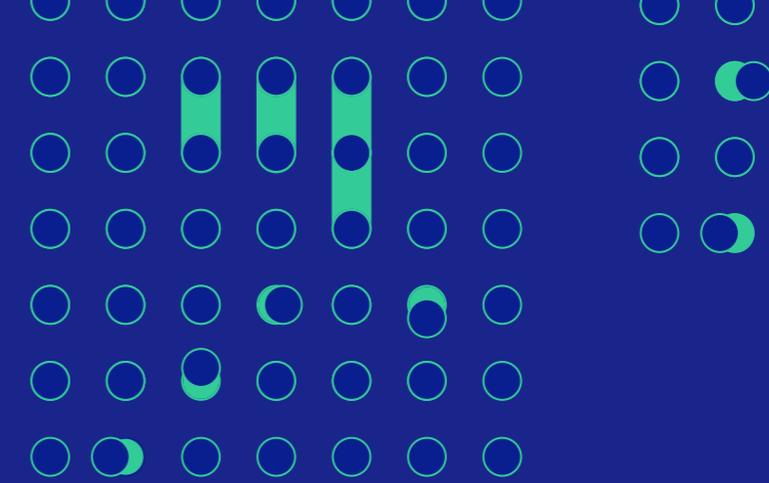
SUMMARY

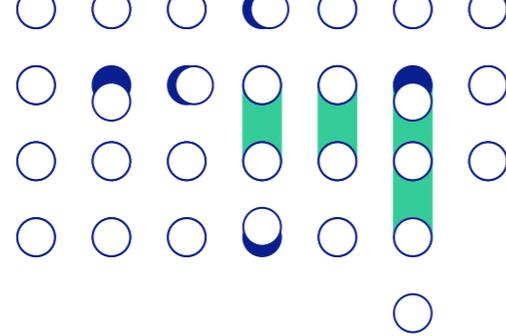
2.7 The Steering Committee deliberated on the above key issues thoroughly, before putting forward the recommendations listed in this report. In particular, the Steering Committee recommended that the Government should introduce a large-scale genome sequencing project so that different stakeholders could be pulled together to take forward the recommendations and work towards the common goal of maximising the benefits of genomic medicine. Details of the eight recommendations are set out in the subsequent chapters.



CHAPTER 3

LAUNCHING THE HONG KONG GENOME PROJECT





Chapter 3

LAUNCHING THE HONG KONG GENOME PROJECT

INTRODUCTION

- 3.1 With technological advancement in sequencing technology where the sequencing of a human genome could be completed in days at the cost as low as USD 1,000, there is an international trend to introduce large-scale genome sequencing project for clinical and scientific advancement. Genome projects of various scales and focuses are being carried out in the United Kingdom (UK), Singapore, Iceland, the United States (US), Israel, Finland, Ireland, etc. For instance, the UK has completed the sequencing of 100 000 genomes by 2018, while the All of Us Research Programme in the US is building a national research cohort of one million or more participants. A summary of the projects is at **Annex B**.
- 3.2 During the deliberation of the Steering Committee, Members unanimously agreed that a large-scale genome sequencing project should be launched to advance the development of genomic medicine in Hong Kong. The project can serve as a catalyst and anchor for showcasing the clinical benefits, piloting related new policy measures, building up talent pool and testing clinical protocols. With reference to international experiences, such project can also provide the necessary data for researchers to conduct genomic research focusing on local population, bringing benefits to patients of our community.
- 3.3 In view of the potential benefits, the Chief Executive announced in her 2018 Policy Address that the Government accepted the Steering Committee's preliminary recommendation and would introduce a large-scale sequencing project, to be named Hong Kong Genome Project (HKGP), with a view to enhancing clinical application of genomic medicine and promoting scientific research. The 2019-20 Budget further reserved \$1.2 billion to take forward the HKGP in six years.
- 3.4 In November 2018, a Working Group on HKGP, comprising experts from the academic, clinical and research sectors, was set up under the Steering Committee to discuss the project framework. The Membership of the Working Group is at **Annex A**.

DISCUSSION AND ANALYSIS

Policy objectives

- 3.5 The HKGP is a catalyst project to establish a genome database of the local population, a talent pool, as well as infrastructure and protocol for genetic and genomic testing, with the following policy objectives –
 - (a) Enhancing clinical application of genomic medicine to benefit patients and their families; and
 - (b) Promoting research in genomic medicine and related field to facilitate future medical development in Hong Kong.

- 3.6 In the longer run, the HKGP would become one of the largest health-related databases in Hong Kong. Clinical and genomic data would be collected at standardised format and processed through standardised bioinformatics pipelines. This could facilitate the use of genomic data for clinical management and research, and pave way for the mainstreaming of genomic medicine in our healthcare system. The HKGP also aspires to become an exemplar in formulating the data security and privacy protocols in the region that are commensurate with international standards.

Project scope

- 3.7 The Working Group recommended the project scope as follows –
 - (a) To perform 20 000 cases (or 40 000 to 50 000 whole genome sequencing¹) in two phases for a period of six years; and
 - (b) The pilot phase (2 000 cases or about 5 000 genomes) will cover patients with undiagnosed disorders, and cancers with clinical clues linked to possible hereditary components. The main phase (18 000 cases or 45 000 genomes) could expand the coverage to other diseases and research cohorts which could benefit from whole genome sequencing.

Coordination by the Hong Kong Genome Institute

- 3.8 The Government will set up the Hong Kong Genome Institute (HKGI), a company limited by guarantee wholly owned by the Government, to coordinate the implementation of the HKGP in partnership with Food and Health Bureau (FHB), DH, HA, universities, private hospitals, and the research and development sector.
- 3.9 The HKGI will be governed by a board of directors with representatives from the Government, HA, universities, private hospitals, experts and lay members, and supported by a full-time professional team. A number of expert advisory committees on science, data access, ethics and data privacy would also be established. The HKGI will set out the protocols of HKGP in consultation with stakeholders.

1. Since some cases may involve two or more samples (e.g. tumour and non-tumour samples for cancer cases) or family members of patients (a trio of patient and parents for undiagnosed disorders), depending on the clinical and research needs, it is estimated that the HKGP would sequence 40 000 to 50 000 genomes in total.

Clinical functions of Partnering Centres

- 3.10** Designated partnering centres would be set up at selected hospitals to perform the clinical functions of the HKGP. Eligible patients (and their family members if necessary) will be recruited as HKGP participants after giving informed consent. They will be invited to provide blood sample (or other samples as appropriate) and be interviewed during one of their regular appointments at the hospitals. The sequencing analysis results will be fed back to participants once available. They will receive genetic counselling and be followed up by relevant specialists as needed.
- 3.11** Considering that most of the clinical geneticists / medical professionals well versed with genetics and genomics are centralised in DH CGS and the two medical schools at present, the Working Group recommended that in the pilot phase, three partnering centres should be set up at the HKCH (where DH CGS is located) and the two teaching hospitals, namely Queen Mary Hospital (QMH) and Prince of Wales Hospital (PWH).

Use of HKGP data for research purpose

- 3.12** Apart from helping the clinical management of patients, another key feature of HKGP is the establishment of a large-scale database of anonymised genomic and clinical data for research. With the informed consent of HKGP participants, the anonymised genomic and clinical data would be pooled for access and analysis by approved researchers in a secure, monitored data environment for medical research purposes. Scientific, data access and ethics advisory committees will be formed to process and approve data access requests.
- 3.13** The data generated by the HKGP would present huge opportunities for investigation into diseases specific to our population. It can contribute to various studies on cancer genomics, pharmacogenomics, other phenotype-genotype association, etc. With the scaling up of the HKGP database, the HKGP will enable big data analysis for biomedical research and innovation, and will create synergies with the biomedical technology and the information and communications technology clusters accommodated in the Hong Kong Science Park. The HKGI would also collaborate with similar projects in the world to maximise the benefit of mutual access of database for research purposes.
- 3.14** The Working Group deliberated on a number of key principles in relation to the HKGP, including participant recruitment criteria, clinical workflow, informed consent, results feedback mechanism, data privacy, security and access. A summary of the key discussion is at **Annex C**.

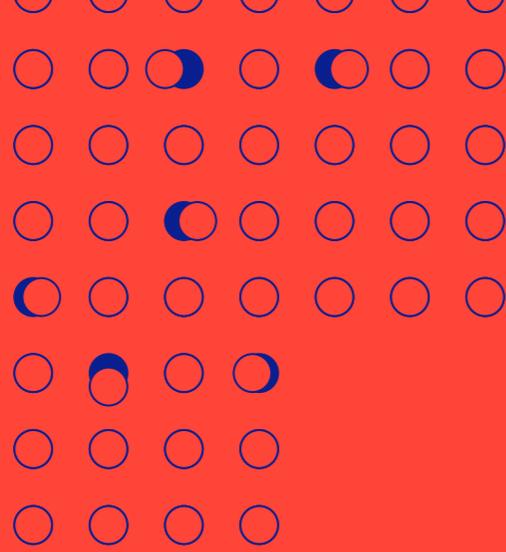
Recommendation 1:

LAUNCHING THE HKGP

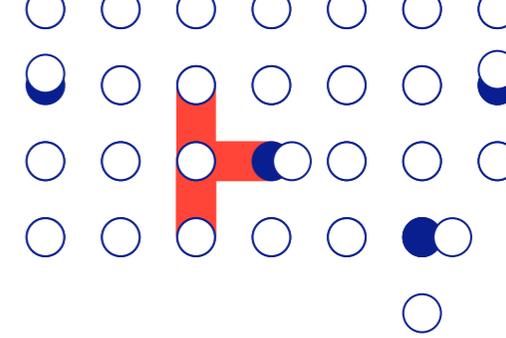
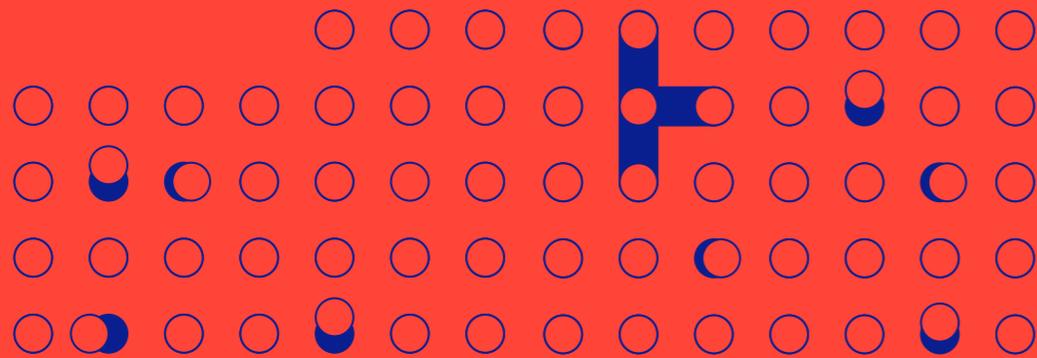
- 3.15** The Steering Committee recommended that the Government should take forward the HKGP as soon as possible in full consultation with relevant stakeholders. Through the HKGP, the genetic and genomic clinical service and research in Hong Kong would be able to excel to a new stage, where ample opportunities will be opened up for healthcare professionals and researchers to realise the potential of genomic medicine. In formulating the project details, the HKGI should take into account the discussions and suggestions of the Steering Committee and its Working Group.

