Hong Kong
Genome Institute
香港基因組中心

Strategic Plan 策略計劃

2022-25

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Hong Kong Genome Institute 香港基因組中心

1. About the Institute

The Hong Kong Genome Institute (HKGI) is established and wholly owned by the Hong Kong Special Administrative Region (HKSAR) Government to propel the city's development of genomic medicine. By launching the Hong Kong Genome Project (HKGP), HKGI is committed to driving the application and integration of genomic medicine into clinical care. Riding on the Project, HKGI also aims at establishing genome database of local population, testing infrastructure and talent pool to advance research in genomic science and enhance public literacy as well as engagement to realise its vision – **to avail genomic medicine to all for better health and well-being**.

HKGP is the first large-scale genome sequencing project in Hong Kong. Through collaboration with the Department of Health (DH), the Hospital Authority (HA) and medical schools of local universities, eligible patients and their family members are recruited on a voluntary basis. The Project, which is implemented in two phases – the pilot phase and the main phase, is set to conduct whole genome sequencing (WGS) for 20,000 cases with the objective to provide participants with more precise diagnoses and personalised treatment. It also plays a significant role in disease surveillance and prevention, advancing the health of the general population in the long run.

1. 機構簡介

香港基因組中心（基因組中心）由香港特別行政區政府成立並全資擁有，肩負推動香港基因組醫學發展的重任。基因組中心透過推行「香港基因組計劃」（基因組計劃），致力推動基因組醫學與臨床護理的應用和融合，並藉此建立本地人口的基因組數據庫、測試設施及人才庫，促進相關領域的研究，同時加強公眾教育和參與，以實現**「普及基因組醫學，共享健康福樂」**的願景。

基因組計劃乃香港首個大型基因組測序計劃，由基因組中心與衞生署、醫院管理局及大學醫學院合作，招募合資格的病人及其家屬自願參與。計劃分兩個階段推行，包括先導階段及主階段，目標為合共 20,000 宗個案進行全基因組測序，期望為參加者提供更準確的診斷及更個人化的治療方案。長遠而言，基因組計劃亦有助加強疾病防控，提升市民大眾的整體健康水平。

With the management team formed and core function teams continued to be in place in 2021, the pilot phase of HKGP focusing on undiagnosed disorders and hereditary cancers was launched in July in the same year. As for the main phase which is set to be rolled out in 2022, the scopes will be expanded to cover cases related to “genomics and precision health” in order to include more diseases and research cohorts that would benefit from WGS.

In launching the Project, HKGI has already set up Partnering Centres at the Hong Kong Children’s Hospital (HKCH), Prince of Wales Hospital (PWH) and Queen Mary Hospital (QMH) to help recruit eligible participants with informed consent through referrals, while also keeping other relevant organisations closely engaged. The results of sequencing analysis will be fed back to patients once available to aid diagnoses and clinical services.

基因組中心的管理層及主要工作團隊於 2021 年陸續履新，並於同年 7 月開展基因組計劃的先導階段，重點涵蓋未能確診病症及與遺傳有關的癌症。至於主階段則會於 2022 年展開，並擴大範疇至與基因組學及精準醫學有關的個案，以涵蓋更多可受惠於全基因組測序的疾病及研究群組。

基因組中心已於香港兒童醫院、威爾斯親王醫院及瑪麗醫院設立夥伴中心，並積極與其他相關機構緊密合作，透過轉介招募合資格參加者，經他們知情同意後參與計劃，而相關測序分析的結果，將回饋予病人作診斷及臨床治療之用。

2. Vision, Mission and Core Values
願景、使命及核心價值

The strategic planning of HKGI is grounded in its vision, mission and core values, which guide its decisions in the planning, development and delivery of services.

基因組中心的策略規劃以其願景、使命和核心價值為基礎，指導基因組中心在其規劃、發展及提供服務所作的決策。

Vision

To avail genomic medicine to all for better health and well-being.

願景

普及基因組醫學，共享健康福樂。

—— 實現基因組醫學的廣泛應用，為大眾帶來健康、幸福和快樂。

Mission

To accelerate the integration of genomics into medicine by driving clinical application, advancing research, nurturing talents and enhancing genomic literacy.

使命

銳意推動基因組醫學的臨床應用、科學研究、人才培育及公眾教育，加快基因組學與醫學的融合。

—— 多管齊下，加快融合基因組學與臨床應用。

Core Values

The core values of HKGI are embedded in its logo of a five-colour double helix structure with dark green as the primary logo colour signifying the fundamental HKGI spirit of **professionalism and reliability**, as well as the lines in green, red, blue, and yellow, which apart from being the colour codes representing ATCG (A - Adenine, T - Thymine, C - Cytosine, and G - Guanine), the four different bases of DNA nucleotides, also symbolise the HKGI values of **"health and new life"**, **"passion and dedication"**, **"hope and happiness"**, and **"versatility and energy"** respectively.

核心價值

基因組中心的標誌設計與其核心價值相互呼應。標誌以深綠色為主調，象徵**專業與可靠**，是團隊所秉持的基本精神。標誌上雙螺旋結構的 DNA 長鏈，由五色線條組成，在深綠色以外，其餘綠、紅、藍、黃四色均各有所喻，不但代表 ATCG (A - Adenine, T - Thymine, C - Cytosine, and G - Guanine) 四種 DNA 代碼，也分別代表基因組中心堅守的四大核心價值及理念，包括**健康與新生**，**熱誠與專注**、**希望與快樂**，及**多元與活力**。

Hong Kong Genome Institute
香港基因組中心

Dark Green = Professionalism and Reliability
深綠色 = 專業與可靠

T (Thymine) Red = Passion and Dedication
紅色 = 熱誠與專注

G (Guanine) Yellow = Versatility and Energy
黃色 = 多元與活力

C (Cytosine) Blue = Hope and Happiness
藍色 = 希望與快樂

A (Adenine) Green = Health and New Life
綠色 = 健康與新生

Core values upheld by HKGI
基因組中心秉持的核心價值

Benefits brought to stakeholders
為持份者帶來的裨益

Professionalism and Reliability

To provide WGS, laboratory, genetic counselling, genetic education, bioinformatics, research facilitation and related services with professionalism and reliability, observing relevant professional guidelines, ethical codes, standardised protocols as well as principles of data privacy and security.

專業與可靠

以專業及可靠的精神，為持份者提供全基因組測序及其他相關服務，包括實驗室、遺傳輔導及教育、生物信息學及研究等範疇，並遵守相關專業指引、道德守則、數據私隱和安全的標準規程及原則。

Passion and Dedication

To achieve HKGI's vision and mission with passion and dedication, working wholeheartedly, thinking positively and taking the initiative to go the extra mile to serve patients and the wider community in a better way.

熱誠與專注

以熱誠和專注的態度實現基因組中心的願景和使命，全情投入，樂觀積極，加倍努力，致力為病人和社會大眾帶來更大裨益。

Versatility and Energy

To adopt a multidisciplinary approach for engaging professionals from various disciplines to promote the development of genomic medicine with vibrant means, energetic efforts and teamwork, embracing the spirit of openness, mutual respect, and acceptance of different ideas.

多元與活力

採取多元及跨專業的方針，廣泛接觸及聯繫不同界別的專家，以充沛的活力及團隊精神攜手推動基因組醫學的發展，並秉持開放的態度，互相尊重，廣納不同意見。

Hope and Happiness

To bring hope and happiness to patients and their families by fostering the integration of genomic medicine into clinical care to improve genomic diagnosis, personalised treatment and prevention of diseases.

希望與快樂

促進基因組醫學與臨床護理的融合，以優化基因組診斷、個人化治療和疾病防控，為病人及其家屬帶來希望和快樂。

Health and New Life

To promote health and better quality of life amongst patients and the people of Hong Kong by facilitating the advancement of knowledge and technology in genomic medicine through vigorous research as well as the translation of research breakthroughs into clinical practice.

健康與新生

透過推動研究及將其相關成果轉化為臨床應用，促進基因組醫學的知識和技術發展，藉此提升病人及市民大眾的健康和生活質素。

Executive Summary 摘要

The Hong Kong Genome Institute Strategic Plan 2022-2025 is the overarching document for guiding all aspects of the Institute's development and planning in the coming three years. In particular, it provides the basis on which its executives develop the annual plan programme initiatives through a longer-term planning approach.

Many of the strategies and key actions set out in the Strategic Plan dovetail with the plans for implementing the recommendations of the Government's Steering Committee on Genomic Medicine (Steering Committee), as part of a coherent and synergistic approach for positioning the HKGI to address key challenges and issues and move towards achieving its vision and mission.

1. Planning Process

Building on the recommendations of the Steering Committee, the planning process of this Strategic Plan was led by the HKGI Board of Directors, some members of which had previously served on the Steering Committee. The Plan has been developed through a process of realistic analysis of HKGI's internal and external environment and consultation with key stakeholders. From the process, four main strategic foci on the development of genomic medicine in Hong Kong have been consolidated along with a number of strategies, which map out the corporate priorities for HKGI to work towards addressing the key challenges it faces in the next three years.

《香港基因組中心2022-2025年策略計劃》(《策略計劃》)是指引基因組中心未來三年全面發展及規劃的總體綱領，透過長遠規劃的方式，為管理團隊制訂年度工作計劃提供基礎。

《策略計劃》臚列的各項策略和主要行動項目，均與落實由香港特別行政區政府成立的基因組醫學督導委員會（督導委員會）所提出的建議一致，有助基因組中心以連貫合一、協同增效的方針應對主要挑戰和問題，為實現其願景及使命向前邁進。

1. 規劃過程

《策略計劃》以督導委員會的建議為基礎，由基因組中心董事局帶領規劃，當中部分成員亦曾參與督導委員會的工作。制訂《策略計劃》的過程務實及全面，深入分析了基因組中心的內外環境，並諮詢了主要持份者的意見。過程中，基因組中心亦就推動香港基因組醫學發展確立了四大策略重點及一系列相關策略，為機構的各項工作訂立優次，以應對未來三年的主要挑戰。

2. Strategic Foci

Integrate genomic medicine into clinical care - Driving the incorporation of genomic medicine into mainstream clinical service in Hong Kong by improving genomic diagnosis, personalised treatment as well as personalised prediction and prevention of disease risks. This is done by showcasing the clinical usefulness of WGS in focused disease areas and the functional/economic benefits of embedding genomics into routine clinical care.

Advance research in genomic science - Facilitating genomic science and discoveries by establishing a flexible platform with rich database for new genomic technologies and multi-omics studies, as well as disease-focused research networks through local and international collaborations. Core to this will be setting up the necessary infrastructure including facilitating the development of a local biobank network and promoting local and international collaborations in genomic research.

Nurture talents in genomic medicine - Developing skilled and competent professionals to deliver genomic medicine through collaborations with local universities, professional bodies and healthcare institutions to enhance training and development of the related professions, including organising continuing professional development programmes for clinicians, genetic counsellors, bioinformaticians and medical laboratory technologists, etc.

Enhance public genomic literacy and engagement - promoting genomic literacy in Hong Kong, particularly amongst healthcare workers and students, by engaging and collaborating with relevant government departments, schools, universities and non-government organisations to initiate public education programmes in genetics and genomics.

