



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要的研究網絡，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.

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.

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. 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，並定期向食物及衛生局匯報。有關的執行進度，亦會刊於基因組中心的年度報告。