WAY FORWARD

- 3.16** As announced in the 2019 Policy Address, FHB plans to establish the HKGI to launch the HKGP in 2020.



ENHANCING CLINICAL SERVICES IN GENETICS AND GENOMICS



Chapter 4

ENHANCING CLINICAL SERVICES IN GENETICS AND GENOMICS

INTRODUCTION

- 4.1 The CGS of DH is the main provider of public clinical diagnostic and counselling services to families with inherited disorders, while follow-up patient treatment and management is provided by HA. CGS accepts only patients by doctor's referral, with the majority of referrals coming from HA. In addition, CGS has been collaborating with HA to expand the newborn screening programme to cover inborn errors of metabolism in addition to CHT and G6PD Deficiency since 2017.
- 4.2 There is no dedicated clinical department providing genetic and genomic services in HA. HA's genetic and genomic services have mostly developed at the local level and on an independent basis by individual clinicians or hospitals according to the interest and expertise of the respective staff and local needs, and have largely been driven through laboratory initiatives. As a result, there is wide variation in the types and complexity of genetic and genomic services provided across all seven Clusters. The specialties that are key providers of genetic and genomic services include O&G, paediatrics, clinical oncology, and medicine. The services mainly cover prenatal genetics, cancer care, certain hereditary diseases and pharmacogenetics, in a limited scope. There is a formulary of genetic tests provided in HA hospitals and DH CGS, but the use of tests and clinical pathways vary among hospitals and clinicians.
- 4.3 Some clinical genetic and genomic services that are not available in HA and DH CGS are provided by the two medical schools, such as services for hereditary breast, ovarian and colorectal cancers as well as paediatric genetic disorders. Such services are funded by research grants, private grants or paid by patients themselves. A small portion of patients also receive genetic and genomic services in the private sector.

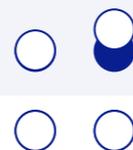
DISCUSSION AND ANALYSIS

HA

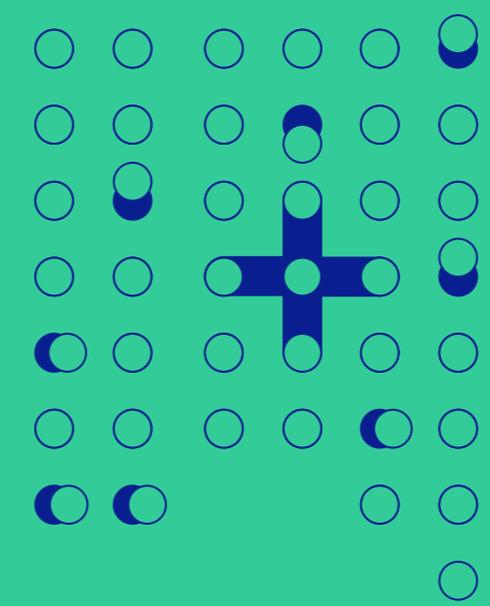
- 4.4** The Steering Committee noted that the provision of genetic and genomic service in HA is not standardised across clusters. To address the current gap as regards provision of genetic and genomic services, the Steering Committee considered that there is a need to strengthen the coordination and collaboration within HA to optimise the use of public resources and enhance equitable access to genetic and genomic tests.
- 4.5** The Steering Committee considered that it is important to have a transparent and efficient mechanism to evaluate and expedite the translation of new tests or technology from research setting into clinical practice. Such mechanism can prioritise public resources for introducing tests with proven effectiveness, cost-effectiveness and local needs. There is also a need to standardise the testing pathway and control on the utilisation of genetic and genomic tests by providing clinical guidelines for clinicians to prescribe appropriate genetic and genomic tests to specific persons or groups of persons under specific clinical scenarios.
- 4.6** The Steering Committee noted that the existing funding mechanism for introducing new genetic and genomic tests to HA through annual budget cycle is not effective, given the long lead time to secure resources and other competing priorities within HA. There is a need for a separate funding regime to support the development of genetic and genomic services, particularly for genetic and genomic tests on uncommon diseases where it is difficult if not impossible to gather sufficient clinical evidence to support routine implementation. The new regime should also allow flexibility for running novel tests with potential to bring significant benefits to patients on a pilot basis to expedite their adoption. The Steering Committee reviewed the mechanisms leveraged in other jurisdictions and the key findings are summarised at **Annex D**.
- 4.7** In tandem with the discussion of the Steering Committee, HA also formulated a SSF GGS, covering issues related to the governance structure for overseeing genetic and genomic service development, organising services based on a tiered approach and a hub-and-spoke model where specialised services will be centralised and provided by the “hubs” and routine services will be provided by the “spokes” to enhance the coordination and collaboration of different services, expediting the introduction of new genetic and genomic tests, etc. The views of the Steering Committee had been conveyed to HA for consideration in the formulation of the SSF GGS, which was issued on 14 October 2019. The executive summary of the SSF GGS is at **Annex E**.

DH CGS

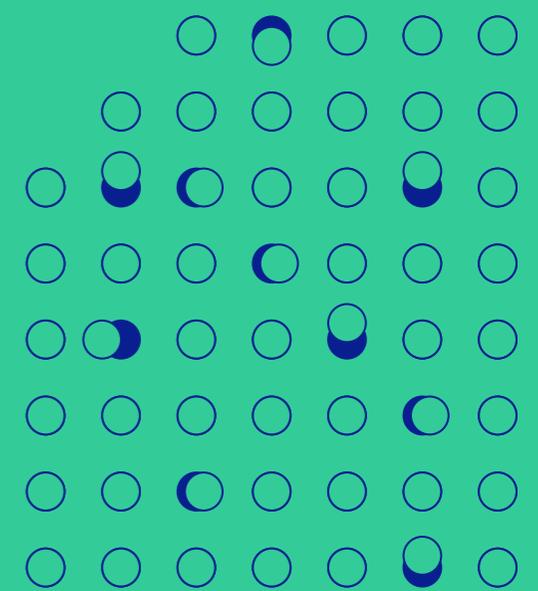
- 4.8** While DH CGS has had frequent collaborations with universities to bring the most advanced genetic technologies like chromosomal microarray, whole exome sequencing and bioinformatics platform into clinical application, and has all along been providing high quality clinical genetic service to the general public, the collaboration with various services of HA could be strengthened to benefit more patients. There is also room for further collaboration with universities to facilitate transfer of high quality research to clinical practice. In addition, any plan for service enhancement or expansion in DH CGS has to go through the annual resource allocation exercise within the Government. This could impede the timely expansion of the necessary workforce and the adoption of new technologies, resulting in long turnaround time of established tests which cannot meet the increasing service demands (i.e. about 1 800 new cases every year).
- 4.9** To foster closer collaboration and synergy with specialists and scientists from HA and universities, CGS has moved to the premises of HKCH in December 2019. Such co-location presents an opportunity for DH CGS to enhance service collaboration with various services of HA and participate in the development of genetic and genomic services of the HKCH.



CHAPTER 5



NURTURING TALENTS IN GENOMIC MEDICINE



Recommendation 2:

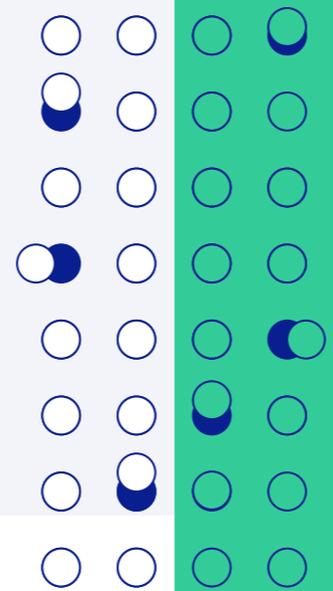
ENHANCING CLINICAL SERVICES IN GENETICS AND GENOMICS

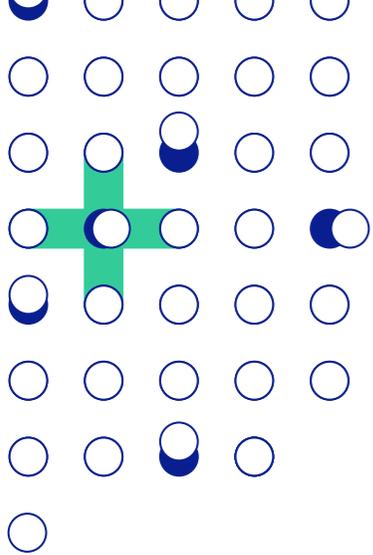
4.10 The Steering Committee welcomed HA's introduction of the SSF GGS, which had covered the concerns raised by the Steering Committee in paragraphs 4.4 to 4.6 above. In implementing the SSF GGS, the Steering Committee invited HA to take into account the following specific suggestions and views –

- (a) Clear career paths for relevant professionals, in particular clinical geneticists, genetic counsellors and bioinformaticians, should be established in HA by the creation of designated posts;
- (b) The HKCH should play a key role in enhancing service collaboration and research translation in genomic medicine among HA, DH CGS and universities. It should also serve as a major training ground for clinical geneticists and other healthcare professionals in genomics;
- (c) The genetic and genomic test formulary should be enhanced to ensure the standardised use of genetic and genomic tests by including clinical indications, restriction on which medical specialties can order the test, etc.; and
- (d) The mechanism for evaluating the introduction of new genetic and genomic tests to clinical service should be as efficient and flexible as possible. A designated fund should be established to expedite the introduction of new genetic and genomic tests for patient benefits.

WAY FORWARD

4.11 FHB would follow up with HA in the implementation of the SSF GGS with reference to the suggestions of the Steering Committee. Meanwhile, FHB would continue to work with DH CGS, HA and universities to map out the detailed collaboration model in HKCH.





Chapter 5

NURTURING TALENTS IN GENOMIC MEDICINE

INTRODUCTION

- 5.1 Genomic medicine is a specialised field which requires dedicated training and experience. The Steering Committee recognised that the key to enhancing the application of genomic medicine in clinical care and research is to build up a talent pool of clinical geneticists, pathologists, genetic counsellors, bioinformaticians and laboratory scientists. In addition to dedicated expertise, the genetic and genomic literacy of general healthcare professionals should also be enhanced in view of the growing relevance of genomic medicine in a wide spectrum of clinical care services.

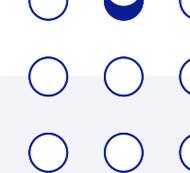
DISCUSSION AND ANALYSIS

- 5.2 The Steering Committee recognised that while universities and the Hong Kong Academy of Medicine (HKAM) have been developing relevant teaching programmes and subspecialties in recent years, the pool of experts in the field of genomics in Hong Kong is still not big enough for the long term development of genomic medicine.