2. 策略重點

融合基因組醫學與臨床護理 — 透過優化基因組診斷、個人化治療及患病風險的預測和預防，推動基因組醫學融入香港主流臨床服務。此策略重點旨在展示全基因組測序於重點疾病領域的臨床成效，以及將基因組學融入常規臨床護理可帶來的實效/經濟效益。

促進基因組科學研究 — 透過為嶄新基因組學技術及多組學研究建立靈活且數據豐富的平台，並加強本地及國際合作，建立以防治疾病為主的研究網絡，從而促進基因組科學研究。此策略重點的關鍵在於為香港建立必要的基礎設施，包括推動設立本地生物樣本庫網絡，及鼓勵本地和世界各地相關機構就基因組研究進行更多交流協作。

培育基因組醫學人才 — 透過與本地大學、專業團體和醫療機構合作，培育知識及技能兼備的專業人才，以提供基因組醫學服務。同時，通過與業界合作強化相關專業的培訓和發展，包括為醫生、遺傳輔導員、生物信息學家及醫學實驗室技術人員等舉辦持續專業發展課程。

加強公眾對基因組學的認識和參與 — 與有關政府部門、學校、大學及非政府機構合作，推行遺傳學及基因組學相關的公眾教育活動，以提高市民大眾，尤其是醫護人員和學生的關注和認識。

3. Strategy for Improving Genomic Diagnosis and Personalised Treatment

- Provide standardised high-quality genomic testing by enhancing WGS capability and capacity; enhancing bioinformatics capability and capacity; obtaining accreditation for HKGI laboratory & WGS pipeline; and enhancing provision of genetic counselling service, including tele-counselling.
- Facilitate provision of more personalised treatment by promoting effective use of genetic diagnosis and pharmacogenomic profiling for targeted treatment.

3. 優化基因組診斷和個人化治療的策略

- 提升全基因組測序及生物信息學的能力及效能；為基因組中心的實驗室及全基因組測序流程取得認證；並加強包括遙距諮詢在內的遺傳輔導服務，以提供標準化及優質的基因組測序服務。
- 提倡善用基因組診斷及藥理基因組分析於針對性的治療，以推動實踐個人化治療。

4. Strategy for Improving Personalised Prediction and Prevention of Disease Risk

- Establish a clinical genomic database of the local population by expanding the HKGP patient cohorts through opening up new recruitment channels; and enhancing guidelines and standardised protocols on data privacy and security.
- Improve risk prediction for diseases prevalent in Hong Kong by developing polygenic risk scores for common diseases in local population; and refining risk prediction by integrating genotyping, deep phenotyping, health-related data and medical records.

5. Strategy for Advancing Research in Genomic Science

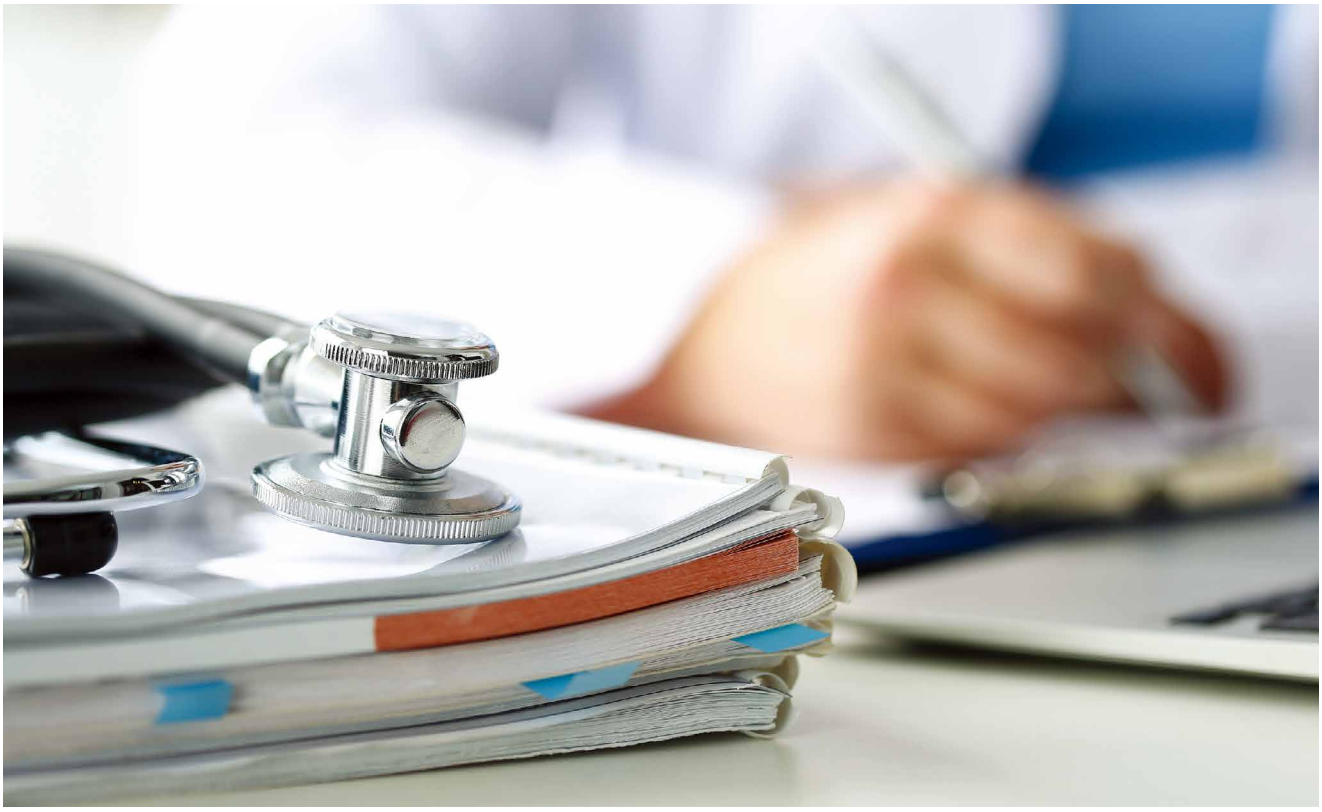
- Establish database and platform to facilitate new genomics technology and multi-omics studies by developing functional assays to characterise, annotate and interpret genes/variants.
- Develop new genomic technologies for clinical implementation.
- Engage industrial partners to translate findings into clinical use.
- Establish disease-focused local and international research networks by identifying specific disease themes for partnering with academic institutes to conduct relevant research studies; and publishing the project implementation experience of HKGP for sharing.

4. 優化個人化疾病預測和預防的策略

- 開闢新的招募渠道，擴大基因組計劃的參與群組，並優化私隱及數據安全指引和標準規程，以建立本地人口的臨床基因組數據庫。
- 制訂香港人口常見疾病的多基因風險評分；整合基因型和深度表型資料、健康相關數據及醫療紀錄，優化患病風險預測，藉此改善香港流行疾病的風險預測。

5. 促進基因組科學研究的策略

- 開發功能性研究，以歸納、註釋及詮釋各種基因變異模式，透過建立數據庫及平台，促進新基因組學技術及多組學研究。
- 開發新基因組技術供臨床應用。
- 與業界夥伴合作，轉化研究成果至臨床應用。
- 鑑辨特定疾病主題，與學術機構合作進行相關的基因組學研究；發布推行基因組計劃的經驗以供分享，藉此建立以防治疾病為主的本地及國際研究網絡。



6. Strategy for Establishing Infrastructure for Implementing Genomic Medicine

- Facilitate the development of a local biobank network for genomic research by establishing the HKGI biobank to enable data sharing for research; and enhancing HKGI's guidelines and standardised protocols on informed consent, collection, storage and responsible sharing of genomic data.

- Facilitate clinical implementation of genomic medicine by evaluating clinical usefulness of WGS in focused disease areas to embed genomic medicine and promote evidence-based research; and evaluating health economics and outcomes of WGS in focused disease areas to embed genomic medicine.

6. 建立發展基因組醫學基礎設施的策略

- 建立基因組中心的生物樣本庫，讓數據可共享作研究之用；以及完善基因組中心就知情同意及基因組數據的收集、儲存及適切分享所制訂的指引和標準規程，藉此促進本地生物樣本庫網絡的發展，推動基因組研究。

- 評估全基因組測序在重點疾病領域的臨床成效，以促進基因組醫學的應用及循證研究；並評估醫療經濟學及全基因組測序結果在重點疾病領域的效益，藉此推動基因組醫學於臨床的應用。

7. Strategy for Enhancing Genetic and Genomic Knowledge and Professional Development

- Engage with professional bodies to strengthen continuing professional development by supporting continuing professional development programmes in genetics and genomics for clinicians, nurses, and allied health professionals, including genetic counsellors and bioinformaticians; and partnering with professional societies to develop genetic counselling guidelines and standards.
- Incorporate experiential learning into continuing education programmes by engaging staff of Partnering Centres in multidisciplinary team meetings; and establishing "genomics champion" in different specialties to contribute and influence genomic education and practice.

7. 強化遺傳學和基因組學知識及專業發展的策略

- 支持及強化醫生、護士、專職醫護人員包括遺傳輔導員及生物信息學家等的持續專業發展計劃，並與專業團體合作，制訂遺傳輔導指引及標準，以促進持續專業發展。
- 鼓勵夥伴中心的人員參與跨專業團隊會議，並於不同專科選出「基因組學傑出人員/團隊」，樹立楷模，為基因組學的教學和實踐提供實例參考，把經驗學習納入持續進修計劃之內。

8. Strategy for Improving Genetic and Genomic Knowledge of Tertiary Students

Engage with local universities to promote genomic medicine by:

- Collaborating and supporting local universities to develop and organise courses in genomics, bioinformatics, biomedical science, and genetic counselling, and promotional events such as information week and career fair.
- Developing and administering enrichment and internship programmes for undergraduate students in genomic medicine related studies.
- Supporting local postgraduate programmes in genomics, bioinformatics and biomedical science.

8. 增進大專學生遺傳和基因組學知識的策略

與本地大學合作推廣基因組醫學，包括：

- 支持本地大學開辦及推廣基因組學、生物信息學、生物醫學及遺傳輔導等課程，並合辦資訊週及職涯規劃講座等宣傳活動。
- 為修讀基因組醫學相關課程的大學本科生，開辦進修課程和實習計劃。
- 支持基因組學、生物信息學及生物醫學的研究生課程。

9. Strategy for Improving Public Awareness and Knowledge of Genomics

- Engage the general public and targeted stakeholders to enhance public understanding of genomic medicine by developing authoritative and user-friendly information and publications on genomic medicine; and formulating strategic engagement plans for targeted stakeholders, including patient groups and professional bodies, to promote awareness of genomic medicine and its benefits.

9. 提升公眾關注和認識基因組學的策略

- 印製具權威性及淺白易明的資訊和刊物，深入淺出介紹基因組醫學，以提升公眾的認識；及制訂策略主動接觸特定持份者，包括病人組織和專業團體，以加深他們對基因組醫學及其效益的認識。

10. Functional and Economic Benefits of Implementing the Strategies

By implementing the strategies for promoting the development of genomic medicine in Hong Kong, HKGI seeks to use the accumulated clinical genomic data of the local population to generate robust functional impacts on the health and well-being of the people of Hong Kong, including deeper insights into disease biology, advancements in biomedical science, better understanding of predispositions to diseases prevalent in Hong Kong, more accurate diagnosis of certain genetic diseases, and significant improvements in the prediction and treatment of diseases.

