



Hong Kong  
Genome Institute  
香港基因組中心



Laying the Foundation  
for Better Health with  
Genomic Medicine

邁步基因組醫學

固植根基 樂享健康

2021-2022 ANNUAL REPORT 年報

The cover design, featuring a flourishing tree, embodies the vision and core values of the Hong Kong Genome Institute (HKGI). It encapsulates HKGI's major achievements in its first year of full operations as the tagline highlights: **Laying the Foundation for Better Health with Genomic Medicine.**

HKGI's five corporate colours are prominently displayed, representing the organisation's unique core values: red signifies Passion and Dedication; yellow evinces Versatility and Energy; blue conveys Hope and Happiness; green underlines aspiration of Health and New Life; and dark green denotes Professionalism and Reliability.

The tree, with strong roots, yields fruits big and small. The DNA strand and colourful icons on it symbolise HKGI's commitment and efforts in advancing genomic medicine in Hong Kong along four strategic foci: integrating genomic medicine into clinical care, advancing research, nurturing talents, and enhancing public genomic literacy.

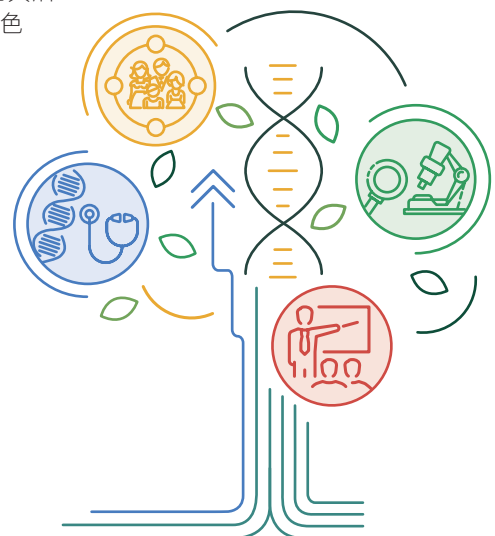
With a solid foundation, the tree will continue to grow and flourish, so as HKGI in its endeavour to avail genomic medicine to all for better health and well-being.

年報封面為一棵枝葉茂盛、碩果纍纍的大樹，由不同圖案組成，與香港基因組中心（基因組中心）的願景和核心價值相互呼應。整體設計象徵了基因組中心首年全面運作後取得的重大成就，一如年報的主題「**固植根基，樂享健康**」——為香港邁步基因組醫學奠下穩固基礎，開創健康未來。

封面的大樹以五種顏色作為主調，各有所喻，代表了基因組中心獨特的核心價值：紅色寓意熱誠與專注；黃色象徵多元與活力；藍色傳達希望與快樂；綠色強調健康與新生；深綠色則展現專業與可靠。

根深葉茂的樹上，繫滿果實，其中的雙螺旋DNA長鏈和主要圖案，凸顯了基因組中心透過四大策略重點，推動基因組醫學的決心和努力，當中包括加快融合基因組醫學與臨床護理、促進科學研究、培育人才，以及加強公眾對基因組學的認識。

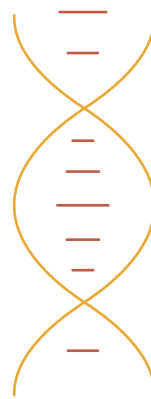
樹木擁有強壯堅固的根幹，方可茁壯成長，基因組中心的發展亦然。憑藉堅實的根基，基因組中心將繼續竭力實踐普及基因組醫學，與市民大眾共享健康福樂的願景。



# Contents

## 目錄

	<b>Corporate Profile</b>
	<b>企業概況</b>
2	About Hong Kong Genome Institute 關於香港基因組中心
6	The Board 董事局
24	Management Team 管理團隊
26	Corporate Information 公司資料
	<b>Management Reports</b>
	<b>管理層報告</b>
27	Chairperson's Statement 主席報告
34	Chief Executive Officer's Report 行政總裁報告
46	<b>Milestones of the Year</b> <b>年度回顧</b>
	<b>Laying the Foundation for Better Health</b>
	<b>固植根基 樂享健康</b>
60	Integrate Genomic Medicine into Clinical Care 融合基因組醫學與臨床護理
69	Advance Research in Genomic Science 促進基因組科學研究
75	Nurture Talents in Genomic Medicine 培育基因組醫學人才
81	Enhance Public Genomic Literacy and Engagement 加強公眾對基因組學的認識和參與
89	Operate with Excellence 卓越營運
	<b>Corporate Governance</b>
	<b>企業管治</b>
95	Principles and Practices 原則與實務
98	Governance Structure 管治架構
108	<b>Financial Report</b> <b>財務報告</b>



## About Hong Kong Genome Institute 關於香港基因組中心

### Corporate Introduction

The Hong Kong Genome Institute (HKGI), established and wholly owned by the Government of the Hong Kong Special Administrative Region (HKSAR Government), commenced full operations in 2021. With the vision *“to avail genomic medicine to all for better health and well-being”* and supported by the Health Bureau, HKGI works in close collaboration with the Department of Health, Hospital Authority, medical schools of local universities and various stakeholders to accelerate the development of genomic medicine in Hong Kong along four strategic foci: integrating genomics into medicine, advancing research, nurturing talents and enhancing public genomic literacy.

As the first step towards achieving its vision, HKGI launched the Hong Kong Genome Project (HKGP) in 2021 focusing on diseases and research cohorts that would benefit from whole genome sequencing. They include undiagnosed diseases, hereditary cancers and diseases related to genomics and precision health. Being the city's first large-scale genome sequencing project, HKGP serves as a catalyst to benefit patients and their families with more precise diagnosis and personalised treatment through whole genome sequencing. It also aims to establish a genome database of the local population, testing infrastructure and talent pool to address the healthcare needs of Hong Kong in the long run.

HKGI has set up Partnering Centres at the Hong Kong Children's Hospital, Prince of Wales Hospital and Queen Mary Hospital to help recruit eligible participants for HKGP with informed consent, while also keeping other stakeholders closely engaged. Once available, the results of sequencing analysis will be fed back to respective clinical leads and patients to aid diagnoses and clinical services.

### 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(特區政府)成立及全資擁有，於2021年正式全面運作。基因組中心致力促進本港基因組醫學發展，在醫務衛生局支持下，與衛生署、醫院管理局、大學醫學院及各方持份者緊密合作，透過聚焦四大策略重點，包括加快融合基因組醫學與臨床應用、促進科學研究、培育人才及加強公眾對基因組學的認識，實現「**普及基因組醫學，共享健康福樂**」的願景。

基因組中心於2021年正式開展香港基因組計劃(基因組計劃)，主要涵蓋三個受惠於全基因組測序技術的疾病及研究群組，包括未能確診病症、與遺傳有關的癌症，以及與基因組學及精準醫學有關的個案。基因組計劃是本港首個大型基因組測序計劃，扮演著催化劑的角色，以全基因組測序讓病人及其家屬受惠於更準確診斷及個人化治療，並透過建立本地人口的基因組數據庫、測試設施及人才庫，應對香港長遠醫療需要，與大眾同創健康未來。

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，並積極與其他持份者緊密合作，透過轉介，招募合資格參加者，經他們知情同意後參與基因組計劃；而相關測序分析的結果，將回饋予有關醫護人員及病人作診斷及臨床治療之用。



## Vision, Mission and Core Values

### Vision

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

### Mission

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

### 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.

## 願景、使命及核心價值

### 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。

### 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。

### 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**。

About Hong Kong Genome Institute  
關於香港基因組中心

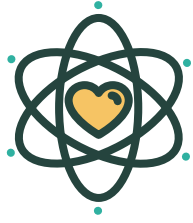


**Professionalism and Reliability**

To provide whole genome sequencing, 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.



### Versatility and Energy

To adopt a multi-disciplinary 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.



### 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.



### 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.

### 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。

### 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。

### 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。

### 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。



## The Board 董事局

Mr Philip TSAI Wing-chung, BBS, JP  
蔡永忠先生, BBS, JP

Chairperson  
主席



Mr Tsai is a former Chairman of Deloitte China and has more than 35 years of experience in planning and managing audits for Hong Kong based operations of multi-national groups, as well as local and overseas listed clients in a wide range of industries. As a Fellow of the Hong Kong Institute of Certified Public Accountants (the "HKICPA"), the Association of Chartered Certified Accountants and the Institute of Chartered Accountants of England and Wales (the "ICAEW"), Mr Tsai is actively involved in the development of the CPA profession and also contributes his efforts in various government, community and social services.

Mr Tsai is a Past President of the HKICPA, a Council Member of the ICAEW, a Member of the Hospital Authority, the Chairman of the Hospital Governing Committee of the Queen Mary Hospital and Tsan Yuk Hospital, the Chairman of the Supplementary Medical Professions Council of the Health Bureau (formerly the Food and Health Bureau), a Member of the Advisory Committee and Audit Committee of the Mandatory Provident Fund Schemes, an Advisor of The Ombudsman, Hong Kong, a Member of the Registration Committee of the Chinese Gold and Silver Exchange, a Member of the Standing Committee on Disciplined Service Salaries and Conditions of Services of the Civil Service Bureau and a Member of the Independent Commission on Remuneration for Members of the Executive Council and the Legislature, and Officials under the Political Appointment System of the HKSAR Government.

Moreover, Mr Tsai is a Member of the University Grants Committee, an Adjunct Professor of the City University of Hong Kong, a Honorary Court Member, the Chairman of the Student Residence Management Board of the Hong Kong Baptist University, the Deputy Chairman of the Alumni Committee of the Hong Kong Baptist University Foundation, a Trustee of the Staff Superannuation Scheme of The Chinese University of Hong Kong and a Member of the Institutional Advancement and Outreach Committee of the University Council of The Hong Kong University of Science and Technology.

Mr Tsai is also the Chairperson of Hong Kong Red Cross.

蔡先生為德勤前中國主席，擁有逾35年的工作經驗，為跨國集團在港業務以及各行各業本土和海外上市客戶實施審計規劃與審計管理。他亦為香港會計師公會、英國特許公認會計師公會、以及英格蘭與威爾士特許會計師公會資深會員。他積極參與註冊會計師行業的發展，為政府、社區和社會服務盡心盡力。

蔡先生亦是香港會計師公會前會長、英格蘭與威爾士特許會計師公會理事會成員、醫院管理局大會成員、瑪麗醫院／贊育醫院醫院管治委員會主席、醫務衛生局(前食物及衛生局)輔助醫療管理局主席、強制性公積金計劃諮詢委員會及審核委員會委員、申訴專員顧問、香港金銀業貿易場註冊委員會成員、公務員事務局紀律人員薪俸及服務條件常務委員會委員及香港特別行政區行政會議成員、立法會議員及政治委任制度官員薪津獨立委員會成員。

他同時擔任大學教育資助委員會成員、香港城市大學兼任教授、香港浸會大學榮譽諮議會成員、浸大學生舍堂管理委員會主席、香港浸會大學基金校友委員會副主席、香港中文大學教職員公積金計劃信託人及香港科技大學大學拓展委員會成員。

蔡先生亦為現任香港紅十字會主席。



Professor Raymond LIANG Hin-suen, SBS, JP

梁憲孫教授, SBS, JP

Deputy Chairperson  
副主席



Professor Liang is a Specialist in Haematology and Haematological Oncology. He is currently Head of Department of Medicine, Director of Comprehensive Oncology Centre and Assistant Medical Superintendent of Hong Kong Sanatorium and Hospital. He is also Emeritus Professor of The University of Hong Kong (HKU) and Honorary Professor of both HKU and The Chinese University of Hong Kong.

Professor Liang was a member of the Hospital Authority Board. Furthermore, he was the Ex-Dean of Li Ka Shing Faculty of Medicine, HKU, and the Past President of the Hong Kong Academy of Medicine.

Professor Liang was one of the founders of Hong Kong Blood Cancer Foundation and Hong Kong Marrow Match Foundation. The latter was responsible for establishing the first all Chinese Unrelated Marrow Donor Registry, serving Chinese patients in need of bone marrow transplantation in Hong Kong, Macau, Taiwan and Mainland China, as well as Chinese patients around the world.

梁教授是一位血液及血液腫瘤科專科醫生，現為養和醫院副院長、內科部主管及綜合腫瘤科中心主任、香港大學(港大)榮休教授和榮譽講座教授，以及香港中文大學榮譽講座教授。

他亦為前醫院管理局委員、港大醫學院前院長和香港醫學專科學院前主席。

梁教授為香港血癌基金和香港骨髓捐贈基金的創會成員之一，後者更成立了全球首個以華人為主的香港骨髓捐贈基金資料庫，為華裔血病患者尋找適合的無血緣骨髓。

## The Board 董事局

### Dr LO Su-vui 羅思偉醫生

Non-Official Director  
非官方董事



Dr Lo joined Hong Kong Genome Institute (HKGI) as the Chief Executive Officer in March 2021. As the CEO, Dr Lo leads HKGI in formulating its strategic direction and development plan. He also provides leadership to the team in launching the Hong Kong Genome Project, the city's first-ever large-scale whole genome sequencing initiative. With the aspiration to benefit the wider community, Dr Lo drives the team to be the change agent in fostering clinical application of genomic medicine and its long-term development in Hong Kong.

Dr Lo is a seasoned professional in public health and administrative medicine, bringing with him a wealth of knowledge in public healthcare system, service provision and relevant policies. He also has a strong blend of experience in corporate affairs, strategic planning, talent development, research and so on. Prior to his current role, Dr Lo had served in the Hospital Authority (HA) for over 20 years during which he had held various senior management positions, including Director of Strategy & Planning in the HA Head Office and Cluster Chief Executive of the New Territories East Cluster.

In addition to his extensive experience with the HA, Dr Lo also held a number of senior positions both locally and overseas. These included being the Head of Research Office in the former Food and Health Bureau (currently the Health Bureau) of the HKSAR Government, and the Director of Purchasing (Specialist Care Services) with the Cardiff Health Authority of the National Health Service in the United Kingdom.

Professionally, Dr Lo is a practitioner in Public Health & Administrative Medicine. He had served as a Part I and II examiner of the Faculty of Public Health, and Censor and examiner of the Royal Australasian College of Medical Administrators.

羅醫生於2021年3月出任香港基因組中心行政總裁，負責領導基因組中心制訂發展策略，帶領團隊發揮推動者的角色，推行本港首個大規模的基因組測序計劃（「香港基因組計劃」），以促進基因組醫學在香港的臨床應用及長遠發展，實現為社會大眾帶來裨益的願景。

羅醫生為資深公共衛生及行政醫學專家，對本地醫療體系、公共服務及相關政策認識深厚，在機構事務、發展規劃、人才培育及調查研究等範疇，均具豐富經驗。出任現職前，羅醫生於醫院管理局（醫管局）服務逾20年，曾擔任不同管理職位，其中包括醫管局總辦事處策略發展總監及新界東醫院聯網總監。

羅醫生亦先後於本地及海外擔任不同要職，包括香港特區政府前食物及衛生局（現為醫務衛生局）研究部主管及英國國民保健服務卡迪夫衛生局採購總監（專科照護服務）。

羅醫生持有公共衛生及行政醫學專業資格，並曾於澳洲皇家醫務行政學院公共衛生學院擔任考核員（甲部及乙部）、審查員及考試委員。

Dr Derrick AU Kit-sing  
區結成醫生

Non-Official Director  
非官方董事



Dr Au is Clinical Professional Consultant (Honorary) and Centre Advisor of the Centre for Bioethics at The Chinese University of Hong Kong. He received medical education at Brown University in the US and postgraduate training in geriatric medicine in Hong Kong. Dr Au has served in clinical service in geriatrics and rehabilitation for two decades before taking up various management positions in the Hospital Authority, including Director of Quality & Safety, overseeing clinical ethics, research ethics, patient risk management and technology assessment among other responsibilities.

Dr Au is also a writer and columnist with publications on bioethics, professional ethics, and history of medicine.

區醫生為香港中文大學榮譽臨床專業顧問及生命倫理學中心顧問，他亦是專科醫生、前醫療管理人及作家，筆名區聞海。區醫生畢業於美國布朗大學醫學院，回港後服務公營醫療30多年，曾任九龍醫院行政總監、醫院管理局質素及安全總監等職務。

區醫生工餘從事人文及倫理寫作，出版的書籍包括《當中醫遇上西醫－歷史與省思》(2004)、《醫院筆記：時代與人》(2016)、《如何走下去－倫理與醫療》(陳浩文、區結成編著，2018)、《生命倫理專題：人工生殖科技》(上、下冊)(區結成編，顏妙融、江萬琪著，2019，2021)，及《生命倫理的四季大廈》(2019)等。

The Board  
董事局

Mr Ray CHAN Chin-ching  
陳展程先生

Non-Official Director  
非官方董事



Mr Chan is the Chief Executive Officer and co-founder of 9GAG, a global multi-platform community for viral content and interests. Founded in 2008, 9GAG's mission is to make the world happier.

9GAG has a global audience of 200 million, including 56 million followers on Instagram, 42 million followers on Facebook, and 16 million followers on Twitter.

Mr Chan graduated from The University of Hong Kong with a degree in Law.

陳先生為9GAG的行政總裁及共同創辦人。他於2008年成立了國際跨平台創意社群9GAG，以「令世界更快樂」為目標，讓用戶能輕易分享新穎有趣的內容和認識志同道合的朋友。

9GAG的每月觀眾逾2億人，包括逾5,600萬Instagram粉絲、逾4,200萬Facebook粉絲和逾1,600萬Twitter粉絲。

陳先生畢業於香港大學法律系。

Professor CHAN Wai-yee  
陳偉儀教授

Non-Official Director  
非官方董事



Professor Chan is the Pro-Vice-Chancellor/Vice President and Li Ka Shing Professor of Biomedical Sciences at The Chinese University of Hong Kong (CUHK). He obtained his BSc (Hon. 1st Class) in Chemistry from CUHK in 1974 and PhD in Biochemistry from the University of Florida in 1977.