Clinicians

- 5.3 Clinical geneticists are clinicians specially trained in genetics and genomics, who play a major role in diagnosing and managing genetic conditions or disorders, and help people who are affected by, or at risk of, inherited conditions to make informed health choices. In some countries, clinical geneticist is an independent specialty. Locally, the Hong Kong College of Paediatricians established the subspecialty in Genetics and Genomics (Paediatrics) in November 2017. As of October 2019, there are five paediatricians accredited in this subspecialty, and three paediatricians are undergoing training. Among the five clinical geneticists, two are serving in DH CGS, two at universities and one in private sector. There is currently no clinical geneticist in HA.
- 5.4 Pathologists' expertise are essential to the implementation of molecular testing and interpretation of the results. Elements of genetics and genomics are already part and parcel of all current specialty programmes under the College of Pathologists. The College of Pathologists established a new specialty in Genetic and Genomic Pathology in July 2019. The first round of fellowship would be conferred in due course.

- 5.5 In addition to training of specialists in genomic and genetics, the Steering Committee acknowledged that there is a need to enhance the genomic and genetic literacy of clinicians in general, as the fields of genomics are diffusing into different clinical care settings. At present, basic elements of genetic and genomic medicine are covered in the curricula of Bachelor of Medicine and Bachelor of Surgery programmes in two medical schools. At specialty level, the College of Paediatricians and the College of O&G have been providing genetic and genomic-related training to clinicians on a regular basis.
- 5.6 Meanwhile, HA also offers overseas corporate scholarship program on specialist training for medical professionals, and organises lectures and short term courses on genomics through Central Commissioned Training Programmes.



Genetic Counsellors

- 5.7** A genetic counsellor is a health professional with specialised training in medical genetics and psychosocial counselling. An experienced genetic counsellor is able to undertake some of the roles of a clinical geneticist. At present, there is no statutory professional regulation for genetic counsellors in Hong Kong.
- 5.8** In HA, a number of doctors and nurses have received basic training in genetic counselling, and are undertaking the role of genetic counsellors to provide genetic counselling prior to referral of patients to clinical geneticists of the CGS of DH or universities for further assessment. The follow-up services for ambiguous or abnormal results are mostly provided by the clinical team of respective specialties, for example, Down syndrome screening by obstetric team. HA also collaborated with HKU School of Professional and Continuing Education on a training programme on genetic counselling for nurses working in paediatric and O&G settings in 2015, 2016 and 2017.
- 5.9** In view of the growing demand for genetic counsellors, local universities have started to offer a number of post-degree programmes with elements of genetic counselling in recent years. Details are summarised in **Annex F**.

Bioinformaticians

- 5.10** Bioinformaticians play an important role in interpreting and analysing genomic data. It requires multidisciplinary knowledge in biomedical sciences, biochemistry, computer sciences, etc. and requires special training.
- 5.11** At DH CGS, genome data analysis and interpretation is solely carried out by clinicians without support of bioinformaticians. In HA, a bioinformatician was recently recruited as a Scientific Officer (Medical) and is undergoing training to support non-invasive prenatal testing service at the HKCH. A number of bioinformaticians are also assisting university academics in genomic research, while some are working in private companies providing genome data analytic services.
- 5.12** While some bioinformaticians receive their training at work, local universities also offer relevant programmes. Details of the programmes are summarised at **Annex F**.

Biomedical scientists / laboratory scientists

- 5.13** The operation of a molecular genetic laboratory requires specific requirements on the testing procedures in addition to the general requirement from medical testing laboratories. Graduates of biomedical sciences and medical laboratory science with knowledge and training on molecular biology techniques and genomic science thus play an indispensable role in developing and providing genetic and genomic tests at local laboratories. Details of the programmes are summarised at **Annex F**.

Genomic and genetic literacy of other healthcare professionals

- 5.14** The Steering Committee acknowledged that there is a need to raise the awareness of other healthcare professionals on genetics and genomics. At present, HA offers training of various duration on genetics and genomics to healthcare professionals in general. HA also organises lectures and short term courses on genomics. To widen the impact on other healthcare professionals, genomic medicine service was selected as one of the major themes of the HA Convention 2019 to raise the general awareness of HA staff on the subject.

Recommendation 3:

NURTURING TALENTS IN GENOMIC MEDICINE

- 5.15** The Steering Committee recommended the following five key measures to build up a comprehensive talent pool for genetics and genomics professionals in Hong Kong

Manpower requirement estimation

- 5.16** As the first step, the Steering Committee recommended that the Government should estimate the manpower requirement for clinical geneticists, pathologists, genetic counsellors, bioinformaticians and medical laboratory scientists, etc. in short, medium and long term, taking into account the future development of genetic and genomic services in the public and private sector. The estimation of manpower requirement would facilitate universities to plan for suitable programmes / curriculum, the assessment of justifications for public funding on such programmes, and attracting overseas talents. This can also provide useful reference for prospective students and professionals to consider their career planning.



Specialty development and training for clinicians

- 5.17** At the invitation of the Steering Committee, HKAM consulted relevant colleges (i.e. College of Obstetricians and Gynaecologists, College of Pathologists, College of Physicians and College of Radiologists) on specialty development and training for genetic and genomic medicine. All consulted colleges proposed that given the wide coverage of genomic medicine, instead of establishing an independent specialty of genetics and genomics, it would be more appropriate for relevant colleges to develop their own subspecialty or enhance training in genetics and genomics under the established mechanism. The Steering Committee agreed that given the insufficient existing expertise and uncertainty over career prospect, it would be more pragmatic to position genetics and genomics as a subspecialty in individual colleges at present stage. For some colleges such as family medicine, genetic and genomic training could be provided at certification or diploma level instead of subspecialty level.
- 5.18** The Steering Committee considered that for the relevant colleges, pre-fellowship training should provide baseline genetic and genomic knowledge, while the post-fellowship training should focus on special skills or intercollege training. Ethics training and guidelines should also be considered. For family medicine, focus should be put on genetic counselling at primary care level to equip family physicians with the ability to identify potential cases for referral to clinical genetic services. The Steering Committee noted that HKAM organised a workshop on the development of best practice guidelines in Genetic and Genomic Medicine in November 2019, and invited HKAM to continue coordinating with relevant colleges on enhancing training in genetics and genomics of clinicians.

Provision of relevant postgraduate programmes

- 5.19** According to international practices, genetic counsellors and bioinformaticians usually require postgraduate level qualifications. In the UK, the US, Australia and Canada, genetic counsellor who holds a master degree with relevant experience is registered or certified under the registration board. A brief outline of the international practices on the qualifications of genetic counsellors is at **Annex C**. For the long term development of these two relatively new professions and other related disciplines in Hong Kong,

the Steering Committee agreed that there is a need to encourage and support universities / academic institutes to offer relevant postgraduate-level programmes which benchmark international qualifications.

Establishment of clear career paths

- 5.20** The Steering Committee highlighted the importance of clear career paths in attracting potential talents to pursue their career in genomic medicine. It recommended that HA and DH should actively explore the creation of dedicated posts for genetic and genomic services (e.g. clinical geneticists, pathologists, genetic counsellors and bioinformaticians) with clear promotional prospect. The proposed HKGI would also provide career opportunities for professionals in this field.

Enhancement of genomic literacy among healthcare professionals

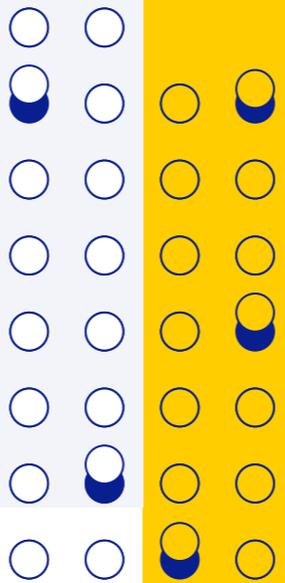
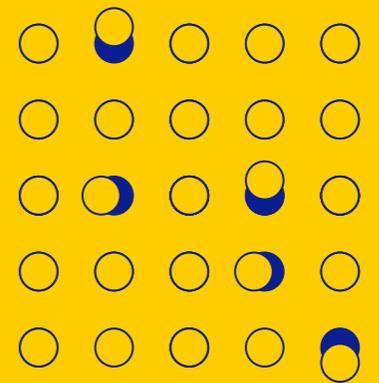
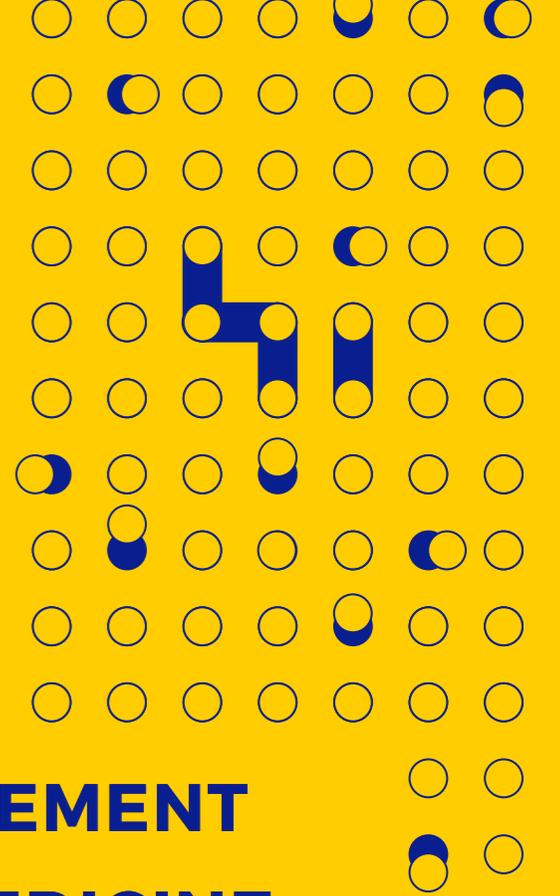
- 5.21** The Steering Committee considered that there is a need to enhance genomic literacy of clinicians, nurses and allied health professionals, in particular those serving in the departments where utilisation and interpretation of genetic and genomic information are expected to become more common, including O&G, paediatrics and oncology, etc. In particular, the Steering Committee considered that HA, by working with other key stakeholders including universities and DH, should provide training ground (such as the HKCH) for genetic and genomic training. The Steering Committee also recognised that the proposed HKGP will play a catalytic role in enhancing the genetic and genomic knowledge of healthcare professionals and attracting both local and international talents.