It is expected that implementation of the strategies would also yield a significant direct economic presence in the economy of Hong Kong by reducing the costs of our healthcare system, helping industries and start-up companies to expand business using the genomic knowledge and data generated, and driving greater growth in research expenditure, investment and employment in the genomics-related fields.

11. Implementation and Monitoring

Strategies and key actions of the HKGI Strategic Plan 2022-25 will be implemented through the annual planning process. The three Annual Plans covering the period 2022-23 to 2024-25 will be the specific action plans for implementing this Strategic Plan.

Monitoring of the implementation of the Strategic Plan will be led and overseen by the HKGI Board and its relevant Committees, and reported to the Food and Health Bureau from time to time. Progress on the implementation will be reported in the Annual Report of HKGI.

10. 推展策略帶來的實效和經濟效益

基因組中心推展上述策略，旨在促進香港基因組醫學的發展，並致力透過建立和善用本地人口的臨床基因組數據庫，期望為社會大眾帶來豐碩成果，與市民共享健康福樂。其中的主要效益將包括：增進對疾病生物學的了解、促進生物醫學的科學發展、加深認識本地流行病的健康風險、為若干遺傳病患提供更準確診斷，及為防治疾病帶來重大改進。

基因組中心預期，推展有關策略有助減低醫療系統的成本、協助業界及初創公司使用積累的基因組知識及數據拓展業務，並帶動基因組學相關範疇的研究開支、投資及就業，直接為香港的經濟發展注入重大動力。

11. 執行和監察

基因組中心將透過其年度計劃，推展擬訂於《策略計劃》內的策略及主要行動項目，並將通過 2022-23 年至 2024-25 年期間的三個年度計劃，轉化各項目為具體行動，以執行《策略計劃》。

監察《策略計劃》執行進度的工作將由基因組中心董事局及相關委員會領導，並定期向食物及衞生局匯報。有關的執行進度，亦會刊於基因組中心的年度報告。

Foreword by Chairperson

Since its inception in May 2020, the HKGI has been fully engaged in setting up the hardware and software for implementing the HKGP to pioneer the development of a clinical genomic database for the population of Hong Kong for integrating genomic knowledge and research breakthroughs into clinical care. With commencement of participant recruitment for the HKGP in July 2021, it is imperative for each and every one of us in the HKGI to use our professional knowledge and experience to plan ahead in a longer term and work together to ensure the best possible outcomes for our efforts to promote the development of genomic medicine.

The exercise to map out our strategic directions for the next three years is a continuation of the journey we have just embarked upon one year ago to make genomic medicine available to more patients and bring health and economic benefits to our society. Our strategic planning process is guided by the recommendations of the Steering Committee on Genomic Medicine as well as the latest local and international developments in genomic medicine, which have enabled us to consolidate our experience and build a solid foundation for future development.

The formulation of this Strategic Plan has involved the contribution of many people and I am indebted to the valuable time and inputs given by the Food and Health Bureau, my fellow Board and Committee members, the HKGI executives, staff of our Partnering Centres and institutions, as well as patient representatives.

Through the collective strength and dedication of all members of the HKGI, I have full confidence that the strategies and priorities set out in this plan will be turned into effective actions for advancing the medical development in Hong Kong by integrating genomic medicine into clinical care, nurturing talents, enhancing public genomic literacy and promoting research in the field.

Philip TSAI Wing-chung, BBS, JP
Chairperson

Introduction by Chief Executive Officer

Building on the solid foundations of the Government's policy directions and recommendations of the Steering Committee on Genomic Medicine, the HKGI Strategic Plan 2022-2025 sets out the strategies and directions we will pursue in the next three years to advance the development of genomic medicine for bringing benefits on the health and well-being of the people of Hong Kong.

During the strategic planning process, we have crystallised four main strategic foci pertaining to the service, research, talent development, and educational aspects of our work along with an array of strategies, which map out the corporate priorities for us to move towards achieving our vision and mission.

On the service aspect, we will focus on integrating genomic medicine into clinical care by going after the strategic goals of improving genomic diagnosis and personalised treatment, enhancing personalised prediction and prevention of disease risk as well as establishing the infrastructure for clinical implementation of genomic medicine. Our vision in this aspect is "to advance genomic medicine for better personalised care", helping patients with undiagnosed disease to "see the unseen" and making personalised treatment available to many more patients.

On the research aspect, we will emphasise on facilitating local and global research on human genomes to contribute to the broader development of biomedical science in Hong Kong. Our vision in this aspect is to provide a flexible platform with rich database for researchers to advance scientific discoveries and support Hong Kong to become the leading research hub of biomedical science.

On the talent development aspect, we will emphasise on nurturing talents in genomic medicine through the pursuit of the strategic goals of enhancing genetic and genomic knowledge and professional development of practitioners in the genomics-related professions, and improving the genetic and genomic knowledge amongst healthcare professionals and university students. Our vision in this area is to have skilled professionals to deliver and develop genomic medicine services.

Regarding the educational aspect, we will seek to enhance public genomic literacy and engagement by raising awareness of the "genomic revolution" amongst targeted stakeholders, including patients, youths and the general public. Our vision in this respect is to achieve a high level of genomic literacy in the varied constituencies comprising the public to facilitate the implementation of genomic approaches to healthcare.

Since commencement of participant recruitment for the HKGP in July 2021, I have heard many untold stories about patients' expectations of genomic medicine, whereby they hope could bring them health, happiness and new life. We may not be able to fully meet these expectations due to various constraints, but I firmly believe that as long as we uphold the core values of HKGI to deliver and develop genomic services with professionalism, reliability, passion, dedication, versatility and energy, we can bring substantial and tangible benefits to patients and our society.

I wish to express my sincere gratitude to members of the Board and Committees for their guidance and support. My heartfelt appreciation also goes to the Food and Health Bureau, my HKGI colleagues as well as our partners in the Hospital Authority, the Department of Health, universities, and patient groups for their valuable contributions to the formulation of this strategic plan. I look forward to the continued support and participation of all in bringing the plan into fruition.

Dr LO Su-vui
Chief Executive Officer

Planning Context

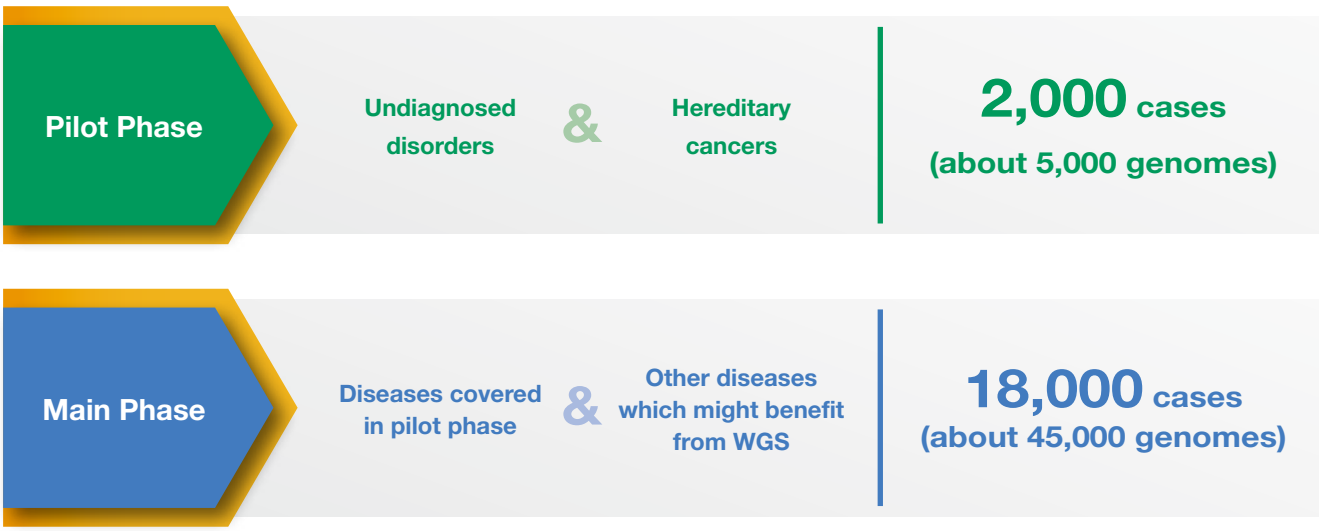
1. Policy Direction for Development of Genomic Medicine

To formulate a roadmap for the next three years, HKGI has developed its first Strategic Plan to provide a framework for everyone in HKGI and its partnering institutions to align their priorities and efforts with the corporate directions and strategies in a consistent way. The objective is to strategically position HKGI so as to enable the organisation to address key challenges, exploit opportunities and move towards achieving its vision and mission.

This first HKGI Strategic Plan, traversing the next three years from 2022 to 2025, builds on the progress of work achieved by the Steering Committee set up by the Secretary for Food and Health of the HKSAR to lead the study on strategies for developing genomic medicine in Hong Kong pursuant to the announcement in the Chief Executive's 2017 Policy Address.

Based on the preliminary recommendation of the Steering Committee that a large-scale genome sequencing project should be conducted in Hong Kong with a view to enhancing the clinical application and promoting innovative scientific research on genomic medicine, the Chief Executive announced in the 2018 Policy Address that the Government would provide funding to introduce the HKGP and establish separately the HKGI, a limited by guarantee company wholly owned by the Government, to coordinate the implementation of HKGP and drive the collaboration of existing infrastructure and expertise for maximum synergy and innovation. Following announcement in the Chief Executive's 2019 Policy Address, HKGI was incorporated in May 2020.

Since its inception, HKGI has followed the Government's policy direction to plan for the implementation of HKGP to perform 20,000 cases involving about 50,000 WGS in two phases. The pilot phase (2,000 cases involving about 5,000 genomes), which has commenced in July 2021, will cover patients with undiagnosed disorders and cancers with clinical clues linked to possible hereditary components. The main phase (18,000 cases involving about 45,000 genomes), scheduled for implementation from July 2022 to 2025, may expand the coverage to other diseases and research cohorts which could benefit from WGS, subject to the progress of the pilot phase.



According to the Government's policy direction, 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.

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.

2. Priority Areas

Guided by the Government's direction set forth by the Steering Committee in its report published in 2019, HKGI has undertaken its strategic planning by according priorities to the following work areas:

- Launching the HKGP
- Enhancing clinical services in genetics and genomics
- Nurturing talents in genomic medicine
- Enhancing public engagement in genomic medicine
- Enhancing the laboratory network with effective referral mechanism and centralisation of advanced genetic and genomic tests
- Facilitating the establishment of a biobank network for genomic research
- Enhancing the regulation on use of genetic data for insurance and employment purposes
- Promoting the proper use of genetic and genomic tests

Planning Process

1. Project Governance

The formulation of the HKGI Strategic Plan 2022-2025 followed the policy directions set by the HKSAR Government in 2020 through acceptance and publication of the Report of the Steering Committee. This policy guidance continued after inception of HKGI in 2020 through the three HKGI official Directors and appointment of five expert and institutional members of the Steering Committee, including its Chairman, Professor Raymond LIANG Hin-suen, to the HKGI Board of Directors. Another four non-official members of the Steering Committee have also been appointed to serve on the functional committees of the HKGI Board. Building on the recommendations of the Steering Committee, the planning process of this Strategic Plan was led and directed by the HKGI Board of Directors, with members of the Board and its relevant committees involved in the development process. Above all, the HKGI Board is the ultimate authority for confirming and approving the strategies and directions of the Strategic Plan.