In June 2009, Professor Chan established CUHK's School of Biomedical Sciences and served as the Founding Director and Chair Professor of Biomedical Sciences. He was appointed Pro-Vice-Chancellor/Vice President of CUHK in August 2018.

Professor Chan is very active in the scientific community, both locally and internationally. He has served as President of the Association of Chinese Geneticists in America and a Member of the Development Committee of the Society for the Study of Reproduction in the US. Besides being a Director of the Board of the Hong Kong Genome Institute, he is also the President of Hong Kong Institution of Science, Council Member of the Shaw Prize Foundation, a Member of the Hospital Authority, a Specialist for the Hong Kong Council for Accreditation of Academic and Vocational Qualifications, and a Member of the Board of Directors of Joshua Hellmann Foundation for Orphan Diseases.

陳教授為香港中文大學(中大)副校長及李嘉誠生物醫學講座教授。他於1974年在中大化學系一級榮譽畢業，並於1977年在美國佛羅里達大學取得哲學博士。

陳教授於2009年6月創立了中大生物醫學學院，擔任首任院長及生物醫學講座教授，並於2018年8月起出任中大副校長。

陳教授積極參與香港及海外多個專業組織的工作，曾任美洲華人遺傳學會主席及美國生殖學會發展委員會委員。除擔任香港基因組中心董事局成員外，他亦是香港科學會主席、邵逸夫獎理事會成員、醫院管理局非官方成員、香港學術及職業資歷評審局專家、香港夏約書孤兒症基金會董事會成員等。



The Board  
董事局

Ms Ivy CHEUNG Wing-han  
張穎嫻女士

Non-Official Director  
非官方董事



Ms Cheung is the Managing Partner and the Head of Audit of KPMG Hong Kong. She was the Past President of the Hong Kong Institute of Certified Public Accountants. She currently serves as member of various public service committees, including the Standing Committee on Disciplined Services Salaries and Conditions of the Service and the Transport Advisory Committee. Apart from being a member of the Advisory Committee, the Honorary Advisory Panel and the Inspection Committee of the Financial Reporting Council, she is the Director of Hong Kong Cyberport Management Company Limited and Insurance Authority.

Ms Cheung has served as Member of the Air Transport Licensing Authority, the Financial Reporting Review Panel, the Independent Commission on Remuneration for Members of the District Councils of the HKSAR, the Non-local Higher and Professional Education Appeal Board, the Occupation Retirement Schemes Appeal Board and the Securities and Futures Appeals Tribunal.

張女士為畢馬威香港區管理合夥人及香港審計服務主管合夥人。她是香港會計師公會前會長，目前於多個公共服務委員會擔任委員，其中包括紀律人員薪俸及服務條件常務委員會和交通諮詢委員會。張女士亦為財務匯報局諮詢委員會、名譽顧問團及查察委員會成員。她也是香港數碼港管理有限公司及保險業監管局之董事局成員。

張女士以往曾擔任空運牌照局、香港財務匯報檢討委員會、香港特別行政區區議會議員薪津獨立委員會、非本地高等及專業教育上訴委員會、職業退休計劃上訴委員會及證券及期貨事務上訴審裁處委員。

Dr CHUNG Kin-lai  
鍾健禮醫生

Non-Official Director  
非官方董事



Dr Chung is currently the Director of Quality and Safety of the Hospital Authority (HA). Under his leadership, the Quality & Safety Division oversees the quality standards, patient safety, clinical incident management, patient relations management, healthcare technology assessment, disasters response and infection control for the public hospitals under HA.

Dr Chung is a Specialist in Emergency Medicine. Prior to his current appointment, he has been the Hospital Chief Executive of North District Hospital from 2016 to 2018, Chief Manager at HA Head Office from 2012 to 2016 and the Hospital Chief Executive of Castle Peak Hospital from 2008 to 2012.

鍾醫生現任醫院管理局(醫管局)質素及安全總監，督導該局轄下公立醫院的質素及標準、病人安全、醫療事故管理、病人關係管理、醫療科技評估、災難應變以及感染控制等工作。

鍾醫生為急症專科醫生，出任現職前曾於2016至2018年出任北區醫院的醫院行政總監；2012至2016年任醫管局總辦事處的總行政經理；及於2008至2012年出任青山醫院的醫院行政總監。



The Board  
董事局

Professor Nancy IP Yuk-yu, SBS, BBS, MH, JP  
葉玉如教授, SBS, BBS, MH, JP

Non-Official Director  
非官方董事



Professor Ip is currently The Morningside Professor of Life Science and the Director of the State Key Laboratory of Molecular Neuroscience at The Hong Kong University of Science and Technology (HKUST). She received her PhD degree in Pharmacology from Harvard University, after which she held the position of Senior Staff Scientist at Regeneron Pharmaceuticals Inc. in New York. Since joining HKUST in 1993, she has served as the Vice-President for Research and Development, Dean of Science, Head of the Department of Biochemistry and Director of the Biotechnology Research Institute.

Professor Ip is world-renowned for her significant contributions to the field of neuroscience. Her outstanding research has resulted in more than 320 scientific papers and 70 patents. She has been elected to the Chinese Academy of Sciences, the US National Academy of Sciences, the American Academy of Arts and Sciences, the Hong Kong Academy of Sciences, and received numerous awards and honours including the National Natural Science Awards, the L'OREAL-UNESCO for Women in Science Award and the 10 Science Stars of China by *Nature*. She currently serves as a Member of the Leadership Group of the Davos Alzheimer's Collaborative, and was also an elected Councillor for the Society for Neuroscience and the Senior Editor of the *Journal of Neuroscience*.

葉教授現任香港科技大學(科大)晨興生命科學教授，以及分子神經科學國家重點實驗室主任。她在美國哈佛大學獲藥理學博士學位，其後在紐約Regeneron製藥公司擔任高級科學家，並於1993年起受聘於科大，先後出任副校長(研究及發展)、理學院院長、生物化學系主任及生物技術研究所所長。

葉教授是國際知名的神經生物學家，她的研究成果獲得了科學界的廣泛認同，在頂尖國際學術期刊發表了逾320篇論文和綜述，並擁有超過70項國際科技發明專利權。葉教授還當選中國科學院院士、美國國家科學院外籍院士、美國人文與科學院外籍院士、香港科學院創院院士；並獲頒多個國內外重要學術獎項，包括國家自然科學獎、歐萊雅聯合國教科文組織「世界傑出女科學家成就獎」及被《自然》科學雜誌選為中國科學之星。她目前擔任世界經濟論壇「達沃斯·阿爾茨海默症協作組織」領導小組成員；亦曾擔任美國神經科學學會理事會成員及《神經科學雜誌》資深編輯。

Professor LAU Chak-sing, BBS, JP

劉澤星教授, BBS, JP

Non-Official Director  
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Professor Lau is Dean and Daniel CK Yu Professor in Rheumatology and Clinical Immunology of the Li Ka Shing Faculty of Medicine at The University of Hong Kong (HKU).

Professor Lau graduated with MBChB from the University of Dundee in 1985. In 1992, he joined HKU as Lecturer in Medicine and successfully rose through the ranks to his current position as Chair and Daniel CK Yu Professor in Rheumatology and Clinical Immunology.

Professor Lau has been a major player in rheumatology in Hong Kong and beyond. Locally, he was President of the Hong Kong Society of Rheumatology (1997-2001) and Founding Chairman of the Hong Kong Arthritis & Rheumatism Foundation (2001). He was also the President of the Hong Kong Academy of Medicine (2016-2020), a statutory accrediting body for medical and dental specialist training in Hong Kong. Besides its academic role, the Academy is an entrusted advisor to the HKSAR Government on multiple health-related policies. In addition, Professor Lau sits on numerous strategic committees/working groups of the Health Bureau (formerly the Food and Health Bureau), Hospital Authority and Department of Health.

Regionally, Professor Lau was President of the Asia Pacific League of Rheumatology Associations (2006-2008) and co-founder of the multi-national, multi-centre Asia Pacific Lupus Collaboration.

Beyond the Asia Pacific region, he is a Member of the Outcomes in Rheumatology, European League Against Rheumatism (EULAR) Scientific Committee and EULAR Task Force on Rheumatoid Arthritis Treatment Recommendations.

劉教授為香港大學(港大)李嘉誠醫學院院長及於崇光基金風濕及臨床免疫學講座教授。

劉教授於1985年畢業於英國鄧迪大學內外全科醫學士課程，其後於1992年加入港大擔任內科學系講師，並晉升至於崇光基金教授席(風濕及臨床免疫學)。

劉教授是本港以至海外的風濕專科權威，由1997至2001年擔任香港風濕學學會主席，並於2001年成為香港風濕病基金會創會主席。他亦由2016至2020年擔任香港醫學專科學院主席。該學院是本地法定認證機構，提供醫學及牙科專科訓練。除了肩負推廣學術專業，學院亦獲香港特區政府委託為顧問，參與多項健康及醫療相關的政策工作。劉教授同時為醫務衛生局(前食物及衛生局)、醫院管理局及衛生署多個策略委員會及工作小組的成員。

在亞太地區方面，劉教授於2006至2008年擔任風濕病協會亞太聯盟主席，並為亞太狼瘡協會的聯合創辦人，致力促進跨地域及跨醫療中心的協作。

在國際層面，劉教授亦為多個專業組織的成員，包括風濕病治療成效小組，以及歐洲抗風濕病聯盟(EULAR)下的科學委員會及類風濕性關節炎治療建議工作小組。

The Board  
董事局

Dr Shawn LEUNG Shui-on  
梁瑞安博士

Non-Official Director  
非官方董事



Dr Leung is the founder, Chairman and Chief Executive Officer of SinoMab BioScience Limited. Currently, he is also a Member of the Biotech Advisory Panel of The Stock Exchange of Hong Kong Limited.

Dr Leung has over 30 years of experience in the field of molecular immunology and therapeutic monoclonal antibodies. He was the first scientist who successfully developed humanised anti-CD22 antibody and introduced the concept of “Functional Humanisation”. Dr Leung currently also serves as an Adjunct Professor at The Hong Kong University of Science and Technology, The Army Medical University (formerly known as the Third Military Medical University) and The Air Force Medical University (formerly known as the Fourth Military Medical University) in Mainland China. He held positions as the Executive Director of a leading US antibody-drug conjugate company, the Managing Director of The Hong Kong Institute of Biotechnology Limited, as well as the Adjunct Professor at Fudan University and The Chinese University of Hong Kong (CUHK).

Dr Leung obtained his BSc and MPhil in biochemistry, as well as EMBA from CUHK. He earned his D.Phil. in molecular biology from the University of Oxford in the UK in May 1989. He was also a postdoctoral fellow at Yale University in the US from July 1989 to June 1991.

梁博士為中國抗體製藥有限公司創辦人、主席兼首席執行官，現時亦為香港聯合交易所有限公司生物科技諮詢小組的成員。

梁博士在分子免疫學及治療單克隆抗體領域擁有逾30年經驗，為首位成功開發人源化抗CD22單抗及提出「功能人源化」概念的科學家。梁博士現時為香港科技大學、中國人民解放軍陸軍軍醫大學（前稱中國人民解放軍第三軍醫大學）及中國人民解放軍空軍軍醫大學（前稱中國人民解放軍第四軍醫大學）客座教授。他亦曾任美國免疫醫學公司行政總監、香港生物科技研究院院長，以及復旦大學和香港中文大學（中大）客座教授。

梁博士於中大取得生物化學學士及碩士學位，以及行政工商管理碩士學位。他於1989年5月在英國牛津大學取得分子生物學博士學位後，在1989年7月至1991年6月在美國耶魯大學從事博士後研究。

Dr Isabella LIU Fang-chun  
劉芳君博士

Non-Official Director  
非官方董事



Dr Liu is the Head of Baker McKenzie's Asia Pacific Intellectual Property (IP) and Technology Group. She advises clients on matters relating to the creation, exploitation and protection of IP rights. She is also responsible for the local IP Group's China and Hong Kong patent prosecution matters. Previously, Dr Liu was the Head of the Firm's Asia Pacific Healthcare and Life Sciences Industry Group for three years, leading a team of legal experts in this field cross multiple practices in the region.

Dr Liu is ranked as a leading lawyer in her field by top legal directories such as *Chambers Asia Pacific* for the Life Sciences category and *IAM Patent*. She has been complimented by clients that she possesses "a superb ability to understand the most complex technologies" and was noted for "advis[ing] in a way that is very commercial and strategic."

劉博士是貝克·麥堅時律師事務所亞太智慧財產權和科技業務部的負責人，為客戶提供有關智慧財產權的創建、運用和保護的諮詢服務，並負責在華智慧財產權的專利起訴事務。她的執業領域主要涉及廣泛的知識產權問題，包括專利和商標起訴、智慧財產權許可、技術轉讓、品牌收購以及智慧財產權的行政和民事執法。劉博士也曾擔任事務所亞太醫療保健產業組的負責人三年，領導跨法律專業的團隊在該領域發展，也為醫療保健行業的客戶提供與該行業有關監管問題的建議。

在頂級法律目錄（例如《錢伯斯亞太》和《IAMPatent》）中，劉博士被評為該領域的領先律師。客戶稱讚她擁有「理解複雜技術的精湛能力」，並因「以商業化和戰略性的方式提供諮詢」而聞名。

The Board  
董事局

Professor Dennis LO Yuk-ming, SBS, JP  
盧煜明教授, SBS, JP

Non-Official Director  
非官方董事



Professor Lo is the Li Ka Shing Professor of Medicine of The Chinese University of Hong Kong. His research interests focus on the biology and diagnostic applications of cell-free nucleic acids in plasma. In particular, he discovered the presence of cell-free fetal DNA in maternal plasma in 1997 and has since then been pioneering non-invasive prenatal diagnosis using this technology. This technology has been adopted globally and has created a paradigm in prenatal medicine. He has also made many innovations using circulating nucleic acids for cancer detection, including the screening of early stage nasopharyngeal cancer.

In recognition of his research, Professor Lo has been elected as Fellow of the Royal Society, Foreign Associate of the US National Academy of Sciences, Fellow of The World Academy of Sciences and Founding Member of the Academy of Sciences of Hong Kong. He has won numerous awards, including the 2016 Future Science Prize in Life Science, the 2014 King Faisal International Prize in Medicine, the 2019 Fudan-Zhongzhi Science Award and the 2021 Breakthrough Prize in Life Sciences.

盧教授現任香港中文大學醫學院李嘉誠醫學講座教授，其重點研究集中於血漿內游離DNA的生物學及診斷應用。於1997年，盧教授成為第一位發表有關孕婦血漿內發現胎兒游離DNA之研究的科學家，自此他一直處於這個嶄新研究領域的最前線。有關技術已被全球廣泛應用，並成為了產前胎兒醫學的範例。盧教授亦利用血漿游離核酸就癌症檢測作出了開創性的貢獻，特別是對於鼻咽癌的早期發現和監察有重大裨益。

盧教授的研究成果對全球醫學及科學界影響深遠，屢獲國際殊榮，當中包括2014年費薩爾國王國際醫學獎、2016年未來科學大獎生命科學獎、2019年復旦一中植科學獎及2021年科學突破獎—生命科學獎。盧教授亦被選為英國皇家學會院士、美國國家科學院外籍院士，以及香港科學院創院院士。



Professor Alfonso NGAN Hing-wan  
顏慶雲教授

Non-Official Director  
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Professor Ngan is currently Kingboard Professor in Materials Engineering and Chair Professor of Materials Science and Engineering at The University of Hong Kong (HKU). He previously held administrative positions including Senior Advisor in the President's Office, Acting Pro-Vice Chancellor (Research), Head of Department of Mechanical Engineering and Associate Dean of Engineering. He obtained his BSc(Eng) degree from HKU in 1989, and PhD from the University of Birmingham in the UK in 1992. After a year of postdoctoral training at the University of Oxford, he joined HKU in 1993, and was promoted through the ranks to Chair Professorship in 2011.

Professor Ngan's interest includes novel stimuli-responsive materials, material defects and their modelling, and nanomechanics including applications to biological systems. His research-related honours include the Rosenhain Medal from the Institute of Materials, Minerals and Mining in the UK, DSc from the University of Birmingham, Croucher Senior Research Fellowship, International Fellow of the Royal Academy of Engineering (FREng), Fellow of the Hong Kong Academy of Engineering Sciences (FHKEng), and Guanghua Engineering Science and Technology Prize. He is currently Honorary Secretary of the Hong Kong Academy of Engineering Sciences, and is serving in a number of advisory/management boards in the HKSAR Government.

顏教授現為香港大學(港大)建滔材料工程教授及材料科學與工程講座教授。他曾擔任的行政職務包括校長辦公室高級顧問、署理副校長(研究)、機械工程系系主任和工程學院副院長。他於1989年獲港大士學位、1992年獲英國伯明翰大學博士學位，並在牛津大學從事博士後工作，於1993年加入港大，2011年晉升為講座教授。

顏教授的研究興趣涵蓋新型刺激響應材料、材料缺陷及其機理，以及納米力學，包括在生物系統中的應用。其研究相關的榮譽包括英國材料、礦物和採礦學會Rosenhain獎章、伯明翰大學理學博士(D.Sc.)、Croucher高級研究員、英國皇家工程院外籍院士(FREng)、香港工程科學院院士(FHKEng)、光華工程科學技術獎等。他目前是香港工程科學院的名譽秘書長，並在香港特別行政區政府的數個顧問和管理委員會擔任委員。

The Board  
董事局

Mr Tim PANG Hung-cheong  
彭鴻昌先生

Non-official Director  
非官方董事



Mr Pang is a registered social worker dedicated in protecting and advocating patient's rights. He works as Community Organizer in Society for Community Organization. He has been a Member of the Hospital Authority (HA) Review Steering Committee, a Member of the Patient Focus Group of the Hospital Accreditation Project under HA, and a Member of the Committee on Promoting Acceptance of People Living with HIV/AIDS under Hong Kong Advisory Council on AIDS.