CHAPTER 6

ENHANCING PUBLIC ENGAGEMENT IN GENOMIC MEDICINE

WAY FORWARD

- 5.22** FHB will work with HA, DH, universities and other relevant parties on an estimate of manpower requirements on genetic and genomic professionals and the support for organising relevant training programmes. HKAM will also coordinate with relevant colleges on specialty training, including the formulation of relevant guidelines or code of practice on genetics and genomics.
- 5.23** In accordance with the SSF GGS, HA will set out the competency requirements and build up relevant expertise for the delivery of advanced genetic and genomic services, e.g. the need for clinical geneticists, pathologists, qualified genetic counsellors and bioinformaticians. Depending on service needs, the creation of relevant posts, such as clinical geneticists, pathologists, post-fellowship training posts in genetics and genomics, genetic counsellors and bioinformaticians will be explored. This will contribute to the establishment of clear career paths for relevant professions in the long run. The Government will provide necessary policy and resources support to HA in accordance with the established mechanism.



Chapter 6

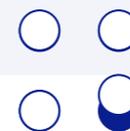
ENHANCING PUBLIC ENGAGEMENT IN GENOMIC MEDICINE

INTRODUCTION

- 6.1** Improved genetic and genomic literacy of the public is conducive to improving the health of overall population and fostering a supportive environment which allows healthcare benefits of genomic medicine to be maximised.
- 6.2** In recent years, the usefulness of genetic and genomic information related to health and disease has become increasingly recognised by the general public through news about research breakthroughs, ethical controversies on the use of genome editing technology on human embryos to modify the traits of the next generation, marketing of direct-to-consumer genetic and genomic tests, etc. Meanwhile, given the complexity of the subject, not all patients and members of the public are equipped with sufficient knowledge in recognising the limitations of genomic medicine under the current technology and the ethical issues behind. Some are hesitant to participate in relevant research projects due to misunderstanding or concern over privacy issues.
- 6.3** DH CGS, under the Genetic Health Promotion Programme (GHPP), promotes genetic and genomic medicine via press interviews, production of promotional materials such as pamphlets and videos, as well as providing professional input to programmes of other non-government organisations. Some academics also developed an educational cartoon booklet for children, adolescents and the families / carers of those who are affected by genetic conditions, and organised events such as focus group meetings, workshops for parents / carers and seminars for professionals who provide genetic services.

DISCUSSION AND ANALYSIS

- 6.4** The Steering Committee recognised the need to engage the public on the relationship between genomics and health, its potential and limitations, as well as the relevant ethical implications.
- 6.5** The Steering Committee recognised the efforts made by the Government, HA, private healthcare sectors, universities, professional bodies and non-governmental organisations on promoting and educating the public on genetic and genomic knowledge and its latest development. However, these education programmes were mostly sporadic and one-off. The Steering Committee considered it necessary to conduct public engagement at a wider and more organised approach, with regular review on the content of the public education programmes to ensure such information is up-to-date.
- 6.6** The Steering Committee considered that the HKGI, to be set up in 2020 to implement the HKGP, would play an important role in connecting related stakeholders including DH, HA, private healthcare sectors, universities, professional bodies and relevant non-governmental organisations, in performing public education on genomic medicine. This network could help rationalise the resources input amongst various parties and offer coordinated, up-to-date and easily accessible genetic and genomic education to the general public.
- 6.7** The Steering Committee also recognised the importance of enhancing public education on the proper use of genetic and genomic tests, including DTCGTs. As a general rule, individuals should be advised to consult healthcare professionals on the use of genetic and genomic tests. Guidelines should be provided to encourage consumers to make informed decisions after considering the clinical validity and utility, limitations of the test results, and privacy and ethical concerns. More discussion on the regulatory aspect of genetic and genomic tests could be found in Chapter 10.



CHAPTER 7

Recommendation 4:

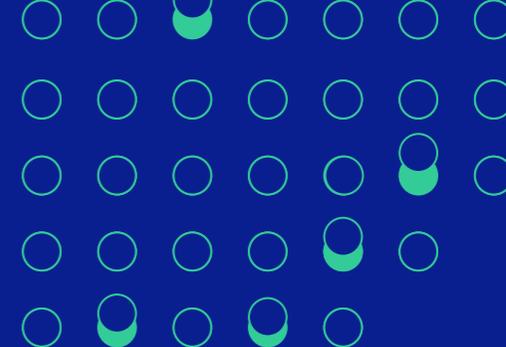
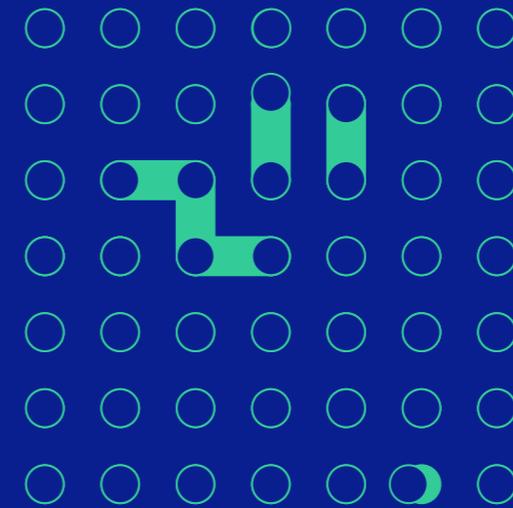
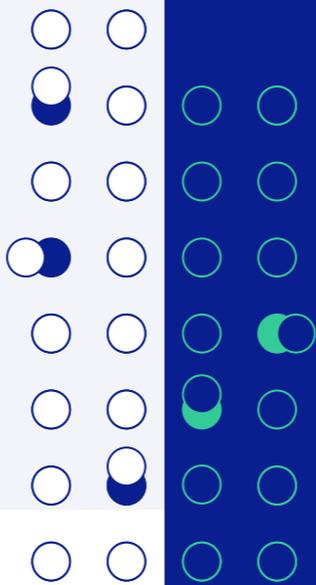
ENHANCING PUBLIC ENGAGEMENT IN GENOMIC MEDICINE

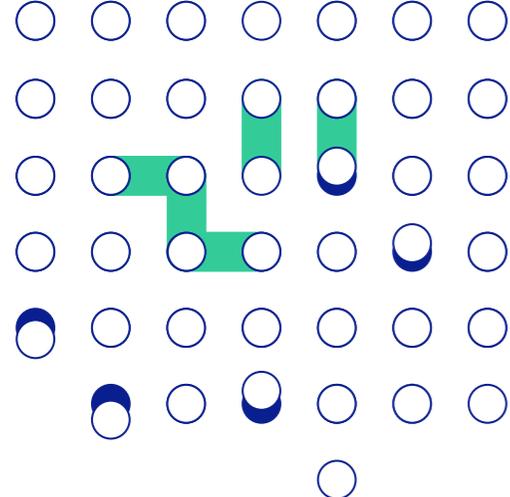
- 6.8 The Steering Committee recommended that –
- (a) The Government should coordinate with professional bodies (e.g. HKAM), academics and other stakeholders (e.g. patient groups) to enhance public understanding on genetics and genomics. DH would have a key role to play under the auspice of Public Health Genomics. Riding on its experience from GHPP, DH is well placed to devise interesting public engagement programmes on genetics and genomics, covering topics including the interaction between genetic materials and behaviour, diet and environment and the proper use of genetic and genomic tests; and
 - (b) The HKGP provides a good opportunity to enhance the understanding of the public on genomic medicine and genome sequencing. The HKGI should ensure there is sufficient publicity, public engagement and participation during the implementation process.

WAY FORWARD

- 6.9 FHB will work with DH to formulate the implementation plan of the genetic and genomic public education programme, in consultation with relevant stakeholders, including the HA and HKAM. FHB will also work closely with the HKGI to promote public understanding on genome sequencing and related issues.

BUILDING A LABORATORY NETWORK FOR GENETIC AND GENOMIC TESTS





Chapter 7

BUILDING A LABORATORY NETWORK FOR GENETIC AND GENOMIC TESTS

INTRODUCTION

- 7.1 Genetic and genomic testing is increasingly used to enhance clinical services in different arenas. It helps diagnosis of diseases, predicts the likelihood of disease onset, informs prognosis and response to treatment, as well as drug reactions. It is also widely used in newborn screening, preimplantation and prenatal genetic testing. Access to quality laboratory services hence becomes the key to realising the expanding benefits of genomic medicine.

- 7.2 The clinical service need of DH and HA patients for genetic and genomic tests is usually met by laboratories in DH and HA as part of the patient care services. For tests not available in DH and HA, patients may order such tests from laboratories in CUHK, HKU, or local / overseas private laboratories, with costs covered by themselves or private donation. HA maintains a formulary of genetic and genomic tests offered by public hospitals and DH CGS. However, there is no formal referral system for ordering genetic and genomic tests among different hospitals in HA. There is also no directory of tests available outside HA and DH CGS, nor a referral mechanism on test ordering among HA, DH and universities. Referral of patients is largely done through the personal connection between clinicians and laboratories. In most cases, patients need to bear the cost of tests conducted by universities in research programmes which are not publicly funded. Such administrative and financial barriers need to be tackled to allow equitable and timely access by patients to the genetic and genomic tests they need.
- 7.3 At the same time, laboratories supported by public funding, including those of DH and HA, as well as some of the laboratories in universities, generally do not coordinate the provision of different types of genetic and genomic tests. This may not only result in unnecessary duplication of public resources, but also hinder the development of advanced genetic tests that require substantial investment and / or specialised expertise due to insufficient volume. Currently, these advanced tests are usually conducted overseas.
- 7.4 A Working Group on Laboratory Network for Genetic Testing² was set up under the Steering Committee, with members from HA, DH, universities and private hospitals, to explore ways to enhance coordination of provision of genetic and genomic testing services.

DISCUSSION AND ANALYSIS

- 7.5 The Working Group considered that there is a need to enhance the cooperation among different laboratories in Hong Kong such that public resources could be utilised in a more cost-effective manner and patients would have more equitable access to appropriate genetic and genomic tests. In particular, the Working Group proposed that an effective referral mechanism should be established among various institutions, and the provision of advanced genetic and genomic tests supported by public funding should be centralised where appropriate.

2. The Membership of the Working Group is at **Annex A**.



Recommendation 5:

ENHANCING THE LABORATORY NETWORK WITH EFFECTIVE REFERRAL MECHANISM AND CENTRALISATION OF ADVANCED GENETIC AND GENOMIC TESTS

To establish a referral mechanism

- 7.6** To enhance coordination among DH, HA and universities, the Working Group recommended that a mechanism should be established in order to –
- (a) Maintain a formulary of approved genetic and genomic tests available in laboratories under DH, HA, universities and the private sector, with clear testing criteria to facilitate clinical decision on test ordering;
 - (b) Allow referral of patients between HA hospitals, and between DH and HA, to receive genetic and genomic test, with proper charging arrangement between the referring institution and the testing institution; and
 - (c) Formalise the ordering and funding arrangement for genetic and genomic tests provided by universities and private laboratories, subject to the dedicated funding mechanism for genetic and genomic tests to be developed under the HA SSF GGS.
- 7.7** The proposed referral mechanism would enhance the access of patients to suitable genetic and genomic tests, expedite testing delivery, optimise the use of public resources and facilitate the development of local genetic and genomic testing in both the public and private sector.