Executive decision was provided through the formation of a working group chaired by the Chief Executive Officer to oversee the planning process. Membership of the Working Group comprised the heads of HKGI's three branches, namely the Scientific Branch, Bioinformatics Branch and Administration Branch, as well as division/unit heads of key functions including laboratory, data security, genetic counselling, corporate communications and Board secretariat.

2. Formulation Process

Overall, the process for formulating the Strategic Plan involved the following components:

Formulation Process of the Strategic Plan

- Environmental scanning and situation analysis of HKGI's internal and external context to identify key challenges and issues.
- Review of the Report of the Steering Committee and deliberations of its working groups as well as other relevant policy documents of the Government to take stock of the progress and map out any prevailing issues and gaps.
- Formulation of a Strategic Framework for the next three years to identify the strategic foci, set the strategic goals, map out the strategic directions and draw up strategies for progressively achieving HKGI's vision and mission.
- Analysis of the key enablers for successfully accomplishing the proposed strategies to support enhancements, drive change and facilitate development of genomic medicine in Hong Kong and setting out a mechanism for monitoring implementation of the strategies through the annual planning process.
- Consultation and discussion with key stakeholders to solicit their views and inputs on the key issues facing HKGI and the proposed strategies for addressing them.

Environment and Key Challenges

1. Overview

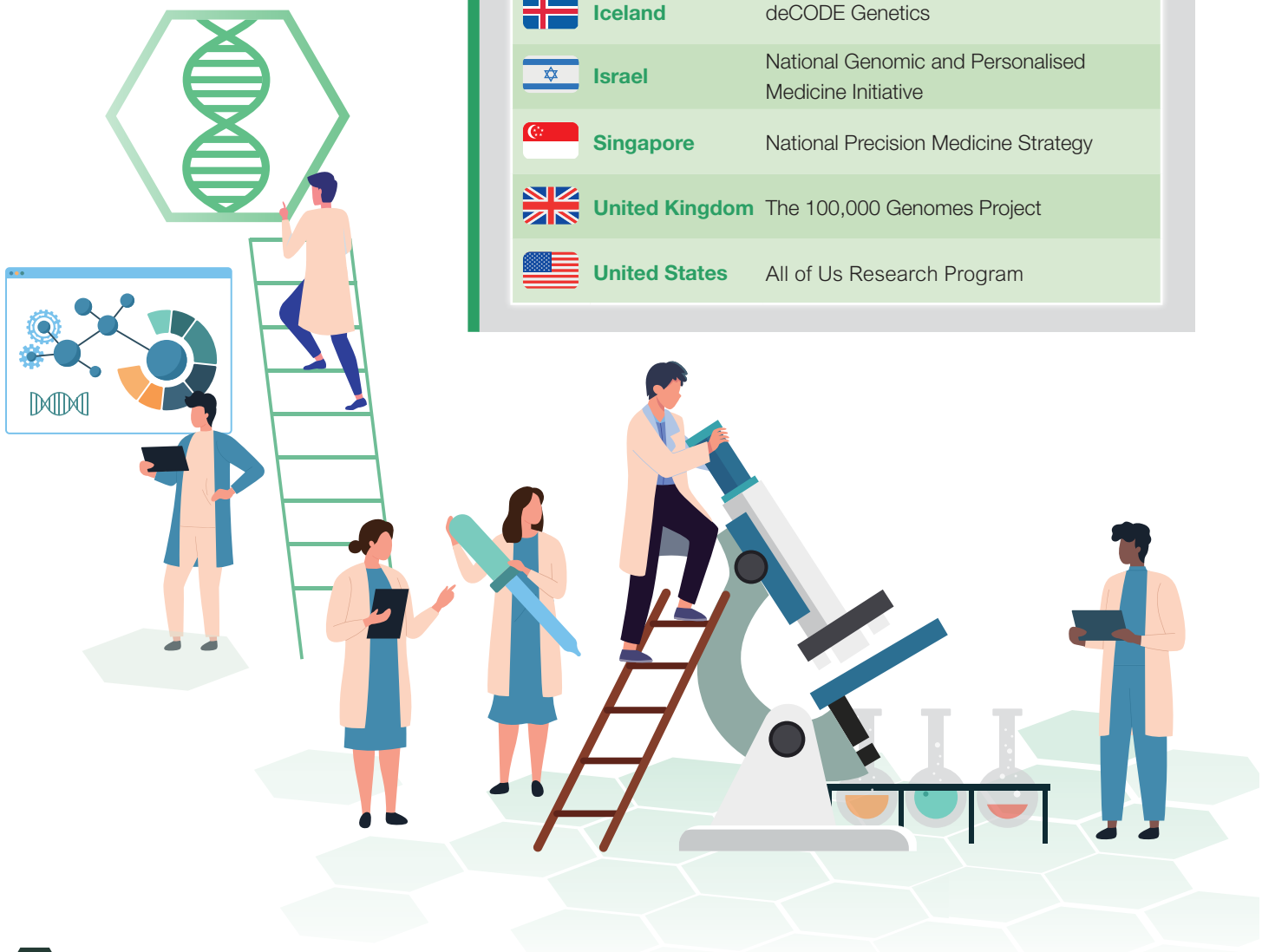
Tasked with the mission of advancing the medical development in Hong Kong by integrating genomic medicine into clinical care, nurturing talents, enhancing public literacy and promoting research in the field, HKGI will need to better understand the key issues and challenges facing the organisation now and in the coming years. HKGI has examined the following aspects of internal and external environments during the strategic planning process:

- Global development of genomic medicine
- Current landscape of genetics and genomics in Hong Kong
- Clinical service provision
- Laboratory services and translation of new technology to clinical use
- Workforce situation
- Genomic literacy
- Ethical, legal and social implications

2. Global Development of Genomic Medicine

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, countries around the globe have formulated policies and strategies for promoting the development of genomic medicine in their jurisdictions with implementation of several large-scale genome projects throughout the last decade. The major projects reviewed during the strategic planning process are briefly described below.

China	100,000 Chinese People Genome Project
Denmark	Danish Reference Genome Project
Iceland	deCODE Genetics
Israel	National Genomic and Personalised Medicine Initiative
Singapore	National Precision Medicine Strategy
United Kingdom	The 100,000 Genomes Project
United States	All of Us Research Program



CHINA – 100,000 Chinese People Genome Project

The 100,000 Chinese people genome project is a precision medicine research project under the national key research and development plan. The project, led by the Harbin Institute of Technology, has started since December 2017. It involves the sequencing of 100,000 healthy subjects from different ethnic backgrounds and regions across China (the Han and nine selected minorities including the Zhuang and the Hui) and collection of their phenotype and environmental exposure data. The primary objective is to establish a reference database of genomic variation and health map for the Chinese population to facilitate development of personalised medicine.

DENMARK – Danish Reference Genome Project

GenomeDenmark is a national platform for sequencing and bioinformatics, which includes universities, hospitals and private firms. The main objective of GenomeDenmark is to establish a platform with research infrastructure to develop know-how, advance national coordination and create synergy within the field of genomics through broad cooperation across research fields and sectors. Genomic references are important and fundamental tools because they facilitate analyses of individual patients and their genes, including how hereditary disorders arise.

One of GenomeDemark's key projects is to establish a high-quality Danish reference genome, in order to generate knowledge that can support the development of personalised treatment, based on genomic information in the healthcare system. The project also generates knowledge that can be applied to the Danish pharmaceutical and food industries. The project de novo assembled the genomes of 150 healthy Danes selected to represent the average citizens in order to uncover the genetic variations present in the Danish population. The catalogue of genomic variations from all donors constitutes a Danish reference genome of high quality. The reference helps determine the structure and development history of the Danish genome and serve as a tool for research and development of genomics and public health.

Denmark has established a Danish National Genome Centre to offer genetic services as part of a more targeted treatment to at least 60,000 patients over the next three years.

ICELAND – deCODE Genetics

Genome sequencing of the Iceland population has been led by deCODE, a genome sequencing and analysis company based in Iceland. Using Iceland's uniquely comprehensive genealogical records, deCODE has also put together a genealogy database covering the entire present-day population and stretching back to the founding of the country. The database has been very useful in research including the detection of de novo mutations (new mutations which are not known before).

Since its establishment in 1996, deCODE has gathered genotypic and medical data from more than 160,000 volunteer participants, comprising well over half of the adult population of Iceland. DeCODE has been identifying disease-related variants since it started, by correlating their genetic database with medical data collected from Iceland.

Some results of deCODE's analysis have already been published in renowned journals such as Nature Genetics, including the identification of a new Alzheimer's-associated gene and the characterisation of structural variants on a population scale, using long-read sequencing technology. These findings will help guide medical research and the understanding of human evolution, through understanding of sequence diversity.

ISRAEL – National Genomic and Personalised Medicine Initiative

The Israeli government started a national initiative in 2018 to develop a genomic and clinical data research platform aiming to improve its digital health technology and infrastructure to benefit the Israeli population. It has planned to spend about NIS 1 billion to support the National Genomic and Personalised Medicine Initiative, which aims to sequence over 100,000 patients' genomes by 2023 with a view to improving healthcare services provided to targeted patients. The multidisciplinary programme will also establish a national database for health researchers in genetics and medical information to identify long-term disease trends of Israeli citizens. Researchers who would like to enquire about participants' genomic data can apply for access to the database.

SINGAPORE – National Precision Medicine Strategy

Singapore's National Precision Medicine (NPM) strategy, being a 10-year plan to enhance and accelerate Singapore's biomedical research, health outcomes and economic growth, was launched in 2017. The NPM Phase I was implemented in the same year to establish an at-scale infrastructure and to map out the genomic profile of Singaporeans according to their three main ethnicities (namely Chinese, Malay and Indian). Completed in October 2019, the NPM Phase I databank comprises the complete genetic data of 10,000 healthy Singaporeans, serving as a reference for Asian genetic normality and underpinning the subsequent development of precision medicine for Singaporeans and patients across Asia.

The NPM Phase II has started in April 2021 with a four-year horizon and it aims to:

- further research insights into the Asian phenotype by analysing the genetic makeup of 100,000 healthy Singaporeans and up to 50,000 patients with specific diseases;
- improve patient outcomes by piloting the implementation of precision medicine in clinical practice; and
- create new economic opportunities for Singapore's healthcare and biomedical technology industry by attracting and anchoring overseas companies while yielding new opportunities for home-grown enterprises.

To achieve this, Precision Health Research, Singapore (PRECISE), has been set up, as the central entity to drive NPM strategy.

UNITED KINGDOM – The 100,000 Genomes Project

The United Kingdom (UK) government announced in December 2012 a programme of WGS, i.e., the 100,000 Genomes Project, as part of its Life Sciences Strategy. The principal objective of the Project was to sequence 100,000 genomes from patients with cancer and rare disorders, etc. and to link the sequence data to a database with standardised, extensible medical information of diagnosis, treatment and outcomes with a view to producing new capability and capacity for genomic medicine to transform the National Health Service (NHS).