Mr Pang is now a Member of the Working Group on Implementation of Modified Referral System for Physiotherapy Services under the Physiotherapists Board, and a Member of the Working Group on eHRSS Partnership under the Steering Committee on Electronic Health Record Sharing.

彭先生是一名註冊社工，於香港社區組織協會擔任社區組織幹事，一直致力維護及倡議病人權益。他曾任醫院管理局（醫管局）檢討督導委員會委員、醫管局轄下醫院認證計劃病人焦點小組成員及香港愛滋病顧問局轄下接納愛滋病者促進委員會委員。

彭先生現為物理治療師管理委員會轄下實施物理治療服務更新轉介系統的工作小組成員，以及電子健康紀錄互通督導委員會轄下電子健康紀錄協作工作小組成員。



Professor YIU Siu-ming  
姚兆明教授

Non-Official Director  
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Professor Yiu is currently a professor and one of the Associate Heads at the Department of Computer Science of The University of Hong Kong (HKU). He is also the Director of the Department's FinTech and Blockchain Laboratory and the Deputy Executive Director of HKU-Standard Chartered Hong Kong FinTech Academy. He was selected three times by Clarivate Analytics as one of the Highly Cited Researchers in the world in 2016, 2017 and 2019, and one of the top 1% researchers in HKU for 11 consecutive years (2011-2021).

Professor Yiu's research areas include bioinformatics, cybersecurity, privacy technology, and FinTech. In the areas of bioinformatics, he served as the conference chair in Hong Kong for RECOMB 2017, one of the flag-ship conferences in the field and as the area programme chair for other prestigious bioinformatics conferences such as ISMB. In addition to academic research, Professor Yiu has been a consultant to various companies in the areas of cybersecurity and data privacy.

姚教授現任香港大學(港大)計算機科學系教授、副系主任及金融科技區塊鏈實驗室主任，同時為港大一渣打香港150週年慈善基金金融科技學院副行政總監。他曾於2016、2017和2019年獲Clarivate Analytics評為全球最廣獲徵引的研究人員之一，亦是港大連續11年(2011-2021年)排名前1%的研究人員之一。

姚教授的研究領域包括生物信息學、安全和密碼學以及金融科技。在生物信息學方面，他曾主持著名旗艦會議RECOMB 2017。除科研外，姚教授亦為不同企業擔任金融技術和網絡安全領域的顧問。

The Board  
董事局

Dr CHUI Tak-yi, JP  
徐德義醫生, JP  
Official Director  
官方董事



Dr Chui is the Former Under Secretary for Food and Health. At the time, his duties were to assist and support the Former Secretary for Food and Health in the setting of policy objectives and priorities on agriculture, fisheries, food safety, veterinary public health, environmental hygiene, medical and health, and related implementation issues, handling Legislative Council business and strengthening the working relationship with Legislative Council, and engaging and liaising with all stakeholders to explain and solicit support for government policies and decisions. Dr Chui graduated from the Faculty of Medicine, University of Hong Kong, in 1981. He has served in the public health care system of Hong Kong for more than 30 years.

(Appointment completed on 30 June 2022)

徐醫生為前食物及衛生局副局長，在任期間協助前食物及衛生局局長訂定漁農、食物安全、禽畜公共衛生、環境衛生、醫療及衛生範疇的政策目標和優先次序，以及處理相關的實施事宜；處理與立法會相關的事務和加強與立法會的工作關係；以及與各持份者溝通和聯繫，以解釋政府的政策和決定，並爭取他們的支持。徐醫生1981年畢業於香港大學醫學院，曾在香港公共醫療體系任職逾30年。

(任期至2022年6月30日)

Ms Shirley KWAN Yu-pik  
關如璧女士

Official Director  
官方董事



Dr Teresa LI Mun-pik, JP  
李敏碧醫生, JP

Official Director  
官方董事

Ms Kwan is currently Deputy Secretary for Health. During her tenure as the Official Director, she oversaw policy matters relating to the development of genomic medicine as well as healthcare manpower planning and professional development in Hong Kong. She was also responsible for the policies on mental health and the regulation of private healthcare facilities. In addition, she was in charge of the strategic planning and implementation of the Voluntary Health Insurance Scheme.

(Appointment completed on 3 July 2022)

關女士現為醫務衛生局副秘書長。在擔任官方董事期間，她的職責包括督導與基因組醫學的發展，以及本港醫療人力規劃和專業發展相關的政策事務。她亦負責精神健康和規管私營醫療機構的政策事宜，並處理自願醫保計劃的策略規劃和實施安排。

(任期至2022年7月3日)

Dr Li is a specialist in Public Health Medicine and currently the Deputy Director of Health. She oversees areas related to health services and administration including elderly health, family and student health, specialised services, administration and policy, finance, health administration and planning, and health information and technology.

李醫生是公共衛生醫學專科醫生，現任衛生署副署長，專責管理與衛生服務及行政相關的範疇，當中包括長者健康服務、家庭及學生健康服務、專科服務、行政及政策、財務、衛生行政及策劃，以及衛生資訊與科技的工作。



## Management Team 管理團隊



### Dr LO Su-vui 羅思偉醫生

#### Chief Executive Officer 行政總裁

MB Bch (Wales), FHKCCM, FHKAM (Community Medicine), FFPHM, MRCP (UK), FRACMA

英國威爾斯大學內外全科醫學士、香港社會醫學學院院士、香港醫學專科學院院士（社會醫學）、英國皇家內科醫學院公共衛生醫學科院士、英國皇家內科醫學院院士、澳洲皇家醫務行政學院院士

Dr Lo assumed office in March 2021, leading Hong Kong Genome Institute in its formulation of strategies and development plan. With the aspiration to benefit the wider community, Dr Lo drives the team to propel clinical application of genomic medicine and its long-term development in Hong Kong.

Dr Lo is a seasoned professional in public health and administrative medicine. Having held a number of senior positions both locally and overseas, Dr Lo brings with him a strong blend of experience in corporate affairs, strategic planning, talent development, research and so on. Prior to his current role, Dr Lo had served in the Hospital Authority (HA) for over 20 years during which he had held various senior management positions, including Director of Strategy and Planning in the HA Head Office and Cluster Chief Executive of the New Territories East Cluster.

羅醫生於2021年3月履新，領導香港基因組中心制訂發展策略，帶領團隊推動基因組醫學在香港的臨床應用及長遠發展，實現為社會大眾帶來裨益的願景。

羅醫生為資深公共衛生及行政醫學專家，先後於本地及海外擔任不同要職，在機構事務、發展規劃、人才培育及調查研究等範疇均具豐富經驗。出任現職前，羅醫生於醫院管理局（醫管局）服務逾20年，曾擔任不同管理職位，包括醫管局總辦事處策略發展總監及新界東醫院聯網總監。

### Mr Richard TSE Kin-pang 謝建朋先生

#### Chief Administrative Officer 首席行政總監

FCPA, FCA (Aus), FCG, HKFCG

香港會計師公會資深會員、澳洲資深特許會計師、特許秘書及公司治理師

Mr Tse joined Hong Kong Genome Institute in October 2020. He oversees corporate services and external affairs, leading on-going efforts to enhance corporate governance and operational efficiency, while raising public awareness on genomic medicine.

Mr Tse has extensive experience in administration and finance, spanning across financial and operations management, corporate services and governance. He was the Chief Financial Officer of the West Kowloon Cultural District Authority and held various senior management positions in established public bodies and multi-national corporations. He is also active in professional and community services and currently serves as a member of Branding and Communication Committee of the Hong Kong Institute of Certified Public Accountants.

謝先生於2020年10月加入香港基因組中心，負責管理機構的行政及對外事務，並專責持續優化企業管治及營運效益，深化市民大眾對基因組醫學的認識。

謝先生為資深行政及財務專業人員，在財務及營運管理、機構事務和公司管治等方面擁有豐富經驗。他曾擔任西九文化區管理局首席財務總監，並於多間大型公營機構及跨國企業出任管理要職。他同時積極參與專業和社會服務，現為香港會計師公會推廣及傳訊委員會成員。





## Dr Brian CHUNG Hon-yin 鍾侃言醫生

### Chief Scientific Officer 首席科學總監

MBBS (Hons, HKU), MSc (Genomics and Bioinformatics, CUHK), MD (HKU), DCH (Ireland),

MRCPCH (UK), FHKAM (Paediatrics), FRCPCH (UK), FCCMG (Clinical Genetics, Canada)

香港大學內外全科醫學士（榮譽畢業）、香港中文大學基因組學及生物信息學碩士、香港大學醫學博士、愛爾蘭皇家醫學院兒科文憑、英國皇家兒科醫學院院員、香港醫學專科學院院士（兒科）、英國皇家兒科醫學院榮授院士、加拿大醫學遺傳學專科學院院士

Dr Chung joined Hong Kong Genome Institute in February 2021, overseeing all scientific and clinical matters related to Hong Kong Genome Project. He also supervises the operation of HKGI's laboratory and works closely with Partnering Centres to mainstream genomic medicine.

Dr Chung was trained in Hong Kong and Canada, specialising in Paediatrics and Clinical Genetics. He was a founding fellow of the subspecialty of Genetics & Genomics (Paediatrics) of Hong Kong Academy of Medicine (HKAM) and was also actively involved in the drafting of respective postgraduate curriculum. With his excellence in research and teaching, Dr Chung has received a number of awards and honours over the years. He is active in international collaborations and is currently the President-Elect of the Asia Pacific Society of Human Genetics.

鍾醫生於2021年2月加入香港基因組中心，管理與香港基因組計劃相關的科學及醫學事務。他亦負責監督基因組中心實驗室的運作，並與夥伴中心緊密合作，加快基因組醫學的普及應用。

鍾醫生在香港及加拿大完成專科訓練，為兒科及臨床遺傳科專家，曾協助香港醫學專科學院成立遺傳學及基因組學專科（兒科），以及草擬相關的研究生課程。鍾醫生在研究及教學方面亦屢獲殊榮，並活躍於業界事務，現為亞太人類遺傳學會的候任主席。



## Professor Ken SUNG Wing-kin 宋永健教授

### Chief Bioinformatics Officer 首席生物信息總監

Ph.D. (Computer Science, HKU)

香港大學計算機科學博士

Professor Sung commenced the appointment in January 2021, overseeing the bioinformatics analysis and data management for Hong Kong Genome Institute to support clinical use. He also supervises the design and implementation of the data platform of Hong Kong Genome Project to facilitate research, while ensuring data privacy and security.

Professor Sung is experienced in Algorithm and Bioinformatics research, who has led the development of a number of bioinformatics software and has published a number of high impact papers in renowned academic journals. In recognition of his research contribution, he was conferred the National Science Award (Singapore) and the Young Researcher Award (NUS).

宋教授於2021年1月出任現職，負責香港基因組中心的生物信息分析及數據管理，支援臨床應用。他亦負責帶領團隊設計及推行香港基因組計劃專用的數據平台，致力確保數據保密及安全，以促進基因組醫學的研究發展。

宋教授在演算法和生物信息學研究擁有多多年經驗，曾開發多個生物信息學軟件，以及於多本國際知名科學期刊及雜誌發表了多篇高水平論文，傑出表現屢獲表揚，包括新加坡國家科學獎及新加坡青年研究員獎等。



## Corporate Information 公司資料

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Email: [enquiry@genomics.org.hk](mailto:enquiry@genomics.org.hk)

Website: [www.hkgp.org](http://www.hkgp.org)

### Auditor

Ernst & Young

### Principal Banker

Bank of China (Hong Kong) Limited

### Legal Advisor

Howse Williams

### Company Secretary

Howse Williams

### 地址

#### 香港基因組中心

香港沙田

香港科學園

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### 核數師

安永會計師事務所

### 主要往來銀行

中國銀行(香港)有限公司

### 法律顧問

何韋律師行

### 公司秘書

何韋律師行

# Chairperson's Statement

## 主席報告





## Chairperson's Statement 主席報告

It is a great honour for me to present the first annual report of the Hong Kong Genome Institute (HKGI) and to share with you the remarkable milestones we have achieved to date in laying a solid foundation for promoting better health and well-being for the people of Hong Kong with genomic medicine.

### Marking a Milestone for Hong Kong's Genomic Medicine Development

Genomic medicine has attracted global attention in recent years with its potential to transform healthcare management. The human genome provides essential clues to fundamental questions, such as why individuals are susceptible to certain diseases, and how these diseases can be detected and treated. This bears substantial implications for the formulation of public health policies, enabling us to more effectively allocate resources and address needs. With such far-reaching impact, Hong Kong is well set to leverage genomics and drive a shift towards precision medicine and personalised treatment for the benefits of the local population.

To this end, the establishment of HKGI by the HKSAR Government and our launch of the Hong Kong Genome Project (HKGP), the city's first large-scale genome sequencing project, marked a watershed moment for the development of genomic medicine in Hong Kong. It was the culmination of years of dedicated efforts and collaboration by many. These include the HKSAR Government and the Steering Committee on Genomic Medicine, the Health Bureau (formerly the Food and Health Bureau), and the Department of Health; the Hospital Authority and our Partnering Centres (PCs) at the Hong Kong Children's Hospital, Prince of Wales Hospital and Queen Mary Hospital; the medical schools of The Chinese University of Hong Kong and The University of Hong Kong; as well as countless medical professionals, patients, their families and friends. By coming together with shared vision and passion, we have embarked on an exciting journey to make genomic medicine a reality for Hong Kong.

### Building Strong Corporate Governance with World-renowned Experts

Starting from scratch is never easy. As a newly founded organisation with bold ambitions, there has been no shortage of challenges along the way. Yet with great pride, we are able to conclude the past twelve months with many impressive firsts. Among them, building strong corporate governance and strategic framework for HKGI within a short period unquestionably top the list.

我非常榮幸，在此與大家分享香港基因組中心(基因組中心)第一份年報，回顧我們首年全面運作至今所取得的重大成就，為本港基因組醫學發展奠定了堅實基礎，並朝着提升全港市民健康和福祉的目標邁進。

### 引領基因組醫學 開創新里程

基因組醫學潛力巨大，足以革新醫療服務，近年備受全球關注。一方面，人類基因組可為重大醫學難題提供關鍵線索，例如為何有些人較易患上某種疾病、如何及早發現並跟進治療等；另一方面，這些資訊對制訂公共衛生政策深具意義，可確保資源有效分配，應對社會所需。有見於基因組醫學的深遠影響，香港亦準備就緒，透過發展基因組學，推動精準醫療和個人化治療，以期為社會大眾帶來裨益。

為此，特區政府成立了基因組中心，並帶領團隊推行本港首個大型基因組測序項目「香港基因組計劃」(基因組計劃)，為香港發展基因組醫學開創新里程。我們得以順利推展基因組計劃，實有賴各方持份者群策群力，籌備多時，包括特區政府、基因組醫學督導委員會、醫務衛生局(前食物及衛生局)及衛生署。角色同樣重要的亦包括醫院管理局、香港兒童醫院、威爾斯親王醫院和瑪麗醫院三間夥伴中心、以及香港中文大學和香港大學的醫學院。當然，不可或缺的還有無數醫療專業人員、病人及他們的親友。我們和各方夥伴懷着共同的願景和熱誠，攜手開展這令人振奮的旅程，致力讓基因組醫學扎根香港，普及發展。

### 匯聚權威 穩健管治

從零開始殊非易事，基因組中心作為一家新成立機構，肩負重任，一路走來亦不乏挑戰。團隊在過去一年同心協力，迎難而上，開創了多項新猷，成績有目共睹，讓我們倍感自豪。其中最大成就之一，無疑是在短時間內，成功為基因組中心建立穩固的企業管治及策略框架。

Effective corporate governance is essential to the success of any organisation. Under the stewardship of the HKSAR Government, appointments to HKGI's first Board of Directors were confirmed in November 2020. Our Board comprises an exceptional roll call of world-class scientists, professors, clinical experts, as well as patient representative and top professionals from the accounting, legal and public relations sectors, providing HKGI with unwavering support and guidance on various fronts.

In addition, we had also formed six functional committees under the Board to ensure the effective implementation and monitoring of HKGI's key operations. These committees, all led by industry heavyweights and field experts, have been providing invaluable advice on a number of matters concerning the operations of HKGI and HKGP, such as scientific and ethical matters, data management, financial and administrative arrangements, audit and risks, communication and public education. The fact that close to 40 Board and committee meetings had been held within just a year and a half since HKGI's establishment speaks for the commitment of each member to establishing a firm footing for Hong Kong to develop genomic medicine. Thanks to the steadfast dedication and support from all members, many firsts were established along the way.

高效的企業管治是所有機構邁向成功的至要關鍵。基因組中心的首屆董事局於2020年11月獲特區政府正式任命，成員包括全球首屈一指的科學家、教授、臨床專家，以及病人組織代表和來自會計、法律及公共關係領域的業界精英，就基因組中心不同範疇的工作給予強大支持和指導。

此外，我們亦在董事局下設立了六個專責委員會，以有效落實和監察基因組中心的主要運作。各委員會均由業界翹楚和行內專才領導，為基因組中心和基因組計劃的相關工作建言獻策，如科學及倫理事務、數據管理、財務及行政管理、審計及風險評估、傳訊及公眾教育等。基因組中心成立至今只有短短一年半，在此期間，董事局和各個委員會已召開了近40次會議，足見各成員矢志推動本地基因組醫學發展，為香港建立鞏固根基的決心。有賴眾成員的堅實支持和寶貴貢獻，基因組中心得以屢創第一，引領發展。



## Chairperson's Statement 主席報告



Hong Kong is well set to leverage genomics and drive a shift towards precision medicine and personalised treatment for the benefits of the local population. The establishment of HKGI by the HKSAR Government and our launch of the Hong Kong Genome Project, the city's first large-scale genome sequencing project, marked a watershed moment for the development of genomic medicine in Hong Kong.