Centralisation of advanced genetic and genomic tests

- 7.8** The establishment of a referral mechanism would facilitate the centralisation of advanced genetic and genomic tests in a few designated laboratories for expertise development and resources optimisation. The Working Group recommended that as a matter of principle, advanced genetic and genomic tests in HA and DH CGS that fulfill the following conditions should be centralised at designated laboratories –

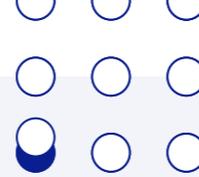
- (a) Tests that require sophisticated technologies or costly equipment, in particular those with short technology life-cycle;
- (b) Tests with low utilisation volume;
- (c) Tests that generate significant amount of data which requires high computational power for analysis and large storage capacity for archive; and
- (d) Tests that require highly skilled experts which are of limited availability in Hong Kong (e.g. clinical geneticists, pathologists with relevant experience and bioinformaticians).

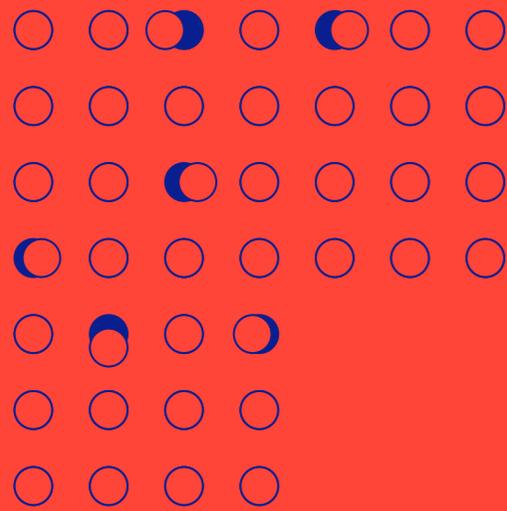
- 7.9** The proposed centralisation could synergise expertise, unify laboratory standards and optimise the use of public resources. It avoids unnecessary competition for limited experts in the field and promotes the healthy development of genetic and genomic testing in Hong Kong in the long run. Meanwhile, centralisation is considered not necessary for other commonly used genetic and genomic tests, which should continue to be provided in laboratories in different hospitals to meet the larger service demand.

- 7.10** The Steering Committee noted that the SSF GGS of HA is exploring a tiered approach and hub-and-spoke model to provide genetic and genomic services in HA, including developing a genetic and genomic service directory to facilitate standardised service provision and information sharing, referring patients to relevant hubs for advanced genetic and genomic tests, and mainstreaming some of the genetic and genomic tests offered by universities in clinical service. The direction is in line with Working Group's recommendations above.

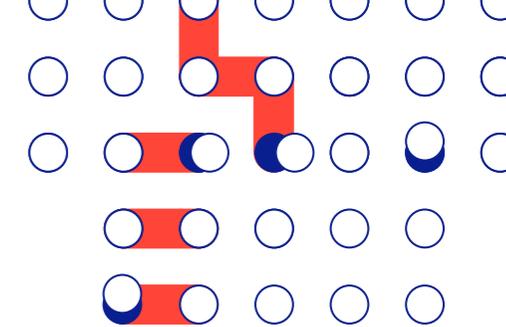
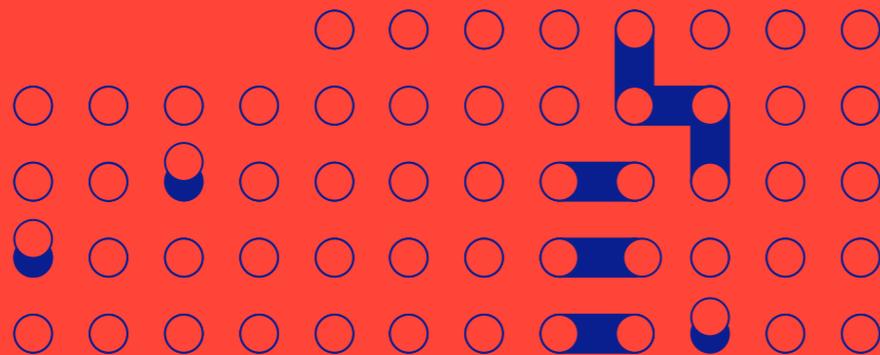
WAY FORWARD

- 7.11** FHB will work with DH, HA, universities and the private sector to develop the proposed referral mechanism and explore the centralisation arrangement of advanced genetic and genomic tests in designated laboratories.





FACILITATING THE ESTABLISHMENT OF A BIOBANK NETWORK FOR GENOMIC MEDICINE



Chapter 8

FACILITATING THE ESTABLISHMENT OF A BIOBANK NETWORK FOR GENOMIC MEDICINE

INTRODUCTION

- 8.1 Human tissue biobanks³ and gene banks with repositories of DNA, broadly referred to as biobanks, play an important role in the analysis of genetic determinants of diseases. Some biobanks, such as the Australia Breast Cancer Tissue Bank, are disease-oriented, the samples of which are usually collected from patients of the same group of disease for disease-specific analysis. Others such as the UK biobank are population-based, where samples are collected from 500 000 people for purposes such as identifying biomarkers for diseases susceptibility and population profiling. A summary table of examples of overseas biobanks is at **Annex H**.
- 8.2 Internationally, there are mainly two options in enhancing the efficient use of fragmented biobank resources, namely the setting up of a centralised biobank or a coordinated network of small biobanks. An example of central biobank is the UK Biobank, which makes biological samples and associated de-identified genomic and longitudinal health data from 500 000 volunteer project participants open to bona fide researchers worldwide. Alternatively, EuroBioBank is a biobank network specifically on rare diseases, and linked up 25 biobanks in different countries across Europe.
- 8.3 There is no territory-wide centralised biobank in Hong Kong. Local biobanks are mostly established by individual researchers to support specific research projects, or by clinicians from donation of residual patient specimens available during routine patient care. These biobank resources mostly belong to the corresponding researchers / clinicians or their institutions and are mainly accessible only by them and their collaborators. For instance, the Hong Kong Diabetes Biobank was set up by the CUHK and the HKUST under the TRANSCEND Consortium to study diabetes mellitus, a biorepository on breast cancer was set up by the HKU, and a biobank on cancer was set up by the Queen Elizabeth Hospital. Population-based biobanks are less common. One of the few examples is the “Children of 1997” birth cohort maintained by the HKU.

3. According to the International Society for Biological and Environmental Repositories, a biobank is a physical or virtual entity for the collection, handling, storage, preservation, retrieval, and distribution of biological samples and their associated data, in support of current or future uses for research purposes.

- 8.4 Given the importance of biobanks to genomic medicine, the Working Group on Biobank⁴ was established under the Steering Committee to discuss the need for a centralised biobank or a biobank network to facilitate the development of genomic medicine in Hong Kong.

DISCUSSION AND ANALYSIS

- 8.5 The Steering Committee recognised that in the era of genomics and big data where scientific development would benefit from the availability of a larger number of samples, there is a need to bring together the fragmented biobank resources in Hong Kong and make them more readily available to the scientific community, with due respect to the privacy of the donors. The benefit can be especially obvious for certain diseases or disorders such as uncommon diseases where the patient population is small and recruitment of research subjects is difficult. Better coordination could also reduce duplication of research efforts in recruiting participants and optimise the use of precious biobank resources.
- 8.6 The Steering Committee appreciated the past developments of biobanks in various institutions in Hong Kong and recognised the challenge in establishing a comprehensive centralised biobank. In particular, merging the institution-based biobanks would overhaul the existing research practices of universities and the HA, and there would be considerable legal, administrative and ethical concerns. For instance, as existing biobanks were mostly established to support particular research projects, the scope of consent obtained is usually specific to one research project, which largely limits the secondary use of donated samples and data for a new research purpose, or restricts the shared use of samples or data with other researchers. Furthermore, different institutions have different biobank and research ethics governance structures, which result in difficulties in sharing samples and data among institutions.
- 8.7 Given the above concern, instead of merging the existing biobanks maintained by different institutions, the Steering Committee considered that priority should be given to establishing a network of local biobanks with guidelines on facilitating the shared use of specimens and data. Meanwhile, the Steering Committee noted that the HKGP would serve as a pilot project to tackle the issues relevant to the shared use of genomic data among institutions, such as broad consent, public education and privacy safeguard.

Recommendation 6:

FACILITATING THE ESTABLISHMENT OF A BIOBANK NETWORK FOR GENOMIC RESEARCH

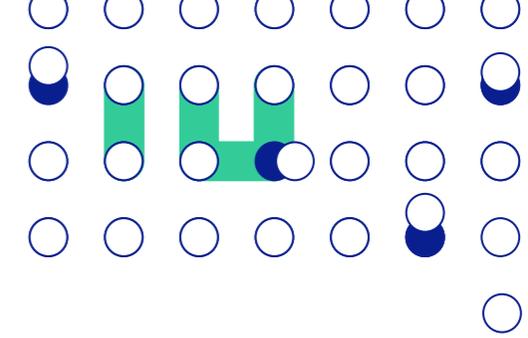
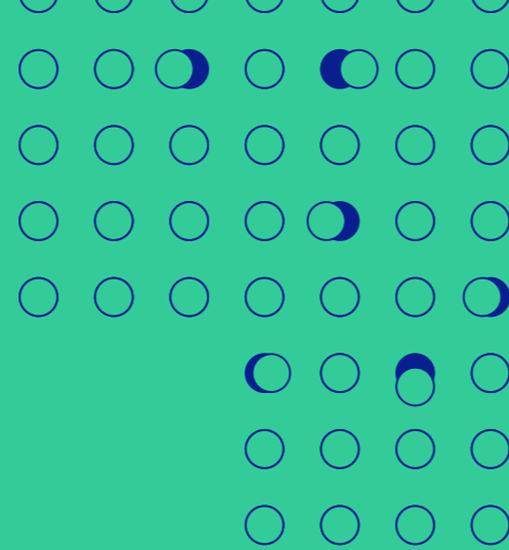
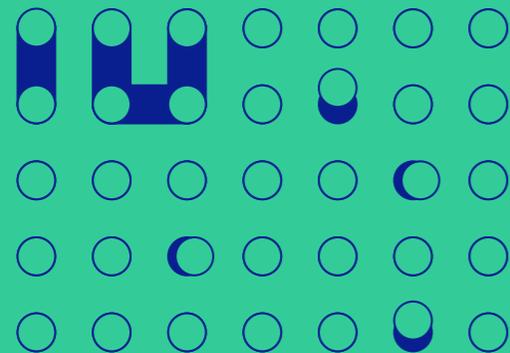
- 8.8 The Steering Committee recommended that –
- A standardised consent template and protocol, covering issues such as quality assurance, ethics and privacy should be developed for participating institutions to follow in order to enable the shared use of de-identified biospecimens and associated data for future genomic research;
 - The HKGP would serve as a pilot project to establish a robust governance mechanism to take forward (a) above, subject to the protection of privacy and interests of participants; and
 - In order to provide incentives for institutions to join the biobank network, the Government should consider making participation in the biobank network as one of the conditions for future genomic research projects to receive government funding (e.g. the Health and Medical Research Fund).