The 100,000 Genomes Project involved 13 newly created NHS Genomic Medicine Centres, 85 NHS Trusts and 1,500 NHS staff at a cost of about GBP 300 million. The targeted 100,000 genomes were completely sequenced in December 2018, taking a total of around six years. NHS is now equipped with enhanced genomic medicine service with the required infrastructure established and has planned to sequence over five million genomes by 2024.

In 2021, the UK government has set out a new vision for genomic medicine, i.e., "to create the most advanced and integrated genomic research healthcare ecosystem in the world" for NHS. It aims to use its genomic research assets to drive the next generation of Life Sciences discoveries, deliver genomics-enabled clinical trials and support the growth and Research & Development of innovative genomics-focused companies by:

- continuing to support and enhance its genomic research infrastructure;
- evaluating variants and their role in prediction and public health;
- utilising new genomic tools to improve prediction and early diagnosis capabilities;
- bringing the best emerging science and technology to bear on cancer diagnosis and treatment; and
- delivering a world class offer on functional genomics.

UNITED STATES – All of Us Research Program

The Precision Medicine Initiative (PMI) of the United States (US) was launched in fiscal year 2016 when USD 130 million and USD 70 million were allocated to the National Institute of Health to build a cohort with national large-scale research participant group and the National Cancer Institute to lead efforts in cancer genomics respectively. As a key element of the PMI, the All of Us Research Program was implemented in 2018 to gather data from one million or more people living in US to accelerate health research and medical breakthroughs with a view to enabling individualised prevention, treatment and care for the whole population. By taking into account individual differences in lifestyle, environment and biology, researchers aim to uncover paths towards delivering precision medicine.

3. Current Landscape of Genetics and Genomics in Hong Kong

Clinicians at DH, 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. During the strategic planning process, the current landscape of services in genetics and genomics in Hong Kong was reviewed to identify the issues and challenges facing HKGI in the next three years.

Clinical Genetic Service of DH

The Clinical Genetic Service (CGS) of DH has been the main provider of public genetic and genomic services on a territory-wide basis in Hong Kong since 1981. CGS handles about 1,800 new cases every year, with a cumulative caseload involving more than 43,000 families (up to 2020) since its establishment. As the CGS does not offer treatment services, patients with treatment needs are referred to respective specialties at HA for follow-up management.

In addition to clinical diagnostic and genetic counselling service, CGS also provides neonatal screening service for Congenital Hypothyroidism (CHT) and Glucose-6-Phosphate Dehydrogenase (G6PD) Deficiency since 1984, and collaborates with HA for the overall planning, coordination and evaluation of the Newborn Screening Programme for Inborn Errors of Metabolism (IEM), which was regularised in 2017 and has been extended to all public birthing hospitals in October 2020.

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.

Genetic and Genomic Services in HA

HA has issued the Strategic Service Framework for Genetic and Genomic Services on 14 October 2019, setting out five strategic directions to improve its genetic and genomic (G/G) services:

- Strengthen the coordination and collaboration of different G/G services to enhance service quality and accessibility;
- Provide timely financial support to G/G service provision and development to help keep pace with G/G advancements;
- Enhance the governance of G/G services for better coordination;

- Nurture skilled and competent G/G professionals in HA; and
- Promote performance monitoring for continuous quality improvement.

By formulating the strategic service framework, HA looks to keep in pace with the global development of genomic medicine and leverage on the huge potential of G/G innovations and advancements to benefit patients of public hospitals in the years to come.

Genetic and Genomic Services and Research in Universities

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.

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's 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.

Genetic and Genomic Services in Private Sector

In addition to the public sector and universities, part of the increasing demands for CGS in Hong Kong have been met by the private sector. In 2016, a private hospital set up its own CGS, 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.

At the same time, direct-to-consumer genetic tests 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.

4. Key Issues and Challenges

A review of the development in genomic medicine around the globe and in the local environment has indicated that in context of the global trend of moving towards integrating genetics and genomics into mainstream clinical practices and the opportunities so generated for undertaking personalised and precision medicine, 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. A number of issues and challenges have to be addressed to set the scene for meeting the service needs of today and laying the foundations for our healthcare system to leverage on the huge potential of genetic and genomic innovations and advancements to benefit patients in the years to come. These include rationalising the clinical service provision, setting a standardised and coordinated mechanism for evaluating the clinical validity and utility of new genetic and genomic tests developed by various institutions to facilitate transfer of new technology into clinical use, improving the workforce situation as well as enhancing genomic literacy amongst healthcare professionals and the general public.

5. Clinical Service Provision

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, HA has just started to provide dedicated genetic and genomic services in public hospitals. Also, 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. HKGI will formulate strategies to address these issues and challenges through the implementation of the HKGP.

6. Laboratory Services

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. Although HA has recently established dedicated G/G services at Hong Kong Children's Hospital, patients and clinicians of different hospitals still do not have equitable access to G/G testing. In addition to inequitable access to G/G testing, there is a mismatch between the provision of laboratory tests and overall clinical needs, particularly in the inadequate development of pharmacogenomics. There are also very few standard protocols or test criteria for G/G services to facilitate the establishment of formal referral networks and the transfer of new technology into clinical use. HKGI will seek to improve this situation by providing standardised high-quality genomic testing to many more patients.

7. Workforce Situation

The provision of G/G 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. At present, there are only a handful of 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 number of pathologists trained in Genetic and Genomic Pathology is also inadequate though the Hong Kong Academy of Medicine has listed the first batch of fellows in this subspecialty since November 2020. 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. HKGI will collaborate with local universities and professional bodies to develop these professions, including the enhancement of training and establishment of clear career paths.

8. Genomic Literacy

The genetic and genomic literacy of healthcare professionals will need to be enhanced to facilitate the application of genomic medicine in different clinical care settings. Owing to inadequate genetic literacy among healthcare professionals, many of them may not be able to manage common G/G cases, recognise conditions with a genetic basis, know when to offer G/G tests or refer patients for specialised assessment and tests, thus contributing to delays in diagnosis and optimal treatment. Lack of genomic literacy amongst members of the public also hinders the application of genomic medicine to clinical practices and wider use of genetic tests to help the diagnosis and treatment of diseases. HKGI will engage targeted stakeholders to enhance the genomic literacy of healthcare professionals and the general public.

Strategic Framework

From the analysis of environment and key issues to be addressed, four main strategic foci pertaining to the integration of genomic medicine in clinical care, advancing research in genomic science, nurturing talents, and enhancing genomic literacy were crystallised along with an array of strategies which map out the corporate priorities for HKGI to work towards achieving its mission and vision. The four strategic foci are as follows:

Integrate Genomic Medicine into Clinical Care

Driving the incorporation of genomic medicine into the mainstream of medical development in Hong Kong by improving genomic diagnosis, personalised treatment as well as personalised prediction and prevention of disease risks. This is done by showcasing the clinical usefulness of WGS in focused disease areas and the functional/economic benefits of embedding genomics into routine clinical care.

Advance Research in Genomic Science

Facilitating genomic science and discoveries by establishing a flexible platform with rich database for new genomic technologies and multi-omics studies, as well as disease-focused research networks through local and international collaborations. Core to this will be setting up the necessary infrastructure including facilitating the development of a local biobank network and promoting local and international collaborations in genomic research.

Nurture Talents in Genomic Medicine

Developing skilled and competent professionals to deliver genomic medicine through collaborations with local universities, professional bodies and healthcare institutions to enhance training and development of the related professions, including organising continuing professional development programmes for clinicians, genetic counsellors, bioinformaticians and medical laboratory technologists, etc.

Enhance Public Genomic Literacy and Engagement

Promoting genomic literacy in Hong Kong, particularly amongst healthcare workers and students, by engaging and collaborating with relevant government departments, schools, universities and non-government organisations to initiate public education programmes in genetics and genomics.

Set out in the ensuing four chapters are the strategic goals, directions and strategies for achieving the four strategic foci. Key actions of the strategies are also highlighted as concrete examples for implementation.

The strategic goals delineate what HKGI wants to achieve; the strategic directions outline the broad directions HKGI will pursue for achieving the goals; and the strategies map out what HKGI needs to do in order to meet the goals. Presented below is the overall framework which illustrates how the strategies relate to HKGI's vision and mission.

Integrate Genomic Medicine into Clinical Care
融合基因組醫學與臨床護理

Strategic Goals 策略目標	Strategic Directions & Strategies 策略方向及具體策略	
Improve genomic diagnosis and personalised treatment **優化基因組診斷和個人化治療**	**Provide standardised high-quality genomic testing** ▸ Enhance WGS capability and capacity ▸ Enhance bioinformatics capability and capacity ▸ Obtain accreditation for HKGI laboratory and WGS pipeline ▸ Enhance provision of genetic counselling service, including tele-counselling	**提供標準化及優質的基因組測序服務** ▸ 提升全基因組測序的能力及效能 ▸ 提升生物信息學的能力及效能 ▸ 為基因組中心的實驗室及全基因組測序流程取得認證 ▸ 加強包括遙距諮詢在內的遺傳輔導服務
	Facilitate provision of more personalised treatment ▸ Promote effective use of genetic diagnosis to enable personalised treatment ▸ Promote effective use of pharmacogenomic profiling for targeted treatment	**促進個人化治療** ▸ 提倡善用基因組診斷，以實踐個人化治療 ▸ 推動善用藥理基因組分析於針對性的治療
Improve personalised prediction and prevention of disease risk **優化個人化疾病預測和預防**	**Establish a clinical genomic database of the local population** ▸ Expand HKGP patient cohorts by opening up new recruitment channels ▸ Enhance guidelines and standardised protocols on data privacy and security	**建立本地人口的臨床基因組數據庫** ▸ 開闢新的招募渠道，擴大香港基因組計劃的參與群組 ▸ 優化私隱及數據安全指引和標準規程
	Improve risk prediction for diseases prevalent in Hong Kong ▸ Develop polygenic risk scores for common diseases in local population ▸ Refine risk prediction by integrating genotyping, deep phenotyping, health-related data and medical records	**優化香港流行疾病的風險預測** ▸ 制訂本地人口常見疾病的多基因風險評分 ▸ 整合基因型和深度表型資料、健康相關數據及醫療紀錄，以優化疾病風險預測

Strategic Goals 策略目標	Strategic Directions & Strategies 策略方向及具體策略	
Establish infrastructure for implementing genomic medicine **建立發展基因組醫學的基礎設施**	**Facilitate the development of a local biobank network for genomic research** ▸ Establish the HKGI biobank to enable data sharing for research ▸ Enhance HKGI's guidelines and standardised protocols on informed consent, collection, storage and responsible sharing of genomic data	**促進建立本地生物樣本庫網絡，以發展基因組研究** ▸ 建立基因組中心的生物樣本庫，以共享數據作研究之用 ▸ 完善基因組中心就知情同意及基因組數據的收集、存儲及適切分享所制訂的指引和標準規程
	Facilitate clinical implementation of genomic medicine ▸ Evaluate clinical usefulness of WGS in focused disease areas to embed genomic medicine and to promote evidence-based research ▸ Evaluate health economics and outcomes of WGS in focused disease areas to embed genomic medicine	**推動基因組醫學的臨床應用** ▸ 評估全基因組測序在重點疾病領域的臨床成效，以促進基因組醫學的應用及循證研究 ▸ 評估醫療經濟學及全基因組測序結果在重點疾病領域的效益，藉此推動基因組醫學的應用

Strategic Intent 策略遠景

Genomic medicine is made available to all for better health and well-being
普及基因組醫學，共享健康福樂