香港已準備就緒，透過發展基因組學，推動精準醫療和個人化治療，以期為社會大眾帶來裨益。我們很高興，特區政府成立了基因組中心，並帶領團隊推行本港首個大型基因組測序項目「香港基因組計劃」，為香港發展基因組醫學開創新里程。



Our strong senior management team is also crucial to our corporate governance, as well as our remarkable progress and accomplishments to date. Despite being a nascent organisation that is relatively small in size, we are proud to have completed the recruitment of the chief officers and senior staff members by the first quarter of 2021, bringing on board veterans in public health, administrative, scientific and bioinformatics professions to steer HKGI towards full operations.

就建立高水平的企業管治而言，基因組中心的管理團隊同樣擔當着重要角色，帶領機構順利推展工作，創出佳績。雖然基因組中心成立的日子尚短，規模亦相對較小，但我們已充分展現工作成效，於2021年第一季便完成管理團隊及主要高級人員的招聘，成功吸納了公共衛生、行政、科學和生物信息學等專業領域的資深人才加入，加快基因組中心全面投入運作，成績令人鼓舞。



## Defining Corporate Vision and Strategic Plan to Accelerate Growth

We well understood setting directions and priorities during the inception stage weighs as much as establishing a robust corporate governing structure. Following the on-boarding of key staff members, we promptly defined HKGI's vision, mission and corporate values, translating our aspiration – *to avail genomic medicine to all for better health* – into laymen language that resonates with our target stakeholders. By articulating and highlighting our objectives, purposes, and guiding principles, we ensure that they are effectively shared across the organisation from the outset and clearly conveyed to the wider community.

Recognising the complexity of our work, we further developed HKGI's first three-year *Strategic Plan* to guide us in all aspects of planning and development for the coming years from 2022 to 2025. With the endorsement from the Board, we identified four strategic foci, including integrating genomic medicine into clinical care, nurturing talents in genomic medicine, advancing research in genomic science, and enhancing public genomic literacy. This well-thought-out roadmap enables us to address key challenges and issues, and move towards achieving our vision and mission. This is undoubtedly a significant achievement for HKGI as a newly established organisation.

## 確立願景 提速發展

我們深明於基因組中心成立初期，除了建立完善的企業管治架構，策劃機構的發展方向和工作優次亦至關重要。因此，我們在管理團隊履新後，隨即為基因組中心確立願景、使命和核心價值，並以簡潔易明的文本展現我們的抱負——「普及基因組醫學，共享健康福樂」，讓持份者有所共鳴。我們透過反覆說明和闡述，確保基因組中心的目標、使命和原則得以有效廣傳，從內而外，從同事以至市民大眾，清晰準確地將訊息傳揚至社區。

與此同時，鑒於基因組中心工作的複雜性，我們特意制訂了基因組中心首個三年策略計劃，旨在為機構在2022至2025年期間各方面的規劃和發展建立明確目標，指引方向。在董事局的支持下，我們確立了四大策略重點，包括融合基因組醫學與臨床護理、促進基因組科學研究、培育基因組醫學人才及加強公眾對基因組學的認識和參與。這份策略計劃思慮周全，勾劃了基因組中心的發展路向，有助團隊應對各項重大挑戰，專心致志朝着願景和使命邁進。作為一家新創機構，這絕對是一項重大成就，讓我們引以為傲。



## Chairperson's Statement 主席報告

### Pioneering Genomic Firsts for Hong Kong

Essential to realising all our aspirations and ambitions is HKGP, which we successfully launched in July 2021. The HKGP is the first-of-its-kind project in Hong Kong set to focus on three key areas: undiagnosed diseases, hereditary cancers and patient cases related to genomics and precision health that would benefit from the technology of whole genome sequencing.

Through rolling out HKGP, we led the effort to establish essential infrastructure, protocols and best practices to integrate genomic medicine into clinical care. We also laid down a strong foundation for building a genome database of the local population that is critical to enhancing clinical applications of genomic medicine and inspiring research locally and worldwide. The database we are building will help improve the under-representation of the Asian/Chinese genomes in the global scientific community. Potential insights drawn from it will facilitate more precise diagnosis, including conditions common to the local population, promote personalised treatment and disease prevention in the long run.

### Establishing a Bridge with the Public

With genomic medicine being a relatively new subject, education is essential to instilling understanding and trust among patients and their families, the medical community, and the general public. Accordingly, one of our first priorities in the first year of operations was to engage with the PCs and medical professionals to provide education and training.

To reach out to the public, in particular the target participants of HKGP, we produced a variety of user-friendly resources to offer clarity on the work of HKGI and HKGP. We collated information packages for adult and children patients participating in HKGP, explaining details and answering common questions clearly and concisely. In addition to this, we produced a series of multimedia content in the form of short introductory videos to convey our messages transparently and raise public awareness. All of these are vital to enhancing genomic literacy and engaging people to our cause.

### 開拓基因組學 屢創新猷

為了實踐使命，實現願景，我們於2021年7月成功推展基因組計劃。該計劃是全港首個同類項目，主要涵蓋三個可受惠於全基因組測序技術的範疇，包括未能確診病症、與遺傳有關的癌症，以及與基因組學及精準醫學有關的個案。

藉着推行基因組計劃，我們率先為業界建立所需基礎設施、制訂規程和最佳實務守則，加快融合基因組醫學與臨床護理。此外，我們亦為建立本地人口基因組數據庫奠定穩固基礎，促進基因組醫學的臨床應用，並啟發本地及國際科研。現今全球科學及醫學研究均十分缺乏華人的基因組數據，而我們致力建立的數據庫，正有助填補這個重大缺口。長遠而言，從中的分析研究所得，將有助實現更精準診斷，尤其是常見於本地人口的疾病，並促進個人化治療及疾病預防，意義深遠。

### 建立橋樑 聯繫各界

基因組醫學作為一門新興課題，必須透過公眾教育，以加深病人及家屬、醫學界和社會大眾的認識，從而建立互信。因此，我們首年運作的重點工作之一，便是與夥伴中心及醫療專業人員建立緊密聯繫，為他們提供持續進修和培訓。

為了廣泛接觸公眾，尤其是基因組計劃的目標參加者，我們製作了各式各樣內容豐富的教育資訊，深入淺出地介紹基因組中心的工作和基因組計劃。我們亦為基因組計劃的參加者編製了資料冊，並特意設計了成人及兒童版本，詳細講解計劃詳情及常見問題。除了刊物外，我們還製作了不同系列的多媒體內容，透過主題短片生動有趣地簡介基因組醫學，提高公眾對這個課題的認知。這些努力，均有助我們加強公眾對基因組學的認識和參與，並對基因組中心的工作予以肯定。

## Achieving More as One

The past year is definitely a challenging yet fruitful and rewarding year for HKGI. We are very fortunate to have received generous support from our stakeholders, partners and friends. My deepest gratitude goes to colleagues from the Health Bureau and former Food and Health Bureau, Department of Health, Hospital Authority, our PCs, the medical schools of The Chinese University of Hong Kong and The University of Hong Kong, and all healthcare professionals for their tremendous support.

I would also like to express my sincere thanks to our committed Board and committee members for their invaluable support and wise counsel. My gratitude also goes to the management team and all HKGI staff members for their dedicated professionalism and magnificent teamwork under the leadership of our inaugural Chief Executive Officer Dr Lo Su-vui. Particularly, I am much grateful for the valuable trust and support from our patients and their families for walking with us to bring genomic medicine closer to everyone in Hong Kong.

May we work together to realise the prospects and benefits of genomic medicine, improve the well-being of the people of Hong Kong, and enrich genomic medicine for the rest of the world. I am fully confident that with our concerted efforts and determination, bolstered by our solid foundations, robust governance, and pragmatic strategic plan, we can learn from the best and brightest, accelerate our growth, and progress as planned.

As one, we will achieve many more firsts in the days and years to come.



Philip TSAI Wing-chung, BBS, JP  
Chairperson

## 團結一致 成果更豐

對基因組中心而言，過去一年縱然滿途挑戰，卻亦成果豐碩。我們非常榮幸，沿途獲不同持份者、合作夥伴和各界友好全心奉獻，傾力支持。我謹在此代表基因組中心，衷心感謝醫務衛生局和前食物及衛生局、衛生署、醫院管理局、三間夥伴中心、香港中文大學和香港大學的醫學院，以及所有醫療專業人員的鼎力支持。

基因組中心在年內成就甚豐，實有賴董事局和委員會各成員一直熱心參與，給予寶貴的支持和指導，貢獻良多，我在此由衷致謝。我亦非常感謝基因組中心的管理團隊和全體同事，在首任行政總裁羅思偉醫生的領導下，上下一心，專業高效，展現了非凡的團隊精神。與此同時，我必須特別感謝病人和他們家人對基因組中心的信任和支持，與我們並肩同行，推動本地基因組醫學發展，以期惠及每一位香港市民。

願我們攜手合作，發揮基因組醫學的巨大潛能，為社會帶來裨益，與市民共享健康福樂，並為全球基因組醫學發展作出貢獻。我深信，只要大家同心同行，加上扎實的根基、穩健的管治和務實的策略計劃，取法乎上，我們定能加快步伐，奮進前行。

憑着團結一心，我們必可繼往開來，再創新猷，成就更多。



主席  
蔡永忠, BBS, JP

## Chief Executive Officer's Report 行政總裁報告





It gives me immense pleasure to share with you the remarkable breakthroughs we made during the first year of full operations at Hong Kong Genome Institute (HKGI) on promoting genomic medicine in Hong Kong.

As the theme of this Annual Report underlines, 2021-22 has been a year of outstanding achievements, with solid foundation built and challenges overcome. We spent the past dozen months diligently laying the groundwork and assembling the building blocks for the development of genomic medicine in Hong Kong. We made strenuous efforts to carry forward with our initiatives and stayed true to our purpose, the HKGI vision – *availing genomic medicine to all for better health and well-being*. Translating words into actions, we started with building a start-up, and with strong footing.

### Beginning with the Basics

Establishing a new institution is nothing short of challenging. It is doubly true for HKGI, one charged with the mission to propel a new branch of science and medicine that has the potential to revolutionise public healthcare services. Commencing operations from ground zero, we focused on the very basics: people and infrastructure.

Human capital is, among other things, fundamental to the success of every organisation. Nothing is possible without people; yet with the right team, everything becomes achievable. This is why talent acquisition has always been one of our top priorities ever since HKGI was incorporated. Although the body of genomic specialists remains small in Hong Kong, during the year, we were able to assemble an exemplary multi-disciplinary team of scientists, clinicians, bioinformaticians, genetic counsellors, genome curators, laboratory professionals and researchers, thanks to the tremendous support from the former Food and Health Bureau (currently the Health Bureau) of the HKSAR Government during our inception stage. In addition to experts from the scientific and bioinformatics fields, we brought on board seasoned veterans to manage HKGI's administrative functions. From finance and human resources to corporate communications; information technology and corporate secretariat to office administration, all have been instrumental in keeping HKGI up and running.

這是香港基因組中心(基因組中心)全面運作後的第一份年報，我很高興在此與大家分享這一年來，我們就推動本地基因組醫學發展取得的重大突破。

正如本年報的主題所展示，2021-22年是基因組中心成就卓越、奠定根基、克服挑戰的一年。我們恪盡職守，傾力為香港基因組醫學發展建立穩固基礎，實踐基因組中心的願景——「普及基因組醫學，共享健康福樂」。我們化願景為行動，從零開始有序推進，堅守宗旨，穩扎根基。

### 建立機構 以人為本

建立一家全新的機構絕非易事，我們既要面對各種挑戰，亦肩負重任，推動足以革新公共醫療服務的新興科學及醫學專科。這對基因組中心來說，絕對是雙重考驗。有見及此，我們在營運伊始便聚焦鞏固根基，全力招募人才及構建基礎設施。

我們深明人才是每家機構成功的關鍵，成事在人，只要擁有精練能幹的團隊，凡事皆可成。因此，自基因組中心成立以來，我們一直以招募人才為首要任務之一。環顧香港，基因組學相關領域的專家屈指可數；儘管如此，全賴香港特區政府前食物及衛生局(現為醫務衛生局)於籌備階段的鼎力支持，助我們在這一年間籌組了一支陣容強大的跨專業團隊，當中包括科學家、醫生、生物信息學家、遺傳輔導員、基因組數據分析員、實驗室專業人員及研究人員等。除了科學及生物信息學專家，我們亦網羅了業界專才執掌行政事務，在財務、人力資源、機構傳訊、資訊科技、機構秘書，以至辦公室行政等方面皆顧及周全，確保基因組中心運作順暢如流。

## Chief Executive Officer's Report 行政總裁報告

By mid-2021, we formed the core team and kicked into high gear to take forward the highly demanding preparation of our inaugural flagship initiative, the Hong Kong Genome Project (HKGP). We attracted the best and brightest to join our ranks, including home-grown experts working abroad to come back for this seminal project. In just a year's time, the HKGI family has expanded from nought to a high calibre and committed team of over 60 and counting.

及至2021年年中，我們的主要團隊陸續履新，並全速為基因組中心首個重點項目「香港基因組計劃」(基因組計劃)展開繁重的籌備工作。我們很高興成功吸納業界精英，包括在海外工作多年的香港專家回流，加入基因組中心，攜手推展這個意義深遠的項目。在短短一年間，我們由零開始默默耕耘，至今已招攬了逾60名業界專才，組成眾志一心的團隊。



### Building Infrastructure to Drive Success

To ensure colleagues are provided with a conducive work environment, we put equal emphasis on setting up essential infrastructure, both hardware and software. From operating out of separate satellite offices across Hong Kong Science Park temporarily at the very beginning, in less than six months, we completed the fitting-out works of our new workplace in the Park in August 2021. The smart office, designed with ample collaborative areas and thoughtful details to ensure occupational safety, efficiency, and productivity, has empowered colleagues to excel at work and benefit from the innovative culture within the technology hub.

### 完善基建 引領成功

良好的工作環境是確保團隊發揮所長，事半功倍的關鍵，故此我們着力為同事提供必要的基礎設備，包括硬件和軟件。在基因組中心成立之初，我們於香港科學園內不同大樓設立了臨時分部，暫作日常營運，並同步籌建全新的辦公總部，在不到半年的時間，便於2021年8月順利完成設計及裝修工程。我們的辦公總部位於科學園，配備了智能設施及寬敞的協作空間，每個細節均照顧到同事們日常工作所需，既確保職業安全，亦有效提升工作效率及生產力。同事們在園內創新文化的薰陶下，思維視野亦更廣更闊。



Establishing a new institution is nothing short of challenging. It is doubly true for HKGI, one charged with the mission to propel a new branch of science and medicine that has the potential to revolutionise public healthcare services. We spent the past dozen months diligently laying the groundwork for the development of genomic medicine in Hong Kong, forging ahead with the HKGI vision – *availing genomic medicine to all for better health and well-being.*

建立一家全新的機構絕非易事，我們既要面對各種挑戰，亦肩負重任，推動足以革新公共醫療服務的新興科學及醫學專科。這對基因組中心來說，絕對是雙重考驗。在過去的一年，我們恪盡職守，傾力為香港基因組醫學發展建立穩固基礎，實踐基因組中心的願景——「普及基因組醫學，共享健康福樂」。



In addition to the setting up of robust hardware, networks and systems, we developed a comprehensive set of corporate governing policies, procedures and protocols addressing operational needs in areas such as procurement and human resources to ensure effective oversight and the highest performance.

除了完善的硬件設施、穩定可靠的網絡和系統，我們亦制訂了全面的企業管治政策、程序及規程守則，以配合基因組中心在採購、招聘等多方面的營運需要，確保團隊在高效的督導下，發揮最佳表現。



## Chief Executive Officer's Report 行政總裁報告

### Setting Precedents with Hong Kong Genome Project

Central to the impressive strides HKGI made during the year was the successful launch of HKGP, the first large-scale whole genome sequencing project in Hong Kong. Following a few months' intensive preparation and assiduous efforts on all fronts, HKGP was soft launched in July 2021 with the first participant recruited. It was followed by the full launch in October 2021, with the core infrastructure, teams, standards and protocols in place. Despite the challenges posed by the pandemic, the Project has progressed well, with over 6,000 samples collected to date.

The smooth roll-out of HKGP not only marks a great leap forward in our bid to integrate genomics into clinical care to benefit patients and the wider community, but also represents a landmark development in genomic medicine for Hong Kong and beyond. The significance is multifaceted. On top of the direct patient benefits HKGP brings about through more precise diagnosis and personalised treatment, it also serves as a catalyst to establish a genome database of the local population, testing infrastructure and talent pool for Hong Kong.

Internationally, HKGP sets precedents, protocols and standards for the end-to-end operations of similar projects in different regions, from establishing partnerships with the authorities and public hospitals, to obtaining informed consent from patients; from setting up testing infrastructure to building bioinformatics platform; from managing data security to conducting training for healthcare professions and many more. The Project also adds the missing piece to the puzzle of the global genome database by increasing the availability of Asian/Chinese genomes, allowing us to participate in global genomics to improve diversity and promote equity.

### Teaming Up with Authorities to Create Synergy

In implementing the first-of-its-kind genome project in Hong Kong, the trust of patients, the medical community and the public all hinges on the ethics, expertise, and professionalism of our team. It is also dependent on the impeccable standards of our operations, the strength and capabilities of our technology and infrastructure, and our proactive outreach to stakeholders in introducing to them this new avenue of healthcare service.

We have lived up to our corporate values and stayed versatile, forging ahead on multiple fronts at lightning speed. Shortly after our core staff members came on board, we worked immediately and closely with the Hospital Authority, The Chinese University of Hong Kong, and The University of Hong Kong to set up three Partnering Centres (PCs) at the Hong Kong Children's Hospital, Prince of Wales Hospital and Queen Mary Hospital to help recruit eligible participants for HKGP. Riding on their patient network, expertise and extended resources, we have been able to create synergy in our common pursuit of catalysing the development of genomic medicine in Hong Kong.