WAY FORWARD

- 8.9 The Government and the HKGI will formulate the details of the standardised protocol and governance structure in paragraph 8.8 above in consultation with major stakeholders, including the HA and universities. Meanwhile, FHB will discuss with relevant bureaux / departments to consider how incentives could be provided for future research projects receiving government funding to participate in the biobank network.

4. The Membership of the Working Group is at **Annex A**.

ENHANCING THE REGULATION ON THE USE OF GENETIC DATA FOR INSURANCE AND EMPLOYMENT PURPOSES



Chapter 9

ENHANCING THE REGULATION ON THE USE OF GENETIC DATA FOR INSURANCE AND EMPLOYMENT PURPOSES

INTRODUCTION

- 9.1 While genetic tests are valuable tools in diagnosis, prevention and treatment of diseases, some people are hesitant in taking genetic tests for fear of privacy concern or genetic discrimination, which refers to the less favourable treatment on the ground of genetic factors that may cause or increase the risk of an inherited disorder or disability. Such fear, mostly in the context of insurance and employment, considerably compromises the benefits brought by genomic medicine to patients and the general population, and hinders the development of genomic research.

DISCUSSION AND ANALYSIS

Local Situation

- 9.2 Locally, processing of genetic data by third parties (i.e. other than the data subject and those authorised directly for data processing) is mainly governed by two existing ordinances, namely the Disability Discrimination Ordinance (DDO) (Cap. 487) and the Personal Data (Privacy) Ordinance (PD(P)O) (Cap. 486). Under the DDO, the definition of disability covers disabilities which may exist in the future or a disability which is imputed to a person. In other words, genetic and congenital predisposition is considered as "disability" under the DDO, and discriminatory treatment in relation to insurance based on genetic test results may be unlawful unless it was reasonable by reference to actuarial or other data from a reliable source, or otherwise justifiable under the DDO. It is also unlawful for employers to request or require information of a medical nature, including genetic information, in connection with discriminating against an employee, unless the employer requires the medical information to determine whether the employee would be able to perform the inherent requirements of the job or would require services or facilities not required by persons without a disability.

9.3 Separately, genetic data is considered as personal data protected under the PD(P)O. Generally speaking, genetic data is considered as sensitive in nature, as it may put a person's fundamental rights and freedoms at risk of unlawful discrimination. Hence, genetic data needs to be treated with greater care. Under the PD(P)O, genetic data must be collected in a lawful and fair way, for a purpose directly related to a function / activity of the data user, and the data collected should be necessary but not excessive. Given that the predictive power of genetic testing results may vary in accordance with the penetration and expressivity of the gene in question, indiscriminate collection of such results from all insurance applicants may deem to be excessive in contravention of the PD(P)O. More details on the regulatory measures under the two ordinances are summarised at **Annex I**.

9.4 The Hong Kong Federation of Insurers (HKFI) issued a Code of Practice on Genetic Testing (the Code) for its members in 2005. The Code of Practice is not available to the public. Given the rapid change in genomic medicine in recent years, HKFI is reviewing the Code with reference to international practices and local circumstances.

International Practices

9.5 Most advanced economies recognise that there is a need to regulate genetic discrimination for privacy and ethical reasons, and for avoiding hindrance to the development of genomic medicine. While recognising the special nature of insurance industry, the international trend is to impose restrictions on the use of genetic information for insurance purpose as a matter of principle, with exceptions allowed in limited scenarios. Some countries adopt a legislative approach, while some opt for self-regulation by the insurance industry.

9.6 For those which provide for exceptions on the use of genetic data in certain circumstances, the principle is that the requisite data processing should only be allowed after independent assessment of conformity with a set of criteria by type of test used and with regard to a particular risk to be insured. For instance, a mechanism for listing out the types of predictive genetic test results having a high positive predictive value that the insurers are allowed to collect but not otherwise is adopted in the United Kingdom. A summary of the international practices in regulating use of genetic data for insurance purpose is at **Annex J**.

Need for a Balance

9.7 The Steering Committee noted that according to the experience of researchers and clinicians, there was generally inadequate transparency on how insurance companies use the genetic data collected for underwriting. A considerable number of patients and their family members expressed worry about the impact of undertaking genetic tests on their eligibility, or the eligibility of their family members, of getting insurance coverage. Such ambiguity often deterred the family members of patients from undertaking genetic tests. Researches would also be hindered by difficulties in recruiting participants.

9.8 Meanwhile, the special nature of insurance requires full disclosure of material facts including health-related data necessary for underwriting decisions. A complete ban on the use of genetic information by insurers may present a risk of anti-selection, where individuals may procure insurance with knowledge about their risks not known to the insurer.

9.9 Hence, a reasonable balance needs to be struck in considering the extent and approach of the regulatory framework on genetic discrimination.

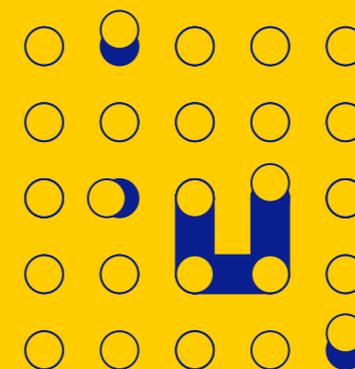
Recommendation 7:

ENHANCING THE REGULATION ON THE USE OF GENETIC DATA FOR INSURANCE AND EMPLOYMENT PURPOSES

- 9.10** While the DDO and the PD(P)O have been providing protection against genetic discrimination and relevant privacy issues to a certain extent, the Steering Committee considered that more specific regulatory measures on genetic discrimination in the context of insurance should be explored, covering life, critical illness and health insurance.
- 9.11** In view of the rapid development of genomic medicine, the Steering Committee considered that it would be more pragmatic and flexible to enhance the regulation through an updated Code of Practice by the HKFI. With further advancement of genomic medicine, the Code could be updated regularly to suit the latest development.
- 9.12** The Steering Committee recommended that in updating the Code, HKFI should be invited to consider adhering to the following principles, which represent the mainstream international practices, with due regard to the local circumstances –
- Insurer should not require or put pressure on an applicant to undertake a genetic test of any kind in order to obtain insurance;
 - Results of diagnostic genetic testing⁵ should be treated as part of the medical information and hence should be disclosed to insurers;
 - Insurer should not ask for the results of a genetic test of any kind conducted for research purpose;

5. Taking the National Institutes of Health (NIH) definition as an example, diagnostic testing is used to identify or rule out a specific genetic or chromosomal condition. In many cases, genetic testing is used to confirm a diagnosis when a particular condition is suspected based on physical signs and symptoms.
Reference: NIH – Genetic Home Reference <https://ghr.nlm.nih.gov/primer/testing/uses>

CHAPTER 10



PROMOTING THE PROPER USE OF GENETIC AND GENOMIC TESTS

- (d) As a matter of principle, insurer should not ask for the results from predictive and pre-symptomatic types of genetic testing⁶. Exceptions may be allowed after meeting certain criteria, e.g. high-end products over a certain protection limit, and where a specific type of genetic test result is necessary for the underwriting decision by reference to actuarial or other data from a reasonable source or otherwise justifiable under the DDO; and
- (e) The Code should set out clearly the principles and practices of insurance companies in using genetic information, and should be available to the general public, in order to enhance transparency.

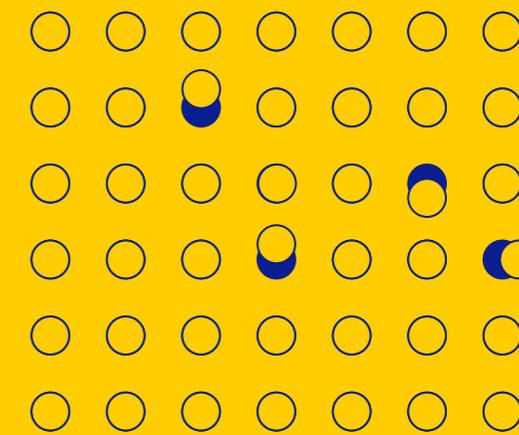
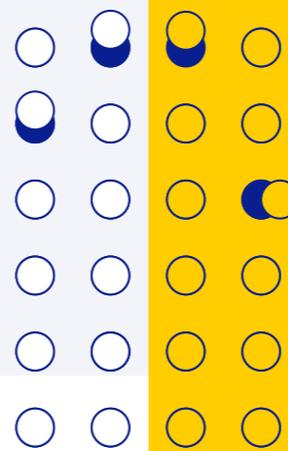
9.13 Appreciating the Code would be conducive to enhancing transparency in the use of the genetic data for insurance purpose, the Steering Committee recommended that the Government should continue to review the situation and consider introducing legislative measures to regulate the use of genetic test results as and when necessary.

9.14 In relation to employment, the Steering Committee in general considered that the existing legislations have provided appropriate framework to guard against genetic discrimination in this aspect; and recommended that no additional regulatory measure is required. However, in view of the potential increase in the adoption of genetic tests, public education should be enhanced on anti-genetic discrimination for employers and employees.

WAY FORWARD

9.15 FHB has invited HKFI to update its Code taking into account the recommendation of the Steering Committee, international practices and local circumstances. FHB will also liaise with the Consumer Council, the Equal Opportunities Commission, the Privacy Commissioner for Personal Data, the Insurance Authority and other relevant parties to enhance public education on anti-genetic discrimination and privacy protection.