Advance Research in Genomic Science
促進基因組科學研究

Strategic Goals 策略目標	Strategic Directions & Strategies 策略方向及具體策略	
Integrate application of multi-omics in personalised medicine **融合多組學研究成果與個人化治療**	**Establish database and platform for new genomics technology and multi-omics studies** ▸ Develop functional assays to characterise, annotate and interpret genes/variants ▸ Develop new genomic technologies for clinical implementation ▸ Engage industrial partners to translate research findings into clinical use	**建立數據庫及平台，促進新基因組學技術及多組學研究** ▸ 開發功能性研究，以歸納、註釋及詮釋各種基因變異模式 ▸ 開發新基因組技術供臨床應用 ▸ 與業界夥伴合作，轉化研究成果至臨床應用
Establish disease focused research networks **建立以防治疾病為主的研究網絡**	**Enhance local and international collaborations in genomic research** ▸ Identify specific disease themes and partner with academic institutes for relevant research studies ▸ Share and publish the project implementation experience of HKGP	**加強本地和國際在基因組研究領域的合作** ▸ 鑑辨特定疾病主題，與學術機構合作進行相關的基因組學研究 ▸ 發表香港基因組計劃的實踐經驗，分享研究成果

Strategic Intent 策略遠景

Hong Kong becomes a leading hub of biomedical science
推動香港成為領先的生物醫學中心

Nurture Talents in Genomic Medicine
培育基因組醫學人才

Strategic Goals 策略目標	Strategic Directions & Strategies 策略方向及具體策略	
Enhance genetic and genomic knowledge and professional development **強化遺傳學和基因組學的知識及專業發展**	**Engage with professional bodies to support continuing professional development programmes** ▸ Support continuing professional development programmes in genetics and genomics for clinicians, nurses, and allied health professionals, including genetic counsellors and bioinformaticians ▸ Partner with professional societies to develop genetic counselling guidelines and standards	**與專業團體合作，支援持續專業發展計劃** ▸ 支持醫生、護士、專職醫護人員包括遺傳輔導員及生物信息學家等的持續專業發展計劃 ▸ 與專業團體合作，制訂遺傳輔導指引及標準
	Incorporate experiential learning into continuing education programmes ▸ Engage staff of Partnering Centres in multidisciplinary team meetings ▸ Establish “genomics champion” in different specialties to contribute and influence genomic education and practice	**把經驗學習納入持續進修計劃之內** ▸ 鼓勵夥伴中心的人員參與跨專業團隊會議 ▸ 於不同專科培育「基因組學傑出人員/團隊」，深化基因組教學和實踐經驗，並推而廣之

Nurture Talents in Genomic Medicine
培育基因組醫學人才

Strategic Goals 策略目標	Strategic Directions & Strategies 策略方向及具體策略	
Improve genetic and genomic knowledge of tertiary students **增進大專學生的遺傳學和基因組學知識**	**Engage with local universities to promote genomic medicine** ▸ Collaborate and support local universities in developing and promoting courses in genomics, bioinformatics, biomedical science, and genetic counselling ▸ Collaborate and support local universities in promoting genomic medicine through events such as information week and career fair ▸ Develop enrichment and internship programmes for undergraduate students in genomic medicine related studies ▸ Support postgraduate programmes in genomics, bioinformatics and biomedical science	**與本地大學合作推廣基因組醫學** ▸ 支持本地大學開辦和推廣基因組學、生物信息學、生物醫學及遺傳輔導等課程 ▸ 與本地大學合辦資訊週及職涯規劃講座等活動，宣傳基因組醫學 ▸ 為修讀基因組醫學相關課程的大學本科生，開辦進修課程和實習計劃 ▸ 支持基因組學、生物信息學及生物醫學的研究生課程

Strategic Intent 策略遠景

Skilled professionals to develop and deliver genomic medicine
培育專業人才，發展和提供基因組醫學服務

Enhance Public Genomic Literacy and Engagement
加強公眾對基因組學的認識和參與

Strategic Goals 策略目標	Strategic Directions & Strategies 策略方向及具體策略	
Improve public awareness and knowledge of genomics **提高公眾對基因組學的關注和認識**	**Engage general public and targeted stakeholders to enhance public understanding of genomic medicine** ▸ Develop authoritative and user-friendly information and publications on genomic medicine for the general public ▸ Identify targeted stakeholders, including patient groups and professional bodies, and develop strategic engagement plans to promote awareness of genomic medicine and its benefits	**與相關持份者合作，提高公眾對基因組醫學的認識** ▸ 為公眾提供可靠易明的基因組醫學資訊和刊物 ▸ 選定相關持份者群組，包括病人組織和專業團體，制訂策略主動接觸他們，以加深他們對基因組醫學及其效益的認識

Strategic Intent 策略遠景

Increased public genomic literacy and engagement
加深公眾對基因組學的認識，積極參與

Integrate Genomic Medicine into Clinical Care

1. Strategic Goals and Strategic Directions

To pursue the strategic focus of integrating genomic medicine into clinical care with a view to realising its vision of making genomic medicine available to all for better health and well-being, HKGI has set three strategic goals:

> **(a) Improve genomic diagnosis and personalised treatment;**
>
> **(b) Improve personalised prediction and prevention of disease risk; and**
>
> **(c) Establish infrastructure for implementing genomic medicine.**

The strategic directions for improving genomic diagnosis and personalised treatment are to provide standardised high-quality genomic testing and to facilitate the provision of more personalised treatment, while those for improving personalised prediction and prevention of disease risk are to establish a genomic and clinical database of the local population and to improve risk prediction for diseases prevalent in Hong Kong. Complementary to these, HKGI will strive to achieve the strategic goal of establishing the necessary infrastructure for implementing genomic medicine through the strategic directions of facilitating the development of a local biobank network for genomic research and facilitating clinical implementation of genomic medicine.

Through the strategies formulated under these strategic directions, HKGI will look to address the following key issues and challenges in the development of genomic medicine in Hong Kong:

- Service gaps in the provision of genetic and genomic services, in particular, the need for offering more standardised and personalised diagnosis and treatment to bring better clinical outcomes and benefits to patients;
- Lack of standard protocols or test criteria for genetic and genomic services;

- Inequitable access to genetic and genomic testing;
- Inadequate development of pharmacogenomics to support precision medicine and targeted therapeutics; and
- The need to keep up with international standards in genetic and genomic testing and its clinical applications.

In implementing the strategies for integrating genomic medicine into clinical care, HKGI seeks to collaborate closely with its partners to use the accumulated genomic and clinical data of the local population to generate robust functional impacts on the health and well-being of the people of Hong Kong by:

- Analysing the WGS and other big data to provide deep insights into disease biology and to identify characteristics associated with health;
- Identifying predisposition to diseases and disorders prevalent in Hong Kong;
- Using biomarkers and gene signatures to diagnose the presence of diseases or disorders that are associated with specific genes or gene products; and
- Using WGS data to enable the prescription of drugs best suited to the patient's genotype to increase efficacy and reduce adverse effects.

It is expected that implementation of the strategies to integrate genomic medicine into clinical care would also yield a significant direct economic presence in the economy of Hong Kong. In the upstream, the development of a clinical genomic database of the local population will be a treasure trove for our healthcare services sector which may use the available data to discover novel target genes for certain diseases prevalent in Hong Kong and to understand genomic factors affecting various phenotypes, like growth rate, blood pressure, as well as the health effects of smoking and alcohol consumption, etc. As the scale of genomic data grows, services further down the stream will be supported, such as helping industries or start-up companies to:

- Identify targeted groups of patients for clinical trials;
- Use the genomic and computational tools developed by HKGI to support their business;
- Reduce the sequencing cost and improve accuracy of their genomic variant calling pipelines;
- Identify off-target integrations to facilitate the production of therapeutic proteins; and
- Identify suitable patients to develop disease cell-lines for understanding or modelling a disease.

In the downstream, HKGI's strategies to develop genomic medicine in Hong Kong will directly impact on the cost of our healthcare system in the following ways:

- Help public hospitals reduce costs by identifying the variants that cause diseases for the HKGP participants;
- Help public health researchers better understand the mutation profile and common hereditary diseases in Hong Kong, which can lead to significant reduction in healthcare costs; and
- Facilitate the development of some gene panels for screening patients, e.g., Asian-specific disease panel, pharmacogenomic panel for drug dosage, and panel for screening carriers of disease variants.

Recent studies commissioned by the American Society of Human Genetics and the UK Office for Life Sciences also indicate that the development of genomic medicine would impact favourably on the economy through greater growth in research expenditure and investment in the field of genomics, as well as creation of direct jobs in the human genetics and genomics domain, and additional jobs for supporting those direct jobs.

Overall, there are three strategic goals along with six strategic directions to enable the integration of genomic medicine into clinical care:

(a) Improve genomic diagnosis and personalised treatment

- Provide standardised high-quality genomic testing
- Facilitate the provision of more personalised treatment

(b) Improve personalised prediction and prevention of disease risk

- Establish a clinical genomic database of the local population
- Improve risk prediction for diseases prevalent in Hong Kong

(c) Establish infrastructure for implementing genomic medicine

- Facilitate the development of a local biobank network for genomic research
- Facilitate clinical implementation of genomic medicine

2. Strategic Goal: Improve Genomic Diagnosis and Personalised Treatment

Provide Standardised High-Quality Genomic Testing

HKGI will strive to provide standardised high-quality genomic testing by enhancing its WGS capability and capacity to improve patients' accessibility to this advanced technology for genomic diagnosis and personalised clinical care through launching the main phase of the HKGP. This requires making corresponding enhancements to the capability and capacity of its bioinformatics platform to process, analyse and store the much larger amount of WGS data for clinical application and research purposes. Another important strategy in this regard is to obtain local and international accreditation of the HKGI laboratory and WGS pipeline to establish high quality standards for its genomic tests. To promote application of the WGS data collected from HKGP participants, HKGI will enhance the provision of genetic counselling services, including tele-counselling, and develop pre-test counselling video materials for WGS.

According to the Report on Strategic Development of Genomic Medicine, there is a need to centralise advanced genetic and genomic tests that fulfil the following criteria for better utilisation of limited resources in Hong Kong:

(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).

HKGI will support Food and Health Bureau in evaluating which genetic and genomic tests may fulfil the above criteria. Subject to Food and Health Bureau's policy direction, HKGI will also strive to provide some of the advanced genetic and genomic tests with its high-quality laboratory and expertise in the long run.

Facilitate Provision of More Personalised Treatment

To move towards delivering precision medicine in the healthcare system of Hong Kong, HKGI will make strenuous efforts to facilitate the provision of more personalised treatment to patients. This will be done through the strategies of promoting the effective use of genetic diagnosis to enable personalised treatment and starting to foster the effective use of pharmacogenomic profiling for targeted treatment.

Genetic or genomic tests not only can determine potential risk for developing a disease, but also they can be highly informative in guiding the diagnosis of a present disease or disorder. With rapid advancements in genomic medicine, genetic and genomic tests for disease diagnosis are now being deployed across a broad range of rare and more common diseases and disorders. Clinicians of Partnering Centres will be encouraged to use the WGS report findings of HKGP participants to provide personalised clinical care to patients.