### 推展「香港基因組計劃」 開創先河

過去一年，基因組中心在多方面深耕細作，其中最重大的里程碑，定是成功推展了基因組計劃。此計劃為香港首個大規模全基因組測序項目，我們能夠於2021年7月順利招募首名參加者，實有賴各方夥伴與我們攜手合作，於短短數月內傾力籌備。及後，隨着各項核心基礎設施、團隊人員、標準及規程守則等逐一完備，我們亦於2021年10月全面推展基因組計劃。縱使疫情帶來挑戰，計劃仍然進展良好，迄今已收集了逾6,000個樣本。

基因組計劃的順利推行，不但象徵香港在融合基因組學與臨床護理方面向前邁進了一大步，造福病人和社會大眾，更為本地和海外的基因組醫學發展寫下重要一頁，影響深遠。就香港而言，基因組計劃除了透過更精準診斷及個人化治療直接惠及病人，亦有助加快步伐，建立本地人口基因組數據庫、測試設施及人才庫。

在國際層面上，我們為基因組計劃所制訂的規程與準則，開創了「一站式」運作的先河，為各地相類計劃提供了寶貴的參考實例。整個運作模式涵蓋全面，由夥拍政府部門及公立醫院，到獲取病人知情同意；由建立測序設施，到構建生物信息平台；由管理數據安全，到培訓醫護人員等，環環相扣。此外，基因組計劃所累積的亞裔／華人族裔的基因組數據，更有助填補全球基因組數據庫中的重大缺口，推動科學和醫學研究多元發展及公平參與，讓香港為國際基因組學發展作出貢獻。

### 夥拍同儕 協同創效

基因組計劃是本港首個同類型項目，如何獲取病人、醫學界及大眾的信任，取決於多項因素；而計劃團隊的操守、知識和專業精神，絕對是關鍵所在。其他決定性因素還包括團隊有否恪守嚴謹的營運準則、所採用的技術和設備是否安全可靠、有否積極聯繫持份者解說這門嶄新的醫療服務等，這些均是建立信任的基石。

在籌備基因組計劃的過程中，我們秉持基因組中心的核心價值，靈活應變，迅速於多個領域取得成果。在主要團隊上任後，我們旋即與醫院管理局、香港中文大學及香港大學緊密合作，於香港兒童醫院、威爾斯親王醫院及瑪麗醫院設立了三間夥伴中心，為基因組計劃招募合資格參加者。我們透過夥伴中心的病人網絡、專業知識及龐大資源，發揮協同效益，攜手加快香港基因組醫學的發展。



### Building Public Trust with Impeccable Ethical Standards

In connection to patient recruitment, apart from securing critical partnerships with authorities most trusted by the local community, we also take much pride in having developed a rigorous informed consent mechanism which ensures the recruitment process is carried out according to the highest standards of medical ethics around the world.

During the year, we produced a comprehensive Informed Consent Package detailing the consent processes for adult, teenage and child participants, the consent forms used, the authorisations required, and the data protection precautions taken under the guidance of the ethics experts on our Ethics Advisory Committee, and in consultation with the PCs as well as professional bodies, including the Office of the Privacy Commissioner for Personal Data and the Equal Opportunities Commission.

As a result of months of sedulous deliberation and consultation, the Institutional Review Boards of the three PCs endorsed the Package and granted ethics approval to HKGP in July and August 2021, while approval from the Department of Health's Ethics Committee was obtained in January 2022. With the positive feedback received, it has been clear our safeguards and high ethical standards established have earned us the trust and confidence of our stakeholders across patients, the healthcare community and the general public.

### 標準從嚴 建立公信

在招募參加者方面，除了與備受社會信賴的政府部門、醫院及大學緊密合作，我們亦制訂了嚴謹的知情同意機制，確保招募過程按照全球最高醫學倫理標準進行，相關工作成果，讓我們深感自豪。

我們在年內為基因組計劃的參加者編製了一套「知情同意資料冊」，仔細闡釋了計劃團隊向成人、青少年和兒童參加者徵詢知情同意的程序，包括參加者需確認的知情同意書及授權的內容等。資料冊亦詳細講解了基因組中心就保護數據所採取的措施，而我們在敲定有關細節前，除了獲倫理諮詢委員會的專家提供指導，亦諮詢了夥伴中心及相關專業機構，包括個人資料私隱專員公署及平等機會委員會，並獲他們正面回應。

經過數月的深入討論和諮詢，三間夥伴中心所屬的研究倫理委員會先後於2021年7月及8月完成審批資料冊的內容，並就基因組計劃發出倫理准許，確認計劃通過道德操守審查；而衛生署倫理委員會亦於2022年1月發出相關准許。從我們收到的正面回饋，足證團隊制訂的保障措施的嚴謹的倫理標準，成功取信於病人、醫護界同儕及社會大眾等持份者，讓我們深受鼓舞。



## Chief Executive Officer's Report 行政總裁報告

### Opening Doors for Genomic Research

Being a city-wide large-scale sequencing initiative, HKGP requires the vital support of an effective and efficient whole genome sequencing service platform and system built according to internationally accredited laboratory standards. I am glad to report that during the year, such support has been progressively developed. While tapping into external expertise and resources from a qualified research centre housed under a well-received local university at the initial stage of HKGP's launch, our team has gradually built up the capacity and capability required to drive long-term success.

On the scientific front, we established our own Genomic Laboratory with state-of-the-art equipment to provide in-house sequencing services and facilitate multi-omics studies. The laboratory commenced operations in December 2021 after timely completion of fitting-out works and installation of advanced sequencing equipment, platforms and systems. This new world-class facility will undoubtedly be the cradle of genomic discoveries, where scientists, clinicians, bioinformaticians, researchers and field experts all work together to explore and inspire, delivering benefits to all from bench to bedside.

### 鞏固實力 啟迪科研

基因組計劃是全港性大規模測序計劃，我們必須依循國際認可的實驗室標準，構建高效的全基因組測序平台和系統作為強大後盾，方可確保計劃順利推行。我很高興在此匯報，團隊在年內已按步完成相關工作，進度理想。在基因組計劃出台之初，我們一方面夥拍本地知名大學旗下的研究中心，借助其專業知識和資源推展計劃；另一方面亦抓緊時機，逐步建立團隊實力，為取得長遠成功奠下基礎。

在科研方面，我們建立了專屬的基因組實驗室及配置了頂尖設備，既有助鞏固團隊內部進行測序的能力，亦可促進多組學研究。隨着裝建工程按時完成，先進的測序儀器、平台及系統迅速就位後，實驗室於2021年12月正式啟用。這個達世界級水平的全新科研設施，定必成為孕育基因組學創新突破的搖籃，讓科學家、醫學專家、生物信息學家、研究人員和不同領域的專家在此並肩合作，交流探索，相互啟發，將研發成果從實驗室帶到臨床應用，惠及大眾。





Central to the impressive strides HKGI made during the year was the successful launch of the Hong Kong Genome Project, which marks a great leap forward in our bid to integrate genomics into clinical care. Apart from bringing about more precise diagnosis and personalised treatment to patients, the Project also serves as a catalyst to establish a genome database of the local population, testing infrastructure and talent pool for Hong Kong.

過去一年，基因組中心在多方面深耕細作，其中最重大的里程碑，定必是成功推行基因組計劃，帶領香港在融合基因組學與臨床護理方面向前邁進了一大步。基因組計劃除了透過更精準診斷及個人化治療直接惠及病人，亦有助加快步伐，建立本地人口基因組數據庫、測試設施及人才庫。



### **Establishing Highly Effective Bioinformatics Platform**

A highly performant bioinformatics platform to support analysis and interpretation of whole genome sequencing data is pivotal to the success of any large-scale genome project, including HKGP. During the year, we spared no effort to develop the necessary bioinformatics infrastructure to support patient recruitment, operation of the PCs as well as sequencing services of HKGP.

### **構建平台 高效運作**

對任何大型基因組項目來說，包括香港基因組計劃，若要取得成功，必須建有高效的生物信息平台，方可支援全基因組測序所涉及的大量數據分析及詮釋工作。為此，我們於本年度全力構建重點生物信息學基礎設施，以支援基因組計劃在病人招募、夥伴中心運作，以及測序服務等多方面的工作。

## Chief Executive Officer's Report 行政總裁報告

By the fall of 2021, we successfully rolled out two sophisticated platforms that are fulcrums of the HKGP operations, the Clinical Frontend and the Laboratory Information Management System (LIMS). While the Clinical Frontend enables the HKGP teams to register patients, obtain their consent, and collect relevant clinical information and bio-samples, the LIMS allows for managing the production of raw next-generation sequencing data from the bio-samples. Bioinformatics pipelines were also developed to process the sequencing data and to annotate genomic variants with known disease associations. A thorough Privacy Impact Assessment on our bioinformatics platforms had also been conducted by an independent auditor, certifying our adherence to stringent international standards in data privacy and cybersecurity. All these laid a strong foundation for the implementation of HKGP and our subsequent work.

### Developing Talent Pool for Genomic Medicine

The complexity and magnitude of genomic medicine necessitate a variety of skilled and competent professionals to maximise potential. In addition to scientists, clinical geneticists, genetic counsellors, and bioinformaticians, we are in great need of genome curators, laboratory professionals, medical technologists and many others. Together, this dream team will make genomic discoveries, develop technologies and translate research findings into clinical applications, realising the advantages of personalised treatment and precision medicine.

To bring this dream team into reality, we have been undertaking the dual role of enabler and educator to catalyse the building of a highly skilled talent pool for developing genomic medicine in Hong Kong. As an enabler, we spearheaded various initiatives to support the continuing professional development programmes in genetics and genomics for clinicians, nurses and allied health professionals, empowering them with the latest knowledge and skillsets to take up new and enlarged roles. For starters, we engaged the clinical and health professionals of our PCs through training and multi-disciplinary team meetings to promote exchange and align professional practices. To extend our reach, we secured collaborations with acclaimed professional bodies, including the Hong Kong Academy of Medicine and the Hong Kong Academy of Nursing to coordinate efforts in promoting genomic medicine.

在同事們同心協力下，我們於2021年第三季成功建立了兩個設定精密的系統，包括臨床資訊管理平台(Clinical Frontend)及實驗室資訊管理平台(Laboratory Information Management System)，作為基因組計劃日常運作的重要支柱。臨床資訊管理平台讓基因組計劃團隊可便捷地為病人登記，向他們徵詢知情同意，並記錄所收集的臨床資訊和生物樣本；實驗室資訊管理平台則協助團隊有效管理以生物樣本進行新一代測序所得的原始數據。我們亦設計了生物信息管理流程，用作處理測序數據，以及標註與已知疾病相關的基因變異。另一方面，我們亦邀請了獨立顧問為相關流程及平台進行全面的私隱影響評估，確定了基因組中心在數據私隱和網絡安全方面均符合嚴謹的國際標準。以上種種努力成果，為我們推行基因組計劃及落實日後的工作奠定了穩固根基。

### 多元培訓 儲備人才

基因組醫學博大精深，其複雜程度和規模之大，需要不同範疇的專家學者群策群力，方可發揮最大潛力。因此，推動香港的基因組醫學發展，我們除了需要科學家、臨床遺傳學家、遺傳輔導員及生物信息學家，基因組數據分析員、實驗室專業人員、醫務化驗師，以及其他相關範疇的人才同樣不可或缺。這支陣容強大的夢之隊，定可為基因組學研究帶來突破、開發嶄新技術，並將科研成果轉化為臨床應用，發揮個人化治療及精準醫療的巨大優勢。

為羅致團隊所需人才，我們自基因組中心成立以來，便一直身兼推動者和教育者，全力為香港建立高技術的人才庫，加快本地基因組醫學的發展。作為推動者，我們牽頭推出多項措施，包括為醫生、護士及專職醫護人員提供持續進修，助他們掌握遺傳學及基因組學的最新知識和技能，為擴闊職能作更佳準備。舉例說，我們透過舉辦培訓講座及跨專業團隊會議，讓夥伴中心的臨床及醫護人員聚首一堂，既促進交流分享，亦有助統一常規實務的流程和標準。為進一步擴大業界網絡，我們亦與香港醫學專科學院，以及香港護理專科學院等獲認可的專業團體合作，同心合力推廣基因組醫學。



As an educator, we are committed to fuelling the talent pool and nurturing leaders of tomorrow in genomic medicine. Apart from providing on-the-job training for the young talents at HKGI, we have particularly focused our efforts on engaging tertiary students. To instil in them the understanding and appreciation of genomic medicine and encourage them to pursue careers in the field, we have been having positive dialogues with several educational institutes. While we had completed our first internship programme in the summer of 2022, we look forward to organising placement programmes alike in the coming years.

作為教育者，我們致力為基因組醫學建立人才庫，培育未來領袖。我們除了為團隊中的年青同事提供在職培訓，亦積極接觸大專學生，與多間院校緊密聯繫，期望透過不同活動加深同學們對基因組醫學的認識，了解這門學科的意義和精粹，並鼓勵他們投身相關工作。隨着基因組中心首個暑期實習計劃於2022年夏季完滿結束，我們將舉辦更多同類計劃，栽培年輕人才。



### Founding Industry Networks to Achieve More

To take talent development further, we went the extra mile to initiate the setting up of the Hong Kong Genetic Counselling Practice Consortium, considering the absence of an accreditation mechanism for genetic counsellors in Hong Kong. The groundwork on the drafting of the Consortium's objectives, membership composition and work plan has already been completed and we are confident in concluding its official formation in the coming year. Another example of our strategic move lay with bioinformaticians, as we formed a collaborative communication network for local experts at our PCs and academia to facilitate regular knowledge and experience exchange. To maximise the network effect, we also set up a network of genomic champions in different clinical specialties of public hospitals to help spread the knowledge.

### 創建網絡 凝聚專才

為進一步培育人才，我們採取主動，構建業界網絡，推動專業發展。有見香港尚未就遺傳輔導設立認可機制，我們於年內牽頭籌備成立香港遺傳輔導專業發展聯席，並已完成多項前期工作，包括草擬組織宗旨、成員架構和工作計劃等，有信心於未來一年正式成立。我們的努力，亦見於為本地生物信息學家建立協作網絡，有效地連繫夥伴中心及各大学院專家學者，定期交流知識和經驗。為了發揮更大影響力，我們亦於公立醫院各個臨床專科，建立了基因組學倡議網絡，透過傑出人員及團隊的實務分享，樹立楷模，加快知識的傳承和應用。

## Chief Executive Officer's Report 行政總裁報告

### Bringing Genomic Medicine Closer to the Community

A parallel operation to developing talents and promoting genomic knowledge is raising public awareness of genomic medicine and HKGI's work. With tireless efforts throughout the year, we have stayed on track to enhance genomic literacy and engagement.

A number of public education and stakeholder engagement initiatives were launched. To start with, a wide range of authoritative and user-friendly publications, videos and animations were produced to introduce the fundamentals of genomic medicine and the establishment of HKGI and HKGP. Engagement events such as corporate visits and media briefing were also hosted to publicise our work among the local community through prominent online and offline coverage in major mass media outlets. All these laid the stage for HKGP and our subsequent achievements.

Above all, in this digital era, continuous enhancement of HKGI's website and online presence has always been the centrepiece of our communication strategy. The website, augmented with lively graphics and a user-friendly design, has served as an imperative engagement tool since its launch in the second half of 2021. Keeping abreast of the ever-evolving communication trends, we look forward to further strengthening HKGI's digital footprint in the future to maximise our publicity effectiveness.

### Engaging Stakeholders Near and Far

Our publicity efforts saw no borders during the year. Globally, we forged close ties with an array of renowned experts from the United States, the United Kingdom, Canada, Australia and so forth through various engagement discussions and sharing sessions. They were all invested in the success of HKGI and HKGP, and fully subscribed to our cause to promote genomic diversity to inspire scientific and clinical breakthroughs.

All these publicity campaigns helped instil positive understanding of genomic medicine and appreciation of our work among targeted stakeholders, building a solid footing for HKGI both locally and internationally. We are well-placed to further deliver the promise and advancement of genomic medicine in the years to come.

### 宣傳教育 深入社區

除了培育人才及促進專業發展，我們亦致力深化大眾對基因組醫學及基因組中心各項工作的認識。有賴同事們全年努力不懈，我們就加強公眾教育及參與的工作已上軌道，成效顯著。

我們透過不同形式和宣傳渠道，廣泛接觸社會大眾，推廣基因組醫學。舉例說，我們編製了一系列既具權威性又顯淺易明的公眾教育刊物、影片及動畫，深入淺出地講解基因組醫學的基本知識，並簡介籌備基因組中心及基因組計劃的背景目的。此外，我們亦舉辦了參觀活動及傳媒簡報會等，透過大眾媒體線上線下的廣泛報道及龐大的讀者網絡，深入社區宣傳基因組中心的工作，為基因組計劃及後續的正面發展打下穩固基礎。

與此同時，身處數碼時代，我們尤其重視網絡宣傳，以此為重點傳播策略，持續優化基因組中心的網站及加強網上曝光。我們的網站圖像解說豐富生動，版面設計方便易用，自2021年下半年推出以來，一直擔當着基因組中心與社會大眾溝通的重要橋樑。我們將繼續緊貼日新月異的網絡傳播趨勢，進一步擴大基因組中心的數碼足跡，務求達致最佳宣傳效益。

### 連繫持份者 不分遠近

在年內，我們連繫持份者的工作，無分國界遠近。除了本地社區，在國際間，我們亦積極與美國、英國、加拿大及澳洲等世界各地不同學科的專家學者建立緊密聯繫，舉辦了多場交流會，分享經驗和業內最新發展。我們樂見各方對基因組中心及基因組計劃均大力支持，熱切期待與我們共同推動基因組學多樣發展，為科學及臨床醫學帶來突破，共享成果。

透過以上各項宣傳推廣，我們成功連繫海內外目標持份者，確立了他們對基因組醫學的了解，並對基因組中心的工作予以肯定。無論是在香港還是國際社會，基因組中心已穩佔一席，團隊亦已作好準備，將繼續推動基因組醫學向前發展，為社會大眾帶來裨益。



## Looking Forward to a Bright Future

Hong Kong's foray into genomics has started later than some other developed jurisdictions. Nevertheless, I firmly believe that by adopting a systematic and coordinated approach, we will be able to deploy a genome project tailored to the needs of the local population.