6. Taking the NIH definition as an example, predictive and presymptomatic types of testing are used to detect gene mutations associated with disorders that appear after birth, often later in life. These tests can be helpful to people who have a family member with a genetic disorder, but who have no features of the disorder themselves at the time of testing. Predictive testing can identify mutations that increase a person's risk of developing disorders with a genetic basis, such as certain types of cancer. Presymptomatic testing can determine whether a person will develop a genetic disorder, such as hereditary hemochromatosis (an iron overload disorder), before any signs or symptoms appear. Reference: NIH - Genetic Home Reference <https://ghr.nlm.nih.gov/primer/testing/uses>



Chapter 10

PROMOTING THE PROPER USE OF GENETIC AND GENOMIC TESTS

INTRODUCTION

- 10.1** In the clinical setting, most of the genetic and genomic tests are ordered by clinicians with prior clinical assessment. Before ordering the test, clinicians usually explain to the patients the clinical validity and implications of conducting the test and obtain their consent. After the test, clinicians would explain the test results to patients carefully with proper genetic counselling, and advise on the appropriate follow-up actions. Such practices are essential to ensuring proper follow-up of the test results and addressing the ethical issues specific to genetic and genomic tests, which are more complicated than other investigations.
- 10.2** In recent years, due to the advancement in gene sequencing technology and sharp decreases in costs, DTCGTs have become increasingly popular. Some are offered as gifts of insurance products, while some are sold directly to the general public at retail stores or online. In addition to health-related genetic and genomics tests such as pre-symptomatic tests for monogenic disorders (e.g. cystic fibrosis), predisposition tests on carrier genes for cancers and susceptibility tests for multifactorial diseases (e.g. cardiovascular diseases, diabetes mellitus), there are also DTCGTs which are not health-related, such as ancestry tests and tests which claim to provide insights to one's talents, weight, diet, athletic ability and wine preferences.
- 10.3** There is at present no overarching regulatory regime for genetic and genomic testing in Hong Kong. The current regulation of Medical Laboratory Technologists (MLTs) under the Supplementary Medical Professions Ordinance (Cap. 359) (SMPO)⁷ essentially restricts the performance of genetic and genomic testing for diagnostic or treatment purposes by MLTs, which can only be conducted with referral from a registered medical, dental and/or veterinary practitioners. There have been suggestions to consider if other professionals like biomedical scientists equipped with genomic knowledge and laboratory techniques can also be allowed to perform the genetic and genomic tests. For genetic and genomic tests which do not fall under the ambit of SMPO, including most of the DTCGTs which are non-health related, there are no restrictions on who can conduct the tests and how they could be offered to consumers.

- 10.4** As regards the laboratory standards, the HOKLAS set up by the Hong Kong Accreditation Service is open to voluntary participation from relevant service providers, including all laboratories in Hong Kong for the accreditation of genetic and genomic testing for diagnosis and treatment. The HOKLAS accredited the laboratories according to *International Organization for Standardization (ISO) 15189: Medical laboratories – Requirements for quality and competence*. The ISO 15189 required the laboratories to meet various requirements, including quality control, techniques and personal data privacy in the whole testing procedure.

DISCUSSION AND ANALYSIS

- 10.5** The Steering Committee reviewed international practices in regulating DTCGTs, and noted that the regulatory regimes for DTCGTs in overseas jurisdictions range from the issue of Code of Practice to legislation. Key findings are summarised at **Annex K**. The Steering Committee considered that as DTCGTs are available outside the clinical setting, there was a need to introduce suitable measures to address healthcare and ethical issues in order to safeguard public health.

Concerns over DTCGTs to be addressed

Insufficient information on clinical validity and limitations

- 10.6** The clinical validity of predictive genetic tests requires support by large volume of data and studies. Each genetic test has its own limitations and some may not provide meaningful results at all. While some providers of DTCGTs may reveal relevant scientific data and limitations for consumer's information, others may not provide sufficient evidence to support the clinical validity of the tests offered, nor the limitations behind. Misleading or ambiguous representation in marketing materials may give consumers false impression on the validity of the tests.

7. Under the SMPO, only persons who have registered as MLT can practise in processing clinical, medical, legal, public health or veterinary specimens for the sole purpose of making and reporting on analysis or examination in vitro and the processing of all matters for human and animal consumption for the sole purpose of making and reporting on analysis or examination in vitro. The Code of Practice of the Medical Laboratory Technologists Board of Hong Kong stipulates that a MLT should not perform any tests for the purpose of medical diagnosis and treatment in the absence of a referral from registered medical, dental and/or veterinary practitioners.

Insufficient professional advice on the suitability of genetic test

- 10.7 Even when providers of DTCGTs provide detailed scientific data on the clinical validity of the tests, most of the consumers may not be able to comprehend the information and assess the suitability of the test for them without professional advice. For instance, there may not be any treatment available for some of the conditions covered by the test; or the treatment is only appropriate after clinical symptoms appear. Taking DTCGTs without assessing the suitability of the test for an individual would not only have financial implications, but may also be considered as unethical when the results are released without proper professional advice, as it might generate unnecessary anxiety or false assurance of one's health conditions.

No specific regulation on safety and quality of DTCGT products

- 10.8 The test kits of some DTCGTs may be regulated by existing legislation, such as the Pharmacy and Poisons Ordinance (Cap. 138) and the Consumer Goods Safety Ordinance (Cap. 456). However, there is no specific regulation targeting at the regulation of safety and quality of DTCGTs at the moment.

Limited genetic counselling

- 10.9 Because of its complicated nature, results of genetic tests need to be interpreted by well-trained healthcare professionals with proper genetic counselling according to individual situation. Without proper counselling and interpretation, a negative test result may impart an unwarranted sense of assurance and lead to continued adoption of unhealthy lifestyles. A positive test result may lead to unwanted anxiety and further tests, adding unnecessary burden to the healthcare system. Not all providers of DTCGTs offer genetic counselling services to their clients, and for some which offer, it is unclear if the counsellor has proper training or qualifications. In some cases, the health advice provided by the so-called counsellors or health trainers on prevention of adverse health conditions is no more specific than the usual advice given to the general public.

Privacy and data security

- 10.10 Genetic and genomic data are sensitive medical data. Individuals who take DTCGTs may not be aware of the potential threat to his / her privacy through data sharing with third party, the risk of data security and genetic discrimination. As the collection of sample for DTCGTs is not regulated, there is also concern on unauthorised collection of samples for other purposes without the data subject's consent.

Recommendation 8:

PROMOTING THE PROPER USE OF GENETIC AND GENOMIC TESTS

- 10.11 The Steering Committee considered that different approaches for health-related and non-health-related genetic tests should be adopted in formulating the regulatory measures and public education strategies, in view of the different level of health risks involved.

Enhancing public education

- 10.12 As many of the DTCGTs could be obtained overseas through online purchase and hence difficult to regulate locally, the Steering Committee considered that it is important to enhance public education to empower people to make informed decisions in collaboration with various stakeholders such as the Consumer Council. As a general rule, individuals should be advised to consult healthcare professionals on the use of genetic and genomic tests. Guidelines should be provided to encourage consumers to make informed decisions after considering the clinical validity and utility, limitations of the test results, and privacy and ethical concerns.

Enhancing regulation of health-related genetic tests

- 10.13 In addition to enhancing public education, the Steering Committee considered that health-related genetic tests (including both diagnostic and predictive ones) could lead to healthcare decisions. Without proper counselling and professional advices, a test result may cause anxiety or false reassurance and would in turn lead to unnecessary investigations or delayed health care and hence entail greater health risks. As such, additional measures should be introduced according to the following principles –
- (a) Evidence supporting the clinical validity and utility of the tests and the related limitations should be clearly specified in advertisement and labelling;
 - (b) The tests should be handled by qualified professionals in accordance with relevant laboratory standards;

- (c) Pre-test genetic counselling should be provided by trained professionals to consumers on issues including the usefulness, suitability and benefit of the testing for the particular individual, as well as the potential implications on privacy, insurance, etc.; and
- (d) Post-test genetic counselling should be provided by trained professionals to the test recipient to advise appropriate follow-up actions, to avoid unwarranted assurance or anxiety, and to properly handle other ethical implications.

WAY FORWARD

- 10.14** The Government has been working on a statutory control framework for medical devices, which would cover those devices intended to be used for human genetic tests in vitro that meet the definition of medical devices⁸, to ensure that they should comply with the requirements on safety, quality, performance and efficacy. As an interim measure, DH has established a Medical Device Administrative Control System.
- 10.15** Meanwhile, the Government will invite relevant regulatory bodies (such as the Medical Laboratory Technologists Board, the statutory body responsible for registration and disciplinary control of MLTs) to review the qualifications and experience requirements of professionals and technologists who are eligible to handle genetic and genomic tests.
- 10.16** HKAM plans to establish a guideline on the good practice of genetic and genomic medicine for medical professionals. The guideline will cover the ethical principles on requesting genetic test, confidentiality and disclosure of genetic test results, best practice on the genetic testing procedures including obtaining informed consent, pre- and post-test counselling. FHB would work closely with HKAM in this regard.

8. Whether or not a product is considered a medical device depends on its intended purpose. If the test kit of genetic tests is intended to examine specimens derived from the human body for the medical purpose stipulated under the definition of medical device, it will be classified as an in vitro diagnostic medical device. For example, in vitro diagnostic test kits for infectious diseases are in vitro diagnostic medical devices. Whereas in the case of DNA ancestry or paternity tests which do not have medical purpose, their test kits are not classified as medical devices.

CONCLUSION AND WAY FORWARD

Genomic medicine is a relatively new development in the medical field. Enhancing its development in Hong Kong requires a forward-looking approach and close co-operation and collaboration among various stakeholders.

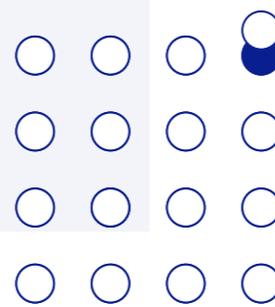
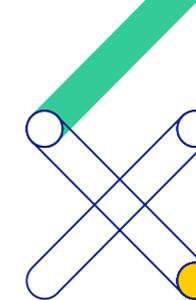
There is ongoing discussion on the readiness and challenges in integrating genomics into routine clinical service, given the practical use of genome analysis can be constrained by the complex origin of many disease conditions, including varying degrees of involvement or penetrance of different gene variants, and complex interaction of genetic, lifestyle and environmental factors, etc. Nonetheless, integration of genomics into healthcare will undoubtedly bring along transformational change in patient care in the longer run.

The Steering Committee thoroughly deliberated the strategies for the development of genomic medicine, including enhancing the genomic literacy of healthcare workforce for supporting patient-centric and equitable access to genetic and genomic services; effective mechanism for technology update and translation of research to clinical practice with proven quality, safety and clinical utility, sustainable financing, as well as prudent and effective use of genome data for clinical practice and research. The Steering Committee also proposed ways to address ethical, legal and social issues.

Looking ahead, the HKGP will serve as the cornerstone to drive the development of genomic medicine by establishing a genome database of the local population, testing infrastructure and talent pool. Along with it, the HA will implement the SSF GGS, which aims to fill the current service gaps and set out a blueprint for its genetic and genomic services with reference to the views of the Steering Committee. Leveraging on the huge potential of genomic medicine, the SSF GGS will bring significant benefits to patients being cared for in the public sector.

The vision of genomic medicine will go beyond benefiting patients in need. The public at large will also enjoy the benefits in the long run. Advances in genomic medicine and research will provide us with information and technology that help understand and analyse individuals and their living environment over the life course more precisely, which in turn will facilitate more personalised preventive interventions for at-risk groups and improve the overall health of the population.

Hong Kong has a strong edge in genomic research and outstanding clinical services in both public and private healthcare systems, which demonstrate our promising potential to excel in genomic medicine. The Steering Committee hopes that FHB will lead and coordinate the implementation of the recommendations detailed in this report in consultation with various stakeholders including DH, HA, universities, medical and other relevant professional bodies as well as the private sector, and as always, with improving the healthcare system and patient care in mind and at heart.