Medications are a cornerstone of the therapeutic armamentarium for most clinicians. The goal of pharmacotherapy is to cure or control a specific condition or disease without causing adverse effects. In recent years, the discipline of “pharmacogenomics” has grown to be able to deploy genetic and genomic knowledge and tools to help clinicians select the “right drug and the right dose” for a patient based on their genome. It is hoped that implementation of the strategies would help develop pharmacogenomics into an important area of research and clinical practice in Hong Kong, that addresses the genetically determined variation in how individuals respond to specific drugs in terms of differences in dose requirement, efficacy, and the risk of adverse drug reactions.

3. Strategic Goal: Improve Personalised Prediction and Prevention of Disease Risk

Establish a Clinical Genomic Database of the Local Population

To provide the necessary data for researchers to conduct genomic research focusing on the population of Hong Kong to bring benefits to patients of our community, HKGI will move towards the strategic direction of establishing a clinical genomic database of the local population in the next three years. HKGI has planned to proceed with this by implementing the strategy of expanding the patient cohorts through opening up new recruitment channels for the HKGP during the main phase of the project while adopting the strategy of developing guidelines and standardised protocols on data privacy and security to enhance public confidence in the project. Meanwhile, HKGI will make use of the database to showcase the clinical benefits and test out the clinical protocols of precision medicine for clinicians to improve personalised prediction and prevention of disease risk amongst their patients.

With advances in WGS, clinicians are facing a new challenge of interpreting the discoveries of Variant of Uncertain Significance (VUS). VUS refers to a variation in the genetic sequence for which its association with disease risk is unclear. While there is still a long way to go before clinicians and scientists can fully understand these variations, the establishment of a large clinical genomic database of the local population will help them make sense of some VUSs based on the available clinical information and phenotypes of patients and their families. Without such a local database, they often need to follow international guidelines in categorising whether a VUS is likely to be benign or pathogenic. However, the appropriateness of applying such guidelines to patients is sometimes questionable, judging from the significant symptoms or family history of the patients. There is scientific evidence that some VUSs are actually benign changes and others are actually Chinese specific disease-causing mutations. The HKGP database will definitely help improve diagnostic strategies and clinical management of such cases, which is precisely the essence of personalised patient care.

Improve Risk Prediction for Diseases Prevalent in Hong Kong

For the strategic direction of improving risk prediction for diseases prevalent in Hong Kong, HKGI will implement two exploratory strategies to achieve the objective. This entails the development of a set of polygenic risk scores for common diseases in the local population and integration of genotyping, deep phenotyping, health-related data and medical records to refine risk prediction.

Our ability to predict and prevent disease risk for individuals has improved rapidly in the last decade with the implementation of many large-scale sequencing projects for many different populations around the world to enable ongoing assembly of robust, evidence-based resources for the identification and classification of genomic variant pathogenicity. With expansion of the library of gene-disease associations, HKGI will be able to start the journey to develop polygenic risk scores for common diseases of the local population and use them to provide personalised prediction and prevention of disease risk for individuals after taking into consideration their genotypes, phenotypes, health-related data and medical records.

4. Strategic Goal: Establish Infrastructure for Implementing Genomic Medicine

Facilitate Development of a Local Biobank Network for Genomic Research

One of the most important infrastructures for implementing genomic medicine in Hong Kong is the establishment of a biobank network for promoting genomic research amongst the local healthcare providers and research institutions. HKGI will advance towards the strategic direction of facilitating the development of such a biobank network for the promotion of genomic research by implementing the strategies of setting up its own biobank to share genomic and other omics data with local researchers via the network, and enhancing guidelines and standardised protocols for obtaining informed consent from HKGP participants to collect, store and share genomic data in a secure and responsible way. With experience gained, HKGI will support Food and Health Bureau in pioneering collaboration with universities to establish the governance and protocol of a local biobank network for genomic medicine.

Facilitate Clinical Implementation of Genomic Medicine

In addition to the development of a biobank network, HKGI will proceed towards the strategic direction of facilitating clinical implementation of genomic medicine in Hong Kong through two additional strategies to enhance the infrastructure for promoting the use of genomic data for clinical care in the coming three years. The first strategy is to evaluate the clinical usefulness of WGS in focused disease areas to encourage clinicians of the related clinical specialties to embed genomic medicine in their clinical practices. Also, HKGI will continue to promote evidence-based research and help translate its findings into clinical use. Complementary to this, HKGI will implement a second strategy to conduct preliminary evaluations on health economics and outcomes of WGS in focused disease areas to showcase the economic and functional impacts of genetics and genomics to clinicians so as to convince them to embed genomic medicine in clinical care.

Advance Research in Genomic Science

1. Strategic Goals and Strategic Directions

To pursue the strategic focus of advancing research in genomic science with a view to supporting Hong Kong to become a leading research hub of biomedical science, HKGI has formulated two strategic goals: (a) integrate application of multi-omics in personalised medicine; and (b) establish disease focused research networks.

The strategic direction for integrating application of multi-omics in personalised medicine is to establish a flexible platform with rich database for new genomics technology and multi-omics studies. In addition, HKGI will seek to achieve the strategic goal of establishing disease focused research networks to promote advanced genomic research through the strategic direction of enhancing local and international collaborations.

Through the strategies developed under these strategic directions, HKGI will strive to address the following key issues and challenges:

- Lack of a clinical genomic database of the local population for researchers to conduct genomic research;
- Keeping pace with the fast global development of new genomics technology and multi-omics studies;
- Accelerating clinical application of multi-omics breakthroughs; and
- Underdevelopment of disease-focused research networks in Hong Kong.

In implementing the strategies for advancing research in genomic science, HKGI will collaborate closely with its partners in both the public and private sectors to bring about benefits of the advancement and application of new genomic technologies to individuals and the society, such as:

- Establishment of a database and analysis platform for genomic and clinical data of the local population to provide important resources for biomedical research, big data analysis and the development of information technology;
- Establishment of local and international knowledge-sharing platforms using a standardised approach for recording, sharing and interrogating fully integrated clinical and genomic databases to provide clinically useful research reports;
- Acceleration of personalised clinical care with application of multi-omics research breakthroughs to provide precise and cost-effective diagnosis, treatment and prevention of diseases for the health and well-being of patients;
- Realisation of reduced healthcare costs for individuals and the healthcare system by making genetic diagnosis of diseases a more accurate and efficient process and by the development of effective and targeted treatment methods;
- Propulsion of economic development not only by reducing productivity losses and decreasing costs of disease treatment, but also by applying omics-technologies to create new biomedical industries and boost business growth of existing enterprises, e.g., developing new medical diagnostic tests and treatment, manufacturing pharmaceuticals and other healthcare products, etc.; and
- Creation of synergies between genomic science and sciences of related disciplines, e.g., systems biology, data science, statistical science, computational technologies, etc. to bring about more revolutionary scientific discoveries.

Overall, there are two strategic goals along with two strategic directions to pursue the strategic focus of advancing research in genomic science:

(a) Integrate application of multi-omics in personalised medicine

- Establish database and platform for new genomics technology and multi-omics studies

(b) Establish disease focused research networks

- Enhance local and international collaborations in genomic research

2. Strategic Goal: Integrate Application of Multi-Omics in Personalised Medicine

Establish Database and Platform for New Genomics Technology and Multi-Omics Studies

Because its products are so useful and beneficial to our healthcare system and society, the process of advancing genomic science is closely intertwined with its applications in clinical care. Conversely, seeking to integrate application of multi-omics into clinical care will contribute to the advancement of scientific knowledge and technological breakthrough in genomics.

HKGI will look to achieve its strategic goal of integrating application of multi-omics in personalised medicine by moving towards the strategic direction of establishing a platform for new genomics technology and multi-omics studies. This will be done through the strategies of upgrading analyses and interpretation of WGS data by the development of functional assays to characterise, annotate and interpret genes/variants, and developing new genomic technologies, such as long-read sequencing, single-cell sequencing technologies as well as transcriptomics and epigenomics analyses, for clinical implementation. HKGI will also search for opportunities to engage industrial partners to translate research findings into clinical use.

HKGI will also strive to be the enabler and facilitator for researchers, providing a rich database and flexible platform for their exploration and discovery. Specifically, HKGI will provide support to researchers in terms of curating the most appropriate dataset for individual research requests, ensuring the reliability and security of the infrastructure, and facilitating innovative research algorithm to be run in the platform.

3. Strategic Goal: Establish Disease Focused Research Networks

Enhance Local and International Collaborations in Genomic Research

The strategic direction for achieving the strategic goal of establishing disease focused research networks is to enhance local and international collaborations in genomic research. HKGI will advance towards this strategic direction by adopting two main strategies in the next three years. Specifically, it will identify specific disease themes for partnering with local and international academic institutes to conduct research studies which are useful in the analysis of diseases. Moreover, HKGI will consolidate the experience of implementing the HKGP and publish it for sharing with local and international partners.

The speed with which personalised medicine becomes a reality for most of the population will be strongly influenced by the sharing of clinical information — in this case, an individual's genomic sequence together with their medical record — as part of large population datasets that are accessible to clinicians and translational researchers. By aggregating and analysing large datasets, it will be possible for researchers to uncover patterns and relationships that would not otherwise be evident. The enormous value of data sharing in the acceleration of progress in genomic medicine is now well recognised. HKGI will make strenuous efforts to establish disease focused research networks to advance research in genomic medicine through enhanced local and international collaborations.

Nurture Talents in Genomic Medicine

1. Strategic Goals and Strategic Directions

To nurture talents in genomic medicine with a view to realising the vision of having skilled professionals to develop and deliver genomic medicine in Hong Kong, HKGI will pursue the strategic goals of (a) enhancing genetic and genomic knowledge and professional development, and (b) improving genetic and genomic knowledge amongst tertiary students. To enhance genetic and genomic knowledge and professional development, a collaborative approach will be adopted along two strategic directions, namely, engaging with professional bodies to support continuing professional development programmes and incorporating experiential learning into continuing education programmes. To improve genetic and genomic knowledge amongst tertiary students, HKGI will move along a similar strategic direction of engaging with local universities to promote genomic medicine.

Through the strategies formulated under these directions, HKGI will seek to address the following key issues and challenges:

- Lack of a sufficient pool of genetic and genomic professionals, including medical geneticists, genetic counsellors and bioinformaticians;
- The need to strengthen training and development of the genetic and genomic professionals;
- Lack of genetic and genomic knowledge amongst other healthcare professionals; and
- The need to enhance literacy and interest in genomics amongst tertiary students.

Overall, there are two strategic goals along with three strategic directions to nurture talents in genomic medicine:

(a) Enhance genetic and genomic knowledge and professional development

- Engage with professional bodies to support continuing professional development programmes
- Incorporate experiential learning into continuing education programmes

(b) Improve genetic and genomic knowledge amongst tertiary students

- Engage with local universities to promote genomic medicine

2. Strategic Goal: Enhance Genetic and Genomic Knowledge and Professional Development

Engage with Professional Bodies to Support Continuing Professional Development Programmes

The training of healthcare professionals is of paramount importance to the transformation of Hong Kong's healthcare system by bringing genomic medicine into the mainstream. In this regard, HKGI will collaborate with the universities, HA, DH, the Hong Kong Academy of Medicine and relevant professional bodies to develop new training and education programmes in genetics and genomics to help healthcare professionals absorb the rapidly evolving pool of genetic and genomic knowledge. In the next three years, HKGI will focus on supporting the professional bodies to organise continuing professional development programmes in genetics and genomics for clinicians, nurses and allied health professionals. It is hoped that these continuing education programmes can enhance healthcare professionals' genetic and genomic knowledge as well as nurture a positive and supportive culture in the clinical setting that facilitates the application of genomics and genomic services into clinical practice.