Along this journey, we are privileged to have received the unstinting support and guidance from stakeholders around us – committed colleagues from the Health Bureau (and the former Food and Health Bureau) and the Department of Health; world-renowned experts on our Board and committees; dedicated cross-disciplinary medical professionals from the Hospital Authority, the PCs and the medical schools of the two globally acclaimed universities; courageous patients and their families, and every single member of the healthcare community and the public who has been supporting us behind the scenes. We would not have been able to navigate through the challenges without each of them standing by us.

My deepest thanks also go to the ever-faithful HKGI colleagues and the management team. None of our work would be possible without the dedication, professionalism and passion of the entire team who has gone above and beyond to contribute to HKGI and HKGP.

In 2021-22, we laid down a solid foundation and turned a new page for the development of genomic medicine in Hong Kong. By coming together, I have every confidence that we will create even more exciting chapters in the years ahead.



**Dr LO Su-vui**  
Chief Executive Officer

## 高瞻遠矚 展望未來

香港發展基因組學的時機，或較同類發達地區稍晚；但我堅信，只要緊循策略藍圖及工作優次，基因組中心定能繼續以穩步有序，高效協作的方針，切實推行本地人口所需的基因組計劃。

在這個從零開始的旅程上，我們非常榮幸獲各界持份者鼎力支持和指導，其中包括醫務衛生局（以及前食物及衛生局）和衛生署的同事、我們董事局及各委員會中享譽全球的專家學者、以及來自醫院管理局、夥伴中心及兩所國際級大學醫學院的跨學科醫學專才。此外，同樣至關重要的是每位勇毅前行的病人和他們的家人，以及在背後默默支持我們的醫護同儕和社會大眾。衷心感激大家與我們並肩同行，基因組中心才能乘風破浪，克服種種挑戰，全速發展。

我亦在此向基因組中心的同事們和管理團隊致以由衷謝意，全賴大家傾力奉獻，以專業及熱誠為基因組中心及基因組計劃竭盡所能，方可成就這豐盛一年。

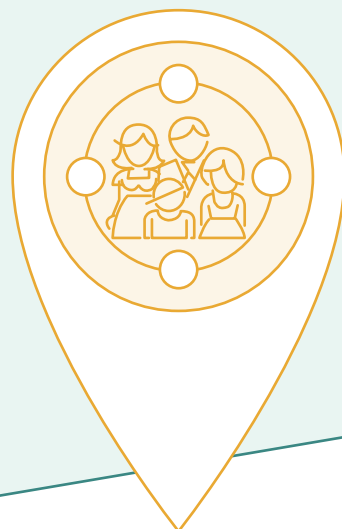
回顧2021-22年度，基因組中心已扎穩根基，為香港的基因組醫學發展揭開了嶄新一頁。我深信未來的日子，在大家同心協力下，我們定能譜寫出更精彩的篇章，共創更健康未來。

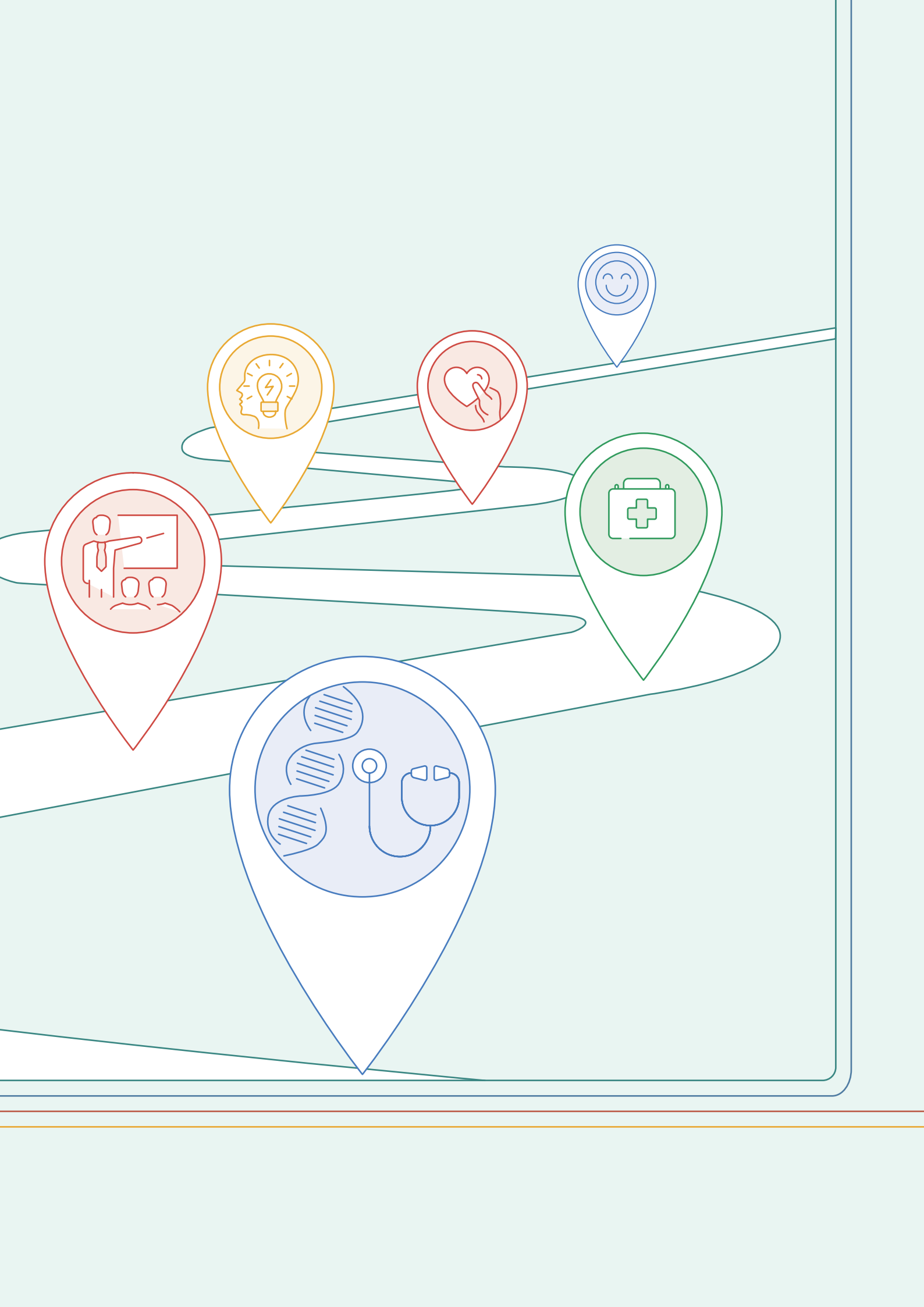


行政總裁  
**羅思偉醫生**

# Milestones of the Year

## 年度回顧





## Established strong corporate governance with world-renowned experts and industry veterans 匯聚國際專家 鞏固企業管治

Having developed and maintained robust corporate governance practices has been the cornerstone of the Hong Kong Genome Institute's (HKGI) accomplishments to date.

成功建立穩健的企業管治，鞏固常規實務，為基因組中心的眾多成就奠下基石。



The HKGI Board comprises an exceptional roll call of world-class scientists, professors and clinical experts, as well as patient representative and top professionals from the accounting, legal and public relations sectors. Together with six functional committees, the members have been providing unwavering support and guidance to HKGI.

基因組中心董事局由國際知名的科學家、教授、臨床專家，以及病人組織代表和來自會計、法律和公共關係的業界精英組成，連同六個專責委員會，一直以來共同為基因組中心提供強大支援和指導。

Signed a Memorandum of Administrative Arrangements with the HKSAR Government, which provided a clear framework for the relationship between HKGI and the HKSAR Government, and the responsibilities of each party.

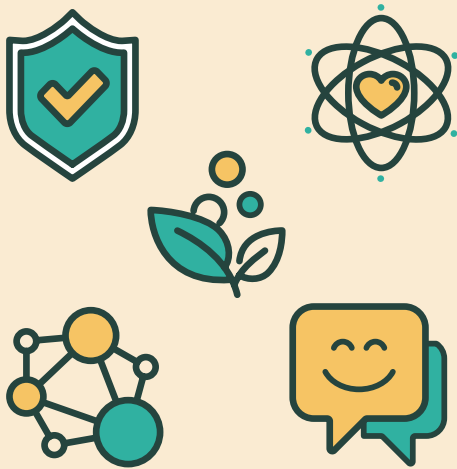
與特區政府簽訂《行政安排備忘錄》，為基因組中心與特區政府之間的關係及雙方的職責，提供清晰框架。



## Defined corporate vision and strategic plan to guide future development 確立願景策略 引領未來發展

Set out a strategic roadmap and priorities for HKGI to capture opportunities and drive long-term success.

為基因組中心奠定策略方向及工作優次，以把握機遇，推動長遠發展。



Defined HKGI's vision, mission and core values for clear articulation across the organisation and among our stakeholders to promote the vision of making genomic medicine available to all for better health and well-being.

確立基因組中心的願景、使命及核心價值，積極向不同持份者清晰闡明，以宣揚「普及基因組醫學，共享健康福樂」的願景。

Formulated HKGI's first three-year development blueprint *Strategic Plan 2022-25* to achieve the corporate vision through four strategic foci: integrate genomic medicine into clinical care, advance research, nurture talents, and promote public genomic literacy.

制訂基因組中心首個三年發展藍圖《2022-25年策略計劃》，並透過四大策略重點，包括融合基因組醫學與臨床護理、促進基因組科學研究、培育基因組醫學人才及加強公眾對基因組學的認識和參與，實現機構願景。





## Started off operations from ground zero 從零開始 運籌帷幄

Jumpstarted corporate operations from scratch with strong support from the former Food and Health Bureau (currently the Health Bureau), kicking HKGI into high gear on all fronts.

基因組中心從零開始，在前食物及衛生局（現為醫務衛生局）的鼎力支持下，各方面均迅速發展。



Completed fitting-out works of the HKGI office at Hong Kong Science Park to provide staff members with an environment conducive to enhanced efficiency and productivity.

完成基因組中心位於香港科學園的辦公室裝修工程，為同事們提供良好的工作環境，以提升效率及生產力。





Brought on board a dedicated multi-disciplinary team of clinicians, scientists, bioinformaticians and administrative professionals. Since its establishment, the HKGI family has expanded from zero to over 60 to date.

廣納不同業界專家、醫生、科學家、生物信息學家等加入，組成跨專業團隊。自成立至今，基因組中心已擴展為逾60人的大家庭。



## Pioneered the first city-wide genome project in Hong Kong 推展首個基因組計劃 開創全港先河

Rolled out the Hong Kong Genome Project (HKGP), setting precedents for end-to-end operations of the first large-scale genome sequencing project in Hong Kong. These ranged from obtaining informed consent from patients, setting protocols and testing infrastructure, to building bioinformatics platform, managing data and much more.

順利推展本地首個大型基因組測序項目「香港基因組計劃」(基因組計劃)，從向病人徵取知情同意到制訂規程；從建立測試設施及生物信息平台到管理數據等，均以「一站式」運作管理，為香港開創先例，堪稱典範。



Set up three Partnering Centres (PCs) at the Hong Kong Children's Hospital, Prince of Wales Hospital and Queen Mary Hospital to help recruit eligible HKGP participants with informed consent. Workflow and protocols were established with training sessions conducted for each PC to align the professional practices. Strong support from the Department of Health, Hospital Authority, the medical schools of The Chinese University of Hong Kong and The University of Hong Kong, and the three PCs had been indispensable to the smooth implementation of HKGP.

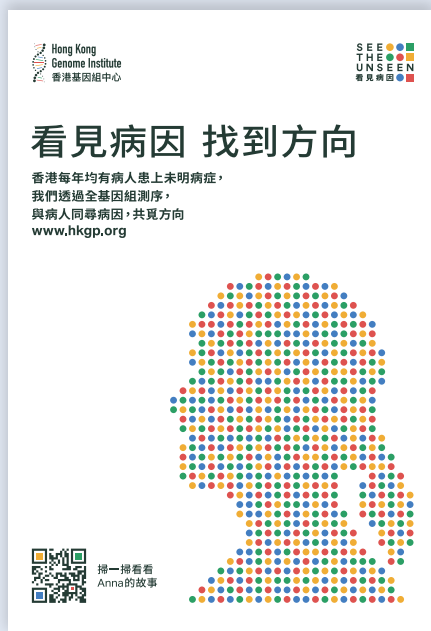
就基因組計劃於香港兒童醫院、威爾斯親王醫院及瑪麗醫院設立三間夥伴中心，在合資格參加者的知情同意下，招募他們參與計劃。基因組中心亦為每間夥伴中心確立工作流程及規程，並提供培訓，以統一實務標準。全賴衛生署、醫院管理局、香港中文大學及香港大學醫學院，以及三間夥伴中心的大力支持，基因組計劃得以順利推行。





Launched HKGP successfully, focusing on undiagnosed diseases, hereditary cancers and diseases related to genomics and precision health. To support patient recruitment and public education, a series of printed and multimedia materials such as information booklet, leaflet and souvenirs tailored for child and adult HKGP participants, as well as a number of videos and animations were produced to promote HKGP, HKGI's work and genomic medicine.

成功推出基因組計劃，重點涵蓋未能確診病症、與遺傳有關的癌症，以及與基因組學及精準醫學有關的個案。為支援病人招募及公眾教育，基因組中心製作了一系列刊物及多媒體資訊，包括為兒童及成人計劃參加者編製的小冊子、宣傳單張和紀念品，以及多部影片及動畫，以推廣基因組計劃、基因組中心的工作及基因組醫學的重要性。





Established HKGI's Genomic Laboratory with state-of-the-art equipment to provide in-house sequencing services to enhance the capability and capacity of whole genome sequencing in Hong Kong in the long run.

建立基因組實驗室，配備頂尖設施，鞏固團隊進行測序的能力，長遠提升本港在全基因組測序領域的實力。



Started the preparation of building an in-house bioinformatics platform with customised analysis tools to enhance HKGI's capability to process whole genome sequencing samples and multi-omics studies.

籌建生物信息平台，配合需要訂制分析工具，以提高基因組中心處理全基因組測序樣本及多組學研究的能力。





## Catalysed the development of talent pool 發揮催化作用 加快人才培育

Enhanced genetic and genomic knowledge of healthcare professionals and attracted both local and international talents through various initiatives.

發揮推動者的角色，加強醫療專業人員對遺傳學及基因組學的認識，並透過不同措施吸納本地及國際人才。



Committed to building a talent pool by enhancing the genetic and genomic knowledge of existing healthcare professionals through on-the-job training, while also initiating positive dialogues with universities and professional bodies to promote student internship, professional training and accreditation.

舉辦在職培訓，助現職醫療專業人員持續進修，增進遺傳學和基因組學的相關知識，同時積極與大學和專業團體商討合作，就實習、專業培訓和認證作長遠規劃，着力建立人才庫。



Supported the professional development of staff members through a series of training on a variety of topics such as cybersecurity and root-cause analysis to empower their performance at work.

為團隊舉辦一系列不同主題的培訓，如網絡安全和根源分析法等，以提升同事的專業發展和工作表現。



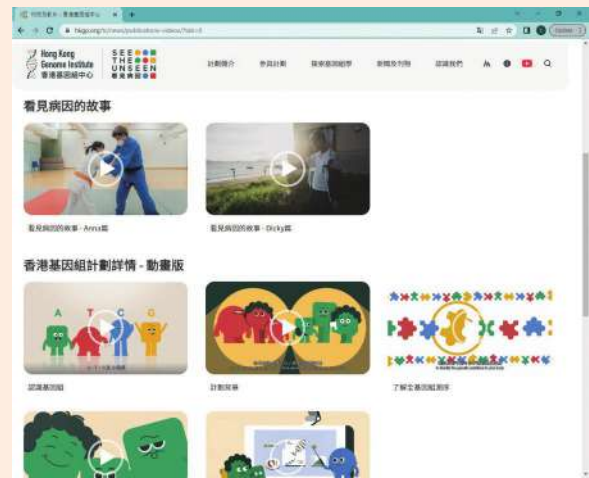
## Built up network with stakeholders through proactive engagement 廣泛接觸持份者 建立業界網絡

Proactively engaged with local stakeholders and international counterparts to publicise HKGI's work and facilitate exchange of industry knowledge and best practices.

積極與本地持份者及國際同業建立聯繫，以推廣基因組中心的工作，促進行業知識及實務交流。

Launched HKGI corporate website as an important tool for a start-up like us to engage with our stakeholders, especially patients, medical professionals and the general public. Designed to be user-friendly, the website gives visitors easy access to information about HKGP and other valuable resources, while also promoting HKGI's significant role in driving the development of genomic medicine in Hong Kong.

建立機構網站，作為基因組中心與持份者，包括病人、醫療專業人員和社會大眾聯繫的重要平台。網站設計方便易用，讓訪客可便捷地閱覽基因組計劃的詳情及相關資訊，並加深了解基因組中心在推動香港基因組醫學發展的重要角色。



Engaged with experts and counterparts from the UK, Australia and other regions proactively to exchange knowledge and explore collaborations. The connections established allowed HKGI to take its work to the international scene and build up Hong Kong's global reputation in the future.

積極與英國、澳洲及其他地區的專家和同儕建立緊密聯繫，交流知識和探討合作，所積累的業界網絡將有助提升基因組中心的國際地位，並為香港在全球建立聲譽。





Hosted the visit for former Secretary for Food and Health, Professor Sophia Chan, at HKGI office and one of the PCs, the Hong Kong Children's Hospital, to report on the implementation progress of HKGP and exchange views about our work.