GOVERNMENT RESPONSE

The Secretary for Food and Health welcomes the report of the Steering Committee and accepts the recommendations in full. The Food and Health Bureau, supported by the Department of Health, will implement the recommendations in collaboration with the Hospital Authority, universities, the Hong Kong Academy of Medicine and other stakeholders.

Looking ahead, the Government has already taken steps to set up the Hong Kong Genome Institute and prepare for the implementation of the Hong Kong Genome Project (HKGP). The HKGP would serve as a catalyst to build up a talent pool, set standards and establish a genome database of local population, which would in turn support research and clinical application of genomic medicine in Hong Kong. It could also promote public understanding on genomic medicine as well as the related ethical issues.

Meanwhile, the Government would continue to work with the Hospital Authority to provide structured and coordinated genetic and genomic services in accordance with the Strategic Service Framework for Genetic and Genomic Services. Support would be provided to academic institutions and professional bodies in training relevant professionals. The Government would also continue the dialogue with relevant organisations to strengthen control on the use of genetic data for insurance and employment purposes.

The report of the Steering Committee marks the milestone of genomic medicine development in Hong Kong. On behalf of the Government, the Secretary for Food and Health thanks the Chairman and Members of the Steering Committee and its Working Groups for their valuable advice and contributions in formulating the strategic directions of the development of genomic medicine, which would help translate scientific advancement to patient benefits in the years to come.

GLOSSARY

Biobank

A biobank is a physical or virtual entity for the collection, handling, storage, preservation, retrieval, and distribution of biological samples and their associated data, in support of current or future uses for research purposes.

Bioinformatics and bioinformatician

Bioinformatics is a subdiscipline of biology and computer science concerned with the acquisition, storage, analysis, and dissemination of biological data, most often DNA and amino acid sequences. Bioinformaticians are scientists who apply computer science and information technology to biological problems, for example by writing data analysis algorithms, managing data storage and using computer programme to help in interpreting genomic testing data.

Cancer liquid biopsy

Cancer liquid biopsy is a less invasive approach for the detection and monitoring of cancer biomarkers through analysis of circulating tumour DNA (ctDNA) in the blood samples of the patient rather than direct analysis of tumour tissue which requires surgery for sample collection.

Chromosomal microarray/ Microarray

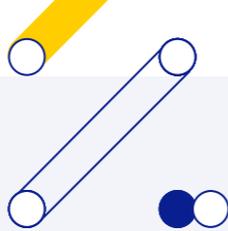
Microarray is a microchip-based testing method that allows simultaneous measurement of many pieces of DNA at once. Microarray can be used, for example, to analyse the gene expression at thousands of regions of the genome or detect extra or missing chromosomal segments (also called copy number variants). The latter use is referred specifically as chromosomal microarray.

Cytogenetics and cytogenetic test

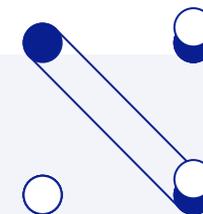
Cytogenetics is a branch of genetics that studies the structure of DNA, or specifically chromosomes (a highly condensed structure of DNA during cell division), within the cell nucleus. Classical cytogenetics uses specialised staining techniques and light microscope to study numerical and gross structural changes in chromosomes. In modern times, cytogenetics is often used with molecular biology techniques (molecular cytogenetics) that allows the study of genetic materials at a much higher resolution.

Deoxyribonucleic acid (DNA)

DNA is a chemical that contains, or 'encodes', genetic information, which is made up of four different chemical bases known as 'A' (adenine), 'C' (cytosine), 'G' (guanine) and 'T' (thymine).



DNA sequencing	DNA sequencing is a laboratory technique used in molecular biology to determine the order of bases (i.e. A, C, G and T) in DNA.	Genomic medicine	Genomic medicine is the use of genomic information and technologies to determine disease risk and predisposition, diagnosis and prognosis (i.e. a forecast of the probable course and outcome of a disease), and the selection and prioritisation of therapeutic options.
Diagnostic genetic testing	According to the US National Institutes of Health, diagnostic genetic testing is used to identify or rule out a specific genetic or chromosomal condition. In many cases, genetic testing is used to confirm a diagnosis when a particular condition is suspected based on physical signs and symptoms.	Genotype	Genotype is the DNA sequence of an organism or individual, which determines (along with environmental influences) the specific characteristics (phenotype) of that organism or person.
Direct-to-consumer genetic tests	Direct-to-consumer genetic tests are genetic tests directly marketed to consumers without involvement of healthcare provider.	Inborn Errors of Metabolism (IEM)	Metabolism is a sequence of chemical reactions that take place in cells in the body. These reactions are responsible for the breakdown of nutrients and the generation of energy in our bodies. IEM are a group of disorders that causes a block in a metabolic pathway leading to clinically significant consequences.
Expressivity	In the context of genetic diseases, expressivity refers to the degree of variation in disease signs and symptoms or their severity across people with the same genetic condition.	Medical Laboratory Technologist	According to the Supplementary Medical Professions Ordinance (Cap. 359), a medical laboratory technologist is “a person trained in the practice of processing clinical, medical, legal, public health or veterinary specimens for the sole purpose of making and reporting on analysis or examination in vitro and the processing of all matters for human and animal consumption for the sole purpose of making and reporting on analysis or examination in vitro”.
Genetics	Genetics is the study of genes and how they are inherited.	Molecular genetic test	Molecular genetic tests (or gene tests) study single genes or short lengths of DNA to identify variations or mutations that lead to a genetic disorder.
Genetic counselling	Genetic counselling is the process of helping people understand and adapt to the genetic, medical, psychological and familial implications of genetic contributions to disease.	Penetrance	In the context of genetic diseases, penetrance refers to the proportion of people with a particular genetic change who exhibit signs and symptoms of a genetic disorder.
Genetic predisposition	A genetic predisposition (also called genetic susceptibility) is an increased likelihood of developing a particular disease based on a person’s genetic makeup.	Pharmacogenomics	Pharmacogenomics is the use of genetic and genomic information to tailor pharmaceutical treatment to an individual.
Genome	Genome is an organism’s complete genetic material, including both genes that provide the instructions for producing proteins (2% of the genome) and the non-coding sequences (98% of the genome).	Phenotype	Phenotype is an organism’s observable physical and biochemical characteristics directly influenced by the genotype and/or environment. In human, this is often the observed signs and symptoms of a condition.
Genomics	Genomics is the study of the genomes of individuals and organisms that examines both the coding and non-coding regions. This term is also used when talking about related laboratory and bioinformatics techniques. The study of genomics in humans focuses on areas of the genome associated with health and disease.		



Predictive and pre-symptomatic genetic testing

According to the US National Institutes of Health, predictive and pre-symptomatic types of testing are used to detect gene mutations associated with disorders that appear after birth, often later in life.

These tests can be helpful to people who have a family member with a genetic disorder, but who have no features of the disorder themselves at the time of testing. Predictive testing can identify mutations that increase a person's risk of developing disorders with a genetic basis, such as certain types of cancer. Pre-symptomatic testing can determine whether a person will develop a genetic disorder before any signs or symptoms appear.

Whole exome sequencing

Whole exome sequencing is a type of DNA sequencing targeting only a small part (around 2%) of the human genome, i.e. the exome that directly codes for proteins.

Whole genome sequencing

Whole genome sequencing is a type of DNA sequencing targeting at the whole genome, i.e. every DNA base in a genome, of an individual.

ABBREVIATIONS

ABCTB	Australian Breast Cancer Tissue Bank	DoH NI	Northern Ireland Department of Health
ABCC	American Board of Genetic Counseling	DTCGTs	Direct-to-consumer Genetic Tests
ABI	Association of British Insurers	EBMG	European Board of Medical Genetics
ACCG	Accreditation Council for Genetic Counseling	ELSI	Ethical, Legal and Social Implications
ACMG	American College of Medical Genetics and Genomics	EOC	Equal Opportunities Commission
CAGC	Canadian Association of Genetic Counsellors	ESHG	European Society of Human Genetics
CDCOPHG	Public Health Genomics under the Centres for Disease Control and Prevention	FDA	Food and Drug Administration
CGS	Clinical Genetic Service	FHB	Food and Health Bureau
CGT	Clinical Genetic and Genomic Testing	FINBB	FinBioBank
CHT	Congenital Hypothyroidism	FSC	Financial Services Council
CUGCs	Clinical Utility Gene Cards	G6PD	Glucose-6-Phosphate Dehydrogenase
CUHK	The Chinese University of Hong Kong	GCRB	Genetic Counsellor Registration Board
DDO	Disability Discrimination Ordinance	GeCIP	Genomics England Clinical Interpretation Partnership
DH	Department of Health	GHPP	Genetic Health Promotion Programme
		GINA	Genetic Information Non-discrimination Act
		GIS	Genomic Institute of Singapore

GMCS	Genomic Medicine Centres	ISO	International Organization for Standardization	QMH	Queen Mary Hospital
GNA	Genetic Non-discrimination Act	IT	Information Technology	SG10K	Singapore 10K Genome Project
GNGC board	Genetic Nurse and Genetic Counsellor Branch Board	LDT	Laboratory-developed Test	SMPO	Supplementary Medical Professions Ordinance
HA	Hospital Authority	LIAJ	The Life Insurance Association of Japan	SSF GGS	Strategic Service Framework for Genetic and Genomic Services
HEE	Health Education England	MLTs	Medical Laboratory Technologists	SSI	Statens Serum Institut
HGSA	Human Genetics Society of Australasia	NGS	Next-generation Sequencing	TRANSCEND Consortium	Transomics Analysis of Complications and Endpoints in Diabetes Consortium
HKAM	Hong Kong Academy of Medicine	NHS	National Health Service	UK	The United Kingdom
HKCH	Hong Kong Children's Hospital	NHSE	NHS England	UKGTN	UK Genetic Testing Network
HKFI	The Hong Kong Federation of Insurers	NIH	National Institutes of Health	UN	United Nations
HKGI	Hong Kong Genome Institute	NPAAC	National Pathology Accreditation Advisory Council	US	The United States
HKGP	Hong Kong Genome Project	NPMS	National Precision Medicine Strategy	WGS	Whole Genome Sequencing
HKU	The University of Hong Kong	O&G	Obstetrics and Gynaecology		
HKUST	The Hong Kong University of Science and Technology	PCPD	Privacy Commissioner for Personal Data		
HMOs	Health Medical Organisations	PD(P)O	Personal Data (Privacy) Ordinance		
HOKLAS	Hong Kong Laboratory Accreditation Scheme	PMI	The Precision Medicine Initiative		
IEM	Inborn Errors of Metabolism	PWH	Prince of Wales Hospital		



Strategic Development of
genomic
MEDICINE IN HONG KONG