Besides the training and development of clinical professionals, HKGI will also provide support to its partners in organising continuing professional training and development programmes for professionals of other related disciplines, such as genetic counsellors and bioinformaticians, who play an essential role in the clinical application of genetic and genomic services. To nurture talents and align standards in genetic counselling, HKGI will partner with the professional societies in this field to promote development of the profession and to set genetic counselling guidelines and standards in Hong Kong.

Incorporate Experiential Learning into Continuing Education Programmes

To achieve the strategic goal of enhancing genetic and genomic knowledge and professional development, HKGI will also move along the strategic direction of incorporating experiential learning into continuing education programmes of the related professional disciplines. Experiential learning is the process of learning through experience, and adopts the approach of "learning through reflection on doing". In this regard, HKGI will implement two main strategies. One is to engage clinicians of the Partnering Centres in discussing the WGS report findings and their clinical application in multidisciplinary team meetings. Through these exchanges of views between professionals of different disciplines, the genetic and genomic knowledge of healthcare professionals will be enhanced and the wider use of genomic approaches in healthcare promoted.

Another strategy to be implemented to promote experiential learning in genomic medicine is to establish "genomics champions" in different clinical specialties, who will be engaged to contribute and influence genomics education and its integration into clinical practice amongst colleagues of their respective specialties. HKGI will provide training and support to these genomics champions to raise awareness, disseminate information and support the mainstreaming of genomics in their specialty. They will be involved in performing the role of:

- Identifying gaps where genomics-related training packages need to be provided, either at a general level or specific to a clinical specialty;
- Raising awareness of genomics amongst their colleagues in the clinical setting and build interdepartmental relationships;
- Providing a point of contact for anyone with a genomics query within their specialty;
- Sharing genomics information with their colleagues to develop an understanding of what genomics is and what it means for them in relation to their patients; and
- Contributing to the development of educational materials for enhancing genomic literacy amongst healthcare professionals.

3. Strategic Goal: Improve Genetic and Genomic Knowledge amongst Tertiary Students

Engage with Local Universities to Promote Genomic Medicine

Apart from the non-genomic healthcare professionals, undergraduate and postgraduate students are by far the most important target audiences of HKGI's genomic-focused educational activities. To achieve the strategic goal of improving genetic and genomic knowledge amongst tertiary students, HKGI will proceed towards the strategic direction of engaging with local universities to promote genomic medicine by progressively implementing the following four main strategies.

First, HKGI will collaborate and support local universities to develop and promote courses in genomics, biomedical science, bioinformatics, and genetic counselling to nurture and train talents necessary for the development of genomic medicine and its integration into clinical practice in Hong Kong. Second, HKGI will join hands with local universities to organise promotional and educational events such as Information Week and Career Fair to attract more tertiary students to study genomics-related subjects or pursue a career in genomics-related professions. Third, HKGI will develop enrichment and internship programmes for undergraduate students in genomics-related studies to improve their genetic and genomic knowledge, and to help nurture talents in genomic medicine. Fourth, HKGI will offer support to local universities in the organisation of postgraduate programmes in genomics, bioinformatics and biomedical science to build up the pool of talents for furthering the development of genomic medicine in Hong Kong.

Enhance Public Genomic Literacy and Engagement

1. Genomic Literacy

The fourth strategic focus of HKGI's Strategic Plan 2022-2025 is to enhance public genomic literacy and engagement. Genomic literacy is commonly defined as the understanding of what a genome is, how genomic science works, and its affordances and limitations, applications, and impacts on society. In general, there are three types of knowledge building: awareness, how-to, and principles-based knowledge. Awareness knowledge refers to having general knowledge or perception of genomics and genomic services. How-to knowledge refers to practical knowledge about the application of genomics and genomic services into clinical practice. Principles-based knowledge pertains to an understanding of the underlying theoretical principles of genomics. While the latter two types of knowledge building are more related to HKGI's third strategic focus of nurturing talents and strengthening genomic literacy amongst healthcare professionals and university students, awareness knowledge building to improve genomic literacy amongst members of the public and youths has become the fourth strategic focus of HKGI's work in the coming three years.

While genomic discoveries will increasingly advance the science of medicine, limited genomic literacy may adversely impact the public's understanding and use of the power of genetics and genomics in healthcare and public health. Genomic literacy and awareness are keys to the effective management of genetic diseases which includes diagnosis, prevention of complications and therapy. Genomic literacy will help the public understand personal health issues involving genomic medicine, make better personal and family health decisions, understand media reports about advancement of genomic research, or participate in public policy discussions relating to the role of genomics in society.

2. Strategic Goal and Strategic Direction

To realise the vision of increased public genomic literacy and engagement in Hong Kong, HKGI will pursue the strategic goal of improving public awareness and knowledge of genomics by engaging the general public and targeted stakeholders to enhance public understanding of genomic medicine.

Through the strategies formulated under this strategic direction, HKGI will look to address the following key issues and challenges:

- Lack of genomic literacy amongst members of the public which hinders the application of genomic medicine to clinical practices and wider use of genetic tests to help the diagnosis and treatment of diseases; and
- The need for promoting genomic literacy amongst youths to enhance long-term genomic health literacy in our society.

3. Strategic Goal: Improve Public Awareness and Knowledge of Genomics

Engage General Public and Targeted Stakeholders to Enhance Public Understanding of Genomic Medicine

Genomic literacy will be achieved only through active engagement between genomics experts and the varied constituencies that comprise the public. To create such engagement opportunities, HKGI will develop strategies for engaging various sectors of the public to enhance their understanding of the explosive growth of genomic approaches to healthcare in recent years. Again, a collaborative approach will be adopted with extensive involvement of targeted stakeholders, including collaborating with patient groups and professional bodies to promote awareness of genomic medicine and its benefits, participating in popular

science events to raise awareness on the development of genomic medicine, and developing authoritative and user-friendly information and publications on genomic medicine to enhance public genomic literacy.

Genomic science has identified grand medical challenges such as making genomics-based diagnostic tests routine, achieving a comprehensive characterisation of cancer genomes, and incorporating genomic data into personalised medical treatment. These significant achievements contrast sharply with the public's level of understanding of basic biology (including genetics) and mathematics (including probability theory, statistics and risk). However, these concepts are important to allow the public to integrate genomics into their personal healthcare.

To inculcate the new scientific concepts evolved from the genomic revolution amongst the youths is therefore crucial to increasing the long-term genomic literacy of our society. There is therefore a need to consider the genomic literacy challenge from both the youth and adult perspectives to highlight the gaps, intersections and areas of opportunity for strengthening genomic literacy in an integrated fashion. In this regard, HKGI will develop strategic public engagement plans and educational materials in collaboration with targeted stakeholders, including youth organisations, to promote awareness of genomic medicine and its benefits to patients and the society.

Key Enablers

1. Information Technology Support

Successful implementation of the strategies set out in this Strategic Plan will require key enablers, in particular Information Technology (IT), to support the generation, storage and dissemination of genomic data, while at the same time providing appropriate access and the necessary computing power for its analysis and interpretation, as well as application to clinical practices. To promote the clinical application of genomic medicine, HKGI's ability to share appropriate and relevant patient information electronically with due diligence to data privacy and security, and in line with patient consent, will become increasingly important.

Moreover, as increasing amounts of genomic information are generated by the HKGP, there will be growing demands on the IT infrastructure for a unified platform which can securely process huge amounts of genomic data for promoting the development of genomic medicine. IT system infrastructure will also be crucial in enabling the workflow, communication and coordination between HKGI and its various partners.

To create favourable conditions for the advancement of genomic medicine, HKGI will seek to play an active role in the development and application of state-of-the-art information technologies in Hong Kong, particularly those related to bioinformatics. It is hoped that the application of advanced IT techniques in the development of a bioinformatics platform for the HKGP will in turn promote the further development of the IT infrastructure in Hong Kong.

2. Progress of Genomic Medicine Research in Local & International Scenes

The key to the development of genomic medicine is the accumulation of knowledge in the field and its application to clinical practices. Therefore, the progress of genomic medicine research in both local and international settings will be a key enabler to the mainstreaming of genomic medicine in the healthcare system of Hong Kong. In order for us to be at the forefront in the development of genomic medicine, HKGI must establish an active and effective local and international exchange and collaboration network to learn from various parties to promote the advancement of genomic medicine in Hong Kong. To this end, HKGI will make strenuous efforts to facilitate the establishment of a local biobank in Hong Kong to promote genomic data sharing and help connecting local genomic researchers with overseas research institutions to keep them abreast of latest developments in genomic medicine and to foster international collaborations in genomic research. Obtaining more resources for HKGI and other local research and healthcare institutions, including talents and funding, to conduct genomic and multi-omics research will also be a crucial contributing factor to its success.

Implementation and Monitoring

1. Implementation

This Strategic Plan serves as the overarching document for guiding all aspects of HKGI's development and planning in the coming three years, including services, facilities, manpower, IT, business support and financial resources, etc. In particular, it provides the basis on which HKGI's executives develop the annual plan programme initiatives through a longer-term planning approach.

Strategies and key actions of the Strategic Plan will be implemented through the annual planning process steered by the Chief Executive Officer to ensure the Annual Plans align with the Strategic Plan. In this regard, the three Annual Plans covering the period 2022-23 to 2024-25 will be the specific action plans for implementing the Strategic Plan.

2. Monitoring

Monitoring of the implementation of the Strategic Plan will be led and overseen by the HKGI Board of Directors and its six functional committees. Specific action plans for implementing strategies of the Strategic Plan will be formulated for each of the three years 2022-23, 2023-24 and 2024-25 for consideration and approval of the Board towards the end of the previous fiscal year. A progress report on the implementation of these annual plan initiatives will be submitted to the Board after each fiscal year for Board members to monitor progress in the implementation of the strategies laid down in the Strategic Plan. Annual plan initiatives under the jurisdiction of individual Board committees and their implementation progress will be presented to the respective committees for consideration before submission to the Board. Moreover, the strategies and key actions set out in the Strategic Plan will be monitored by the Food and Health Bureau.

Abbreviations

ATCG	The colour codes representing the four different bases of DNA nucleotides (A - Adenine, T - Thymine, C - Cytosine and G - Guanine)
CGS	Clinical Genetic Services
CHT	Congenital Hypothyroidism
CUHK	The Chinese University of Hong Kong
DH	Department of Health
DNA	Deoxyribonucleic acid
G/G	Genetic and Genomic
G6PD	Glucose-6-Phosphate Dehydrogenase
HA	Hospital Authority
HKCH	Hong Kong Children's Hospital
HKGI	Hong Kong Genome Institute
HKGP	Hong Kong Genome Project
HKSAR	Hong Kong Special Administrative Region
HKU	The University of Hong Kong
HKUST	The Hong Kong University of Science and Technology
IEM	Inborn Errors of Metabolism
PWH	Prince of Wales Hospital
QMH	Queen Mary Hospital
Steering Committee	Steering Committee on Genomic Medicine
VUS	Variant of Uncertain Significance
WGS	Whole Genome Sequencing

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