接待前食物及衛生局局長陳肇始教授，並一同參觀夥伴中心之一的香港兒童醫院，向陳教授匯報了基因組計劃的進度，以及就基因組中心的工作交流意見。



Hosted HKGI's first media briefing to introduce the organisation and HKGP. As a result of targeted media pitching, prominent press coverage was generated in print and online to foster understanding and appreciation of HKGI's work among the general public.

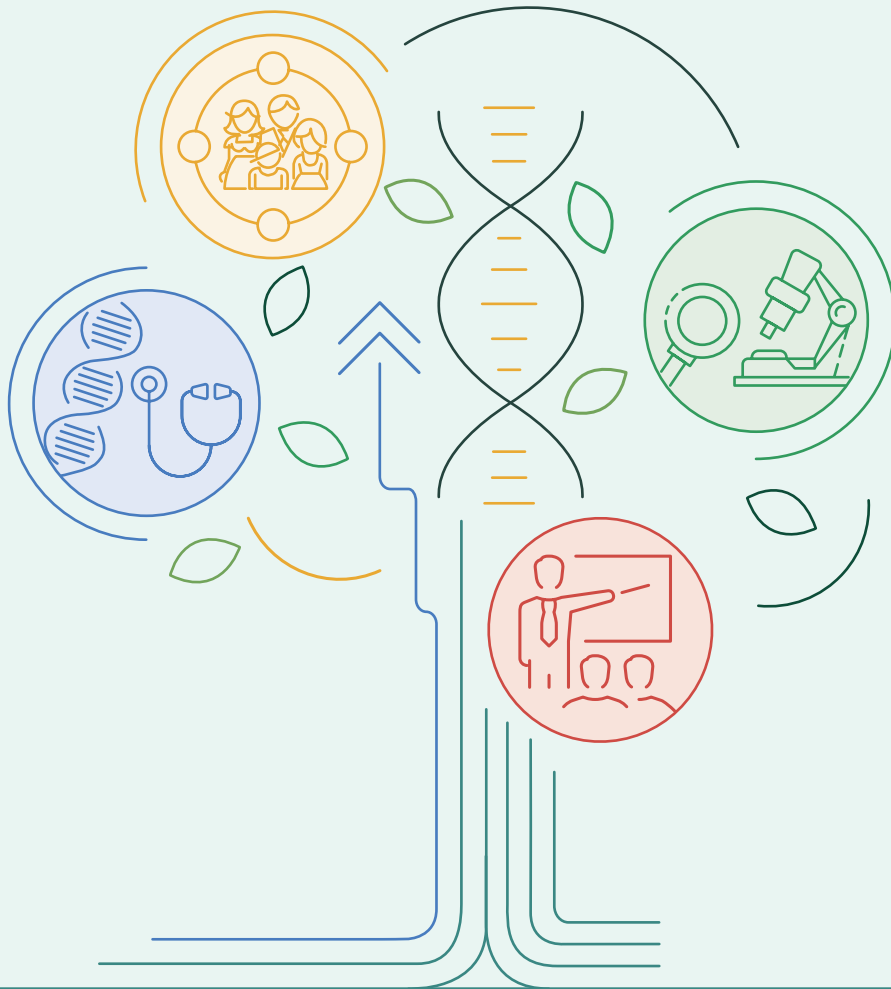
舉辦首個傳媒簡報會，向目標主流媒體詳盡解說基因組中心的成立背景和基因組計劃詳情，獲得多篇正面報道，有助深化公眾對基因組中心工作的了解。

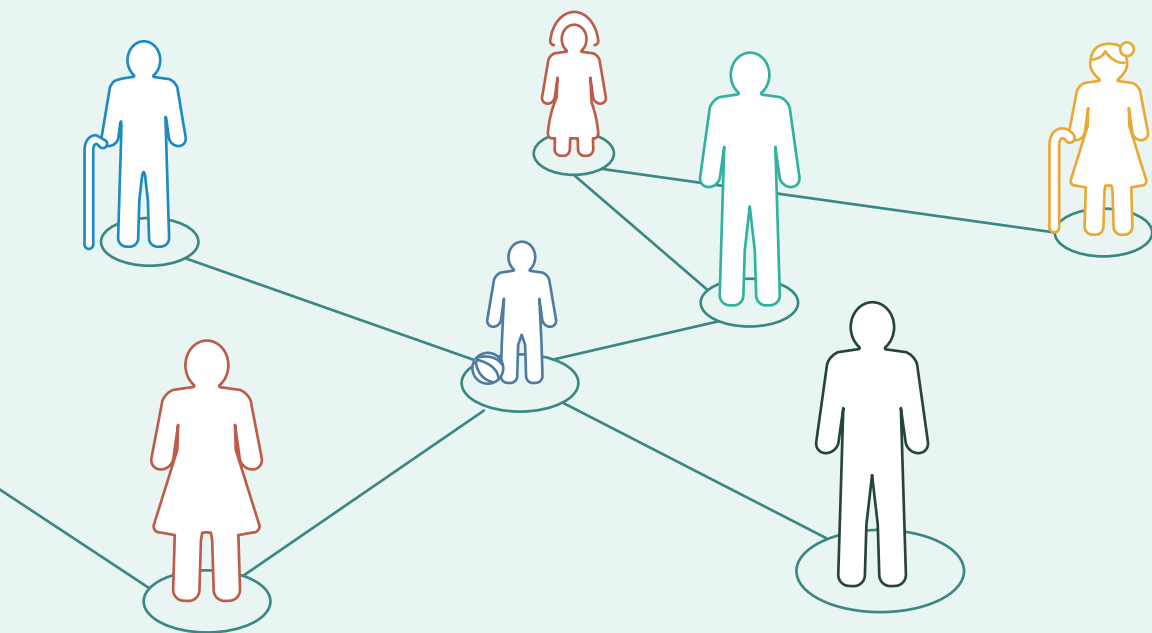




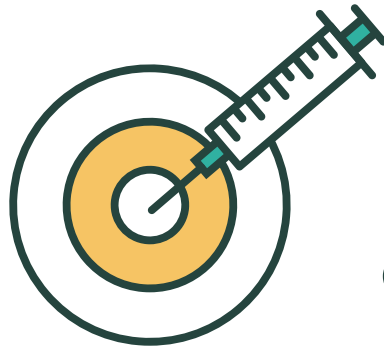
# Laying the Foundation for Better Health

固植根基 樂享健康









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



# Integrate Genomic Medicine into Clinical Care

## 融合基因組醫學與臨床護理

Since the Human Genome Project, a significant international research undertaking, was completed and published the first sequence of the human genome in 2003, genomic medicine has made great strides worldwide. Advances in genomic research and whole genome sequencing have vastly expanded our understanding of DNA, genetics and genomics, while research outcomes and medical breakthroughs have opened new doors for disease screening, accurate diagnosis, personalised treatment, and better disease prevention.

### Genomic Medicine Picking Up Pace in Hong Kong

In Hong Kong, passionate clinicians at the Department of Health, the Hospital Authority, universities, and private hospitals have provided high-quality clinical genetics services to the public for many years. Yet, the overall development of genomic medicine in Hong Kong remains scattered compared to other advanced jurisdictions. Patients with undiagnosed diseases often have to consult multiple specialists before their diseases can be identified. Those in need of testing have to sometimes turn to overseas testing services, which is costly and causes undue anxiety and inconvenience.

To take Hong Kong's genomic medicine development to the next level, the HKSAR Government's Steering Committee on Genomic Medicine called for a comprehensive strategy to boost the development, clinical application, and research of genomic medicine. It recommended the launch of a large-scale genome sequencing project focusing on the local population, which will "serve as a catalyst and anchor for showcasing the clinical benefits, piloting related new policy measures, building up a talent pool, and testing clinical protocols."

### Launching Infrastructure for Genomic Medicine

The Hong Kong Genome Institute (HKGI) was thus established to accelerate the development of genomic medicine in Hong Kong. As the first step towards achieving this mission, HKGI commenced full operation and launched the Hong Kong Genome Project (HKGP) in 2021 with the plan to conduct whole genome sequencing for 20,000 cases in five years. As the city's first large-scale genome sequencing project, HKGP serves as a catalyst to benefit patients and their families with more precise diagnosis and personalised treatment through whole genome sequencing. It also aims to establish a genome database of the local population, testing infrastructure and talent pool to address the healthcare needs of Hong Kong in the long run.

國際研究項目人類基因組計劃於2003年完成，並公布首個人類基因組序列，自此，基因組醫學於全球取得長足發展。憑着基因組研究和全基因組測序日漸成熟，我們可加深了解DNA、遺傳學和基因組學，其研究成果和醫學突破亦提升了疾病篩查、準確診斷、個人化治療及疾病防控等不同層面的潛力。

### 策劃藍圖 促進發展

衛生署、醫院管理局、大學和私家醫院的醫生一直竭誠為香港市民提供優質的醫學遺傳服務。然而，相較其他先進國家和地區，香港基因組醫學的整體發展較為分散。患有未能確診病症的病人通常要在諮詢多名專家後才能找到病因，而有需要進行測序的病人有時只能尋求海外的服務，不僅費用高昂，亦帶來不必要的擔憂和不便。

為促進本港基因組醫學進一步發展，香港特區政府轄下的基因組醫學督導委員會制訂了全面的策略，以促進相關領域的發展、臨床應用及研究。督導委員會亦建議推行以本地人口為主的大型基因組測序計劃，作為「催化基因組醫學發展的基石，展示臨床裨益、試行相關的新政策措施、建立人才庫，以及試行相關的臨床規則。」

### 完善基建 應對需要

香港基因組中心(基因組中心)旨在推動本港基因組醫學發展。作為實現願景的第一步，基因組中心於2021年正式全面運作，並開展了香港基因組計劃(基因組計劃)，目標於五年內為20,000宗個案進行全基因組測序。基因組計劃作為本港首個大型基因組測序計劃，扮演著催化劑的角色，以全基因組測序讓病人及其家屬受惠於更準確診斷及個人化治療，並透過建立本地人口的基因組數據庫、測試設施及人才庫，應對香港長遠醫療需要，與大眾同創健康未來。

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

# Human Genome Project 人類基因組計劃

The Human Genome Project (HGP) is one of the most ambitious and significant biomedical research endeavours in human history.

In 1990, scientists from different countries and disciplines, such as biology, physics, chemistry and engineering, came together to launch this highly collaborative initiative to study and sequence all the DNA (i.e. the genome) in the human body.

With more than a decade and tremendous efforts from thousands of scientists, the first draft sequence of the human genome was completed in 2003. The DNA samples from the anonymised volunteers were analysed and combined to form a “human reference genome”. Scientists and clinicians from around the world can now compare an individual’s DNA with this reference to identify disease-causing differences (i.e. genetic variants).

The Human Genome Project has profoundly transformed the study of human biology, and accelerated the development and practice of medicine.

人類基因組計劃是人類史上最偉大及具意義的生物醫學研究項目之一。

1990年，來自不同國家及學科（包括生物學、物理學、化學及工程學）的科學家聚首一堂，開始這項需高度協作的計劃，為人體中所有DNA（即基因組）進行研究及測序。

數千名科學家經過十多年的不懈努力，於2003年完成了第一個人類基因組測序的藍圖。他們分析和結合了多名匿名志願者的DNA樣本，組成一個「人類參照基因組」。全球的科學家和醫生現在能以此作基準，進行比對並找出導致疾病的差異（即基因變異）。

人類基因組計劃改變了人類生物學的研究，並加速了醫學的發展和實踐。



## Building Critical Local Partnerships

Shortly after HKGI was established and core teams were recruited, it moved immediately to set up the infrastructure for implementing genomic medicine. HKGI worked closely with the Hospital Authority, The Chinese University of Hong Kong and The University of Hong Kong to set up three Partnering Centres (PCs) at the Hong Kong Children's Hospital, Prince of Wales Hospital and Queen Mary Hospital. While playing a pivotal role in HKGP's patient recruitment, the PCs enable HKGI to drive Hong Kong's genomic medicine development.

## 夥拍同儕 普及應用

在基因組中心成立及核心團隊履新後不久，基因組中心隨即建立推行基因組醫學的基礎設施及專業網絡，並與醫院管理局、香港大學及香港中文大學緊密合作，於香港兒童醫院、威爾斯親王醫院及瑪麗醫院設立三間夥伴中心。在招募基因組計劃參加者方面，夥伴中心發揮着舉足輕重的作用，同時支援基因組中心推動本港基因組醫學發展。

# Genomic Medicine

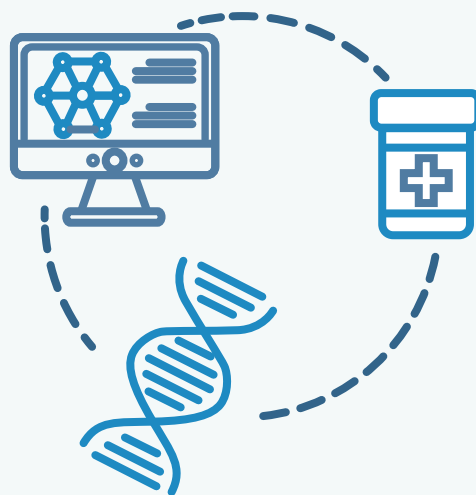
## 基因組醫學

Genomic medicine is the future of medicine. It is an interdisciplinary medical specialty involving the use of an individual's genomic information to inform, personalise and improve healthcare.

By understanding a person's complete set of DNA, i.e. genome, and how it can affect one's health, clinicians and scientists will be able to make more precise diagnoses and provide more tailor-made treatments to patients, instead of a one-size-fits-all approach. Genomic medicine may also be utilised in predicting disease risks and developing effective prevention plans.

基因組醫學是醫學的未來，是一門跨學科的醫學專業，主要運用個人的基因組訊息以改善醫療方案，實現個人化治療。

透過了解一個人的完整 DNA(即基因組)以及它如何影響健康，醫生和科學家將能給病人作出更精準的診斷及提供更多量身定制的治療方案，而非只用同一種方法醫治全部病人。基因組醫學也可以用於預測疾病風險，和制訂有效的預防計劃。



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

### Establishing Standards and Protocols for HKGP

To ensure patient recruitment is carried out according to the highest standards of medical ethics around the globe, HKGI produced a comprehensive Informed Consent Package detailing the consent processes for adult, youth and child participants, the consent forms used, the authorisations required, and the data protection precautions taken under the guidance of the ethics experts on its Ethics Advisory Committee and in consultation with relevant institutions of its PCs as well as public bodies, including the Office of the Privacy Commissioner for Personal Data. After several months of intensive preparation, deliberation and consultation, the Institutional Review Boards of the three PCs endorsed the Informed Consent Package and granted ethics approval to HKGP in July and August 2021, while approval of the Department of Health's Ethics Committee was obtained in January 2022. This was a very important milestone for the implementation of HKGP as it established high ethical standards and secured the support of its key stakeholders for this large-scale genome project.

In addition to the setting of high ethical standards, it is of utmost importance that HKGP is implemented according to a set of unified and internationally recognised protocols on clinical, consent, clinical data use and research to instil public confidence, generate stakeholders' support and build credibility in the outcomes of the project. Under the guidance of the distinguished scientists of its Scientific Advisory Committee, HKGI developed a set of internationally acclaimed research protocols on undiagnosed diseases and hereditary cancers to guide the implementation of HKGP.

### Genetic Counselling Takes Centre Stage

Genetic Counselling plays a pivotal role in building the necessary infrastructure for translating genetic and genomic research findings into clinical practices. As the genetic counsellors in Hong Kong come from a variety of professions with different backgrounds of education and training, there is a need to align the genetic counselling practices for HKGP to ensure that its sequencing analysis results are correctly interpreted and fully understood by participants and their family members. This facilitates patient decision and clinical application of sequencing findings for precise diagnosis and personalised treatment. It is also important to unify genetic counselling practices for HKGP in accordance with international benchmarks to ensure that participants' experience of the entire process will not be significantly different and their confidence in the sequencing findings and interpretation is enhanced.

To align genetic counselling practices and streamline workflow according to international benchmarks during the informed consent, pre-test counselling and post-test counselling processes of HKGP, two rounds of meetings/workshops with the three PCs were conducted, the first round being held in August and September 2021 and the second round in February 2022.

### 制訂規程 樹立公信

為確保按全球最高的醫學倫理標準招募病人，基因組中心制訂了全面的「知情同意資料冊」，詳細列明成人、年輕人及兒童參加者的參加流程、採用的同意書、需要授權的資料，以及在倫理諮詢委員會的專家指導下，經諮詢相關夥伴中心和公共機構（包括個人資料私隱專員公署）後採取的數據保護措施。經過數月的密集籌備、商議及諮詢，三間夥伴中心的研究倫理委員會分別於2021年7月及8月就基因組計劃知情同意書給予倫理准許，而衛生署倫理委員會亦於2022年1月發出相關准許。此乃基因組計劃籌備過程中非常重要的里程碑，不但為基因組中心樹立了最高的倫理標準，亦獲得主要持份者對此大型基因組計劃的支持。

除制訂最高的倫理標準外，最重要是在臨床、同意書、臨床數據使用和研究等方面，建立統一且獲國際認可的規程，藉以推行基因組計劃，樹立公眾信心，獲得持份者支持，增加計劃成果的公信力。依循科學諮詢委員會多名傑出科學家的指導，基因組中心就未能確診病症及與遺傳有關的癌症，制訂了一套獲國際認可的研究規程，從而指導基因組計劃的落實過程。

### 遺傳輔導 不可或缺

遺傳輔導在推動建立必要的專業網絡，以及將遺傳與基因組研究成果投入臨床應用方面，發揮着重要作用。由於本港的遺傳輔導員來自不同專業，他們的教育及培訓背景各不相同，因此在推行基因組計劃時，需要統一遺傳輔導實務，確保輔導員能夠向參加者及其家屬正確解釋測序分析結果，讓病人全面理解以作出臨床相關的決定，實現準確診斷及個人化治療。此外，根據國際標準而統一基因組計劃的遺傳輔導實務亦相當重要，此舉確保每位參加者於整個過程的體驗不會出現顯著差異，同時增強他們對測序結果和解讀的信心。

基因組中心與三間夥伴中心舉行了兩次會議／研討會，統一了遺傳輔導實務，以及簡化基因組計劃的知情同意書、測序前及測序後的流程，從而切合國際標準。首輪會議於2021年8月及9月舉行，第二輪則於2022年2月舉行。



## Providing State-of-the-art Sequencing Services

Being a large-scale sequencing project, HKGP needs to have the support of an effective and efficient whole genome sequencing service platform and system built according to internationally accredited laboratory standards. During the past year, such sequencing service support has been progressively developed with the award of contract to a qualified university research centre for the provision of sequencing services for the pilot phase of HKGP in May 2021 and the completion of the trial run of its service platform in October 2021. After validation of the sequencing workflow, the outsourced sequencing service platform was successfully launched in November 2021 with batches of bio-samples being dispatched continuously to the service provider for processing and sequencing.

The establishment of HKGI's state-of-the-art laboratory for the provision of in-house sequencing services was completed with its operations commenced in December 2021 after timely completion of fitting-out works, acquisition of the necessary laboratory equipment, installation of the sequencing platform, and setting up of the long-read sequencing system for advanced studies.

The whole genome sequencing process involves sample registration at the HKGI Laboratory, sample processing and sequencing at the outsourced service provider, sequencing alignment and variant calling with HKGI's bioinformatics pipelines, and variant annotation and interpretation by Scientific Officers of HKGI. Quality indicators have been developed and implemented for each of these steps to ensure that the processing, sequencing, and analysis of samples are performed to the highest quality and in compliance with relevant international standards.

## 頂尖測序服務 建立標準

基因組計劃是大規模的測序計劃，必須依循國際認可的實驗室標準，構建高效的全基因組測序平台和系統，作為計劃的強大後盾，以確保計劃順利推行。基因組中心於過去一年陸續開展測序服務，包括於2021年5月與合資格的大學研究中心合作，為基因組計劃先導階段提供測序服務，並於2021年10月完成服務平台的測試。測序流程獲確認後，外判測序服務平台於2021年11月成功推出，可以將生物樣本送交研究中心進行處理及測序。

此外，基因組中心的實驗室按時完成裝建工程，並配備先進設備和測序平台，亦建立了長序列測序系統來進行研究。實驗室於2021年12月正式投入運作，專責提供測序服務。

全基因組測序過程首先於基因組中心實驗室辦理樣本登記，隨後由服務供應商進行樣本處理及測序，再按基因組中心的生物信息管理流程進行序列比對及變異識別，並由中心的科學主任為相關變異加入註釋及詮釋。每個步驟均制訂及落實了質素指標，確保以最高標準進行樣本處理、測序及分析，並符合相關的國際標準。



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

### Establishing a Highly Effective and Scalable Bioinformatics Platform

A highly performant bioinformatics platform to support analysis and interpretation of whole genome sequencing data is vital to any large-scale genome project. During the past year, HKGI has successfully developed the necessary bioinformatics infrastructure and platform for supporting patient recruitment, operation of the PCs, and sequencing services of HKGP. The Clinical Frontend for HKGP teams to register patients, obtain their consent, collect relevant clinical information and bio-samples, and the Laboratory Information Management System (LIMS) for managing the production of raw next-generation sequencing data from the bio-samples, were rolled out in October 2021. Bioinformatics pipelines have also been developed to process the sequencing data and to annotate genomic variants with known disease associations. The Privacy Impact Assessment on the bioinformatics platform was completed by the appointed external security auditor in early 2022.

In addition, HKGI has developed flexible bioinformatics pipelines that can readily be plugged with various in-house developed or publicly available bioinformatics tools. They allow job resumption from failed steps without the need to start from the very beginning to save the execution time and cost of the sample runs. The pipelines are scalable to facilitate the potential increase in sample processing throughput in the future.

HKGI has also developed an in-house web-based analysis tool, the Bioinformatics Analysis Browser, for performing tertiary analyses on HKGP cases. This tool has seamlessly included the well-known knowledge bases developed by advanced overseas countries with comprehensive annotations for virtual gene panel analysis according to the phenotypes collected. It incorporates a prioritisation functionality on potential disease-causing variants and provides visualisation to enable users to assess the sequencing quality of variants. It has gone through a privacy impact assessment to ensure its secure application.

### 構建生物信息平台 高效運作

對任何大型基因組項目來說，若要取得成功，必須建有高效的生物信息平台，方可支援全基因組測序所涉及的大量數據分析及詮釋工作。為此，基因組中心於過去一年成功構建重點生物信息學基礎設施和平台，以支援基因組計劃在病人招募、夥伴中心運作，以及測序服務等多方面的工作。2021年10月，基因組中心率先推出臨床資訊管理平台，讓基因組計劃團隊可便捷地為病人登記、向他們徵詢知情同意，並記錄所收集的臨床資訊和生物樣本；中心同時建立了實驗室資訊管理平台，協助團隊有效管理以生物樣本進行新一代測序所得的原始數據。中心亦設計了生物信息管理流程，用作處理測序數據，以及標註與已知疾病相關的基因變異，並於2022年初邀請了獨立顧問為相關流程及平台進行全面的私隱影響評估。

此外，基因組中心制訂了靈活的生物信息管理流程，方便與各種內部開發或公用的生物信息學工具同時使用，並允許工序從步驟失敗中恢復運作，無需從頭開始，此舉可以節省樣本分析的執行時間和成本。有關流程亦採用可擴展性設計，應付將來樣本處理量可能增加的需要。

基因組中心亦開發了網絡分析工具「生物信息學分析瀏覽器」，以便對基因組計劃的個案進行高端解讀分析。此工具無縫對接其他先進國家或地區開發的知名知識庫，並可根據收集的遺傳表型對虛擬基因組合分析進行全面註釋；亦結合了潛在致病基因變異的優先排序功能，令用戶能透過視像化模式來評估基因變異的排序質素。分析瀏覽器已通過私隱影響評估，確保安全可靠。



## Personalised and Precision Medicine on the Horizon

With the core infrastructure, teams, and standards and protocols in place, HKGP had its soft launch in July 2021, and the first participant was recruited. The project was fully unveiled in October 2021 at the three PCs with the launch of the Clinical Frontend and the bioinformatics platform. Despite the subsequent hiatus in patient recruitment caused by the fifth wave of COVID-19 outbreak in early 2022, over 6,000 samples have been collected to date.

The importance of multi-disciplinary team meetings for the implementation of genome projects is well recognised around the world. It is important at all points: from the analysis and interpretation of whole genome sequencing results through diagnosis, clinical decision making and finalisation of treatment plans. After thorough discussions with the overseas experts and local clinicians, HKGI has finalised operational details of its multi-disciplinary team meetings with the roles of different team members, including clinical leads/referring clinicians, clinical geneticists, genetic counsellors, bioinformaticians, scientific officers and trainees, clearly defined. The first two multi-disciplinary team meetings of HKGP were held in May and June 2022 for exchange of views on the whole genome sequencing findings with respect to individual patients' clinical indications amongst HKGI and HKGP team members of the three PCs. Both meetings yielded fruitful results regarding the refinement of diagnosis and clinical treatment plans for individual patients to move towards the practice of personalised and precision medicine.

## 邁向個人化治療及精準醫療時代

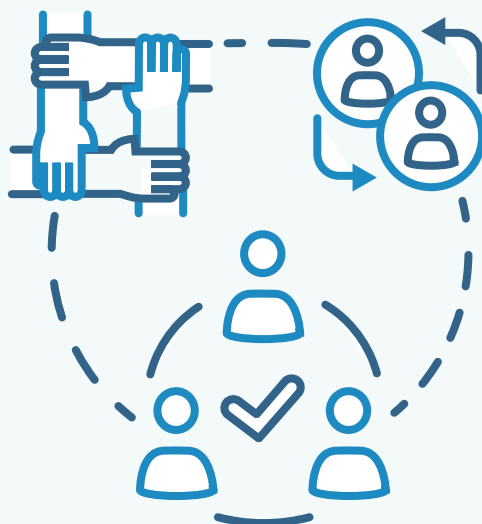
隨着核心基礎設施、團隊、標準及規程準備就緒，基因組計劃於2021年7月正式開展，並招募了首名參加者。臨床資訊管理平台及生物信息平台推出後，三間夥伴中心亦於2021年10月全面開展基因組計劃的相關工作。縱使在2022年初，2019冠狀病毒病爆發第五波疫情，導致病人招募暫停，基因組計劃迄今已收集超過6,000個樣本。

跨專業團隊會議對實施基因組計劃相當重要。無論是全基因組測序結果的分析和解讀，還是診斷、臨床決定以及確定治療方案，跨專業團隊會議對各個環節均舉足輕重。基因組中心與海外專家及本地醫生進行深入探討後，確定其跨專業團隊會議的運作細節，明確界定臨床醫生／轉介醫生、臨床遺傳學家、遺傳輔導員、生物信息學家、科學主任及受訓人員等不同團隊成員的角色。基因組計劃分別於2022年5月及6月舉行首次及第二次跨專業團隊會議，與三間夥伴中心的基因組計劃團隊成員就個別病人臨床情況及全基因組測序結果交流意見。兩次會議顯著改善個別病人的診斷和臨床治療方案，有效推動個人化治療和精準醫療。

## Multi-disciplinary Team 跨專業團隊

Multi-disciplinary Team (MDT) has an indispensable role to play in integrating genomics into clinical care. As the name suggests, a MDT is composed of experts from various disciplines, including clinicians, genetic counsellors, genome curators, laboratory professionals, bioinformaticians, and scientists. They discuss the whole genome sequencing results to make diagnoses and clinical decisions, and to formulate treatment plans for patients.

跨專業團隊在將基因組學融入臨床護理方面發揮着不可或缺的角色。顧名思義，跨專業團隊由來自不同領域的專家組成，包括醫生、遺傳輔導員、基因組數據分析員、實驗室專業人員、生物信息學家及科學家。他們透過深入探討全基因組測序結果，以作出診斷及臨床決策，並制訂病人的治療方案。



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

There were many successful diagnoses amongst the first batch of HKGP participants with undiagnosed disease or hereditary cancers. In these cases, the findings of the whole genome sequencing analysis helped end individual patients' diagnostic odysseys and their prolonged periods of medical investigation and testing, leading to the formulation of better and more precise treatment and care plans for them.

Patients and their family members were appreciative of the patient-centred services provided by HKGI which addressed their unmet needs. They were also grateful for HKGI's efforts and the strict measures it has put in place to safeguard the privacy and security of their information and genomic data.

### Addressing Everyday Diseases, Changing Lives

Preparations for launching the main phase of HKGP started in 2021-22. While the pilot phase focused on undiagnosed diseases and hereditary cancers, HKGI expanded the cohort of undiagnosed diseases and hereditary cancers and incorporated additional disease cohorts in genomics and precision health. These included the cohorts of aortic dissection, neuro-developmental disorders, childhood acute leukaemia, childhood solid tumours, acute myeloid leukaemia, and gynaecological sarcoma. Meetings with interested clinicians and their clinical research teams were held during the year to discuss the engagement of these potential disease cohorts. Through these discussions, clinical experts from each PC have put forward proposals for bringing their disease cohorts to participate in HKGP. Findings of these collaborative research projects will assist in medical management and treatment, as well as disease prediction and prevention.

### Looking Forward to Our Bright Future Together

The solid foundation built for integrating genomic medicine into clinical care for personalised and precision medicine is set to yield fruitful results. In addition to implementing the main phase of HKGP and further expanding the genomic database of the local population, HKGI will initiate more in-depth studies, e.g., multi-omics studies, in collaboration with relevant stakeholders, incorporating utility and outcome evaluations to promote genomic medicine. To expand the reach and impact of HKGP, new channels will be opened for boosting patient recruitment.

縱觀首批患有未能確診病症或與遺傳有關癌症的參加者，許多個案皆成功獲得診斷，全基因組測序分析結果讓他們可結束曲折漫長的診斷過程，省卻長時間的醫學診查及檢測，亦能為他們制訂更適切、更精準的治療及護理方案。

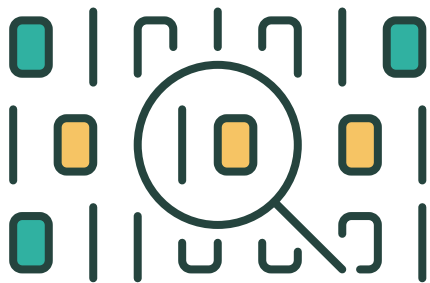
基因組中心的服務以病人為本，致力解決病人的需要，獲得病人及家屬的認同。他們感謝基因組中心對保障病人資料和基因組數據私隱及安全而付出的努力，以及所採取的嚴謹措施。

### 應對常見疾病 擴展計劃

2021-22年期間，基因組計劃主階段的準備工作如火如荼。除了先導階段涵蓋的未能確診病症和與遺傳有關的癌症，基因組中心於主階段擴展了這兩個群組，並擴大範疇至與基因組學和精準醫學有關的疾病群組，包括主動脈撕裂、神經發展障礙、兒童急性淋巴白血病、兒童腫瘤、急性骨髓性白血病及婦科肉瘤。基因組中心亦於年內與相關範疇的醫生及臨床研究團隊舉行會議，探討這些潛在疾病群組的參與性質。經過多次討論，各夥伴中心的臨床專家亦就相關疾病群組納入基因組計劃提出建議，這些研究成果將有助醫療管理及治療，同時加強疾病預測和防控。

### 精誠合作 放眼未來

基因組醫學與臨床護理互相結合，實屬推動個人化治療和精準醫療的重要基石，並會帶來豐碩的成果。除了推行基因組計劃的主階段及進一步擴展本地人口的基因組數據庫，基因組中心亦會與相關持份者合作，展開多組學等更深入的研究，結合實用層面及研究成果，以推動基因組醫學發展，並會繼續開拓新平台招募病人，從而擴大基因組計劃的涵蓋範圍，提升影響力。



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





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

Collecting local genomic data and establishing a local biobank are the first steps in the development of genomic and personalised medicine. To truly realise its full potential, we must strategically advance genomic science research and capitalise on local and international research collaboration. This will offer deeper insights into disease biology and biomedical science, allowing us to develop new genomic technologies and multi-omics studies, broaden the diversity of research outcomes, implement pharmacogenomics, and ultimately produce greater benefits for patients.

### Multi-omics Playing a Broader Role in Personalised Medicine

Soon after HKGP was launched, HKGI began analysing genomic data to obtain a better understanding of predispositions to diseases prevalent in Hong Kong. Its initial focus was on diseases that are endemic to the region but not well studied, such as Thalassaemia. We fully expect that, as more data is collected, the findings will improve our ability to produce more accurate diagnoses of certain genetic diseases. This will enable us to perform better disease prediction and treatment, with greater capacity to improve the health and well-being of the Hong Kong people.

### Developing a Greater Ability to Analyse Genes

To supplement its research capabilities, HKGI tapped into the data and knowledge bases of leading genome projects around the world, such as those developed by Genomics England, the Australian Genomics Health Alliance, and the National Institutes of Health of the United States. HKGI's capability and capacity for performing tertiary analysis have been enhanced by the successful development of the in-house web-based analysis tool, Bioinformatics Analysis Browser in 2021-22. This tool integrates different knowledge bases with comprehensive annotations for virtual gene panel analysis. Its functions will be expanded on an ongoing basis by introducing more new feature enhancements, including linking it to other useful databases such as the Genome Aggregation Database (gnomAD).

### Establishing International Research Networks

Many of the genetic and genomic studies are currently conducted locally and independently by individual clinicians, researchers, or universities. They have largely been driven by the interest and expertise of the respective staff, local needs, or laboratory initiatives. As a result of this, disease-focused research networks in Hong Kong have been underdeveloped.

收集本地基因組數據及建立本地生物樣本庫是發展基因組醫學及個人化治療的第一步。為充分發揮其潛力，基因組中心須有策略地促進基因組科學研究，深化本地與國際間的研究合作，以便對疾病生物學和生物醫學有更深入的了解，從而促進嶄新的基因組技術發展和多組學研究，擴大研究成果的多元性，實踐藥理基因組學的應用，為病人帶來更大裨益。

### 推動多組學應用 實踐個人化治療

基因組計劃推出後不久，基因組中心隨即開始分析基因組數據，務求深入了解本港流行疾病的趨勢。分析工作初期集中於區內流行但研究有限的疾病，例如地中海貧血。隨着收集的數據增多，分析結果將有助基因組中心對某些遺傳病作出更準確的診斷，從而達到有效的疾病預測和治療，強化本地醫療實力，讓市民享有健康福樂。

### 基因分析 發展實力

基因組中心與世界各地主要的基因組計劃均有合作，如Genomics England、Australian Genomics Health Alliance及美國國立衛生研究院開展的計劃，透過運用其數據庫和知識庫來提升研究能力。隨着團隊於2021-22年度成功開發網絡分析工具「生物信息學分析瀏覽器」，大大提升了基因組中心解讀測序結果的實力及能力。瀏覽器可融合多個具有綜合註釋的知識庫，用於虛擬基因組合分析，亦可引入更多新的強化功能，包括連接至其他數據庫，如基因組聚合數據庫 (gnomAD)。

### 建立國際網絡 啟發科研

目前，許多遺傳及基因組研究由個別醫生、研究人員或大學於本港獨立進行，主要取決於學術興趣及專業知識、本地需要或實驗室的計劃。因此，以疾病主導的研究網絡於香港仍有龐大的發展空間。

To this end, HKGI is establishing disease-focused local and international research networks under specific disease themes, partnering with academic institutes to conduct relevant research studies. In 2021-22, we initiated a series of research collaborations with local universities. This included working with The Hong Kong University of Science and Technology to study Alzheimer's disease using genomics; The Chinese University of Hong Kong to study diabetes; The University of Hong Kong to study leukaemia; and the Hong Kong Children's Hospital to study children's cancers.

有見及此，基因組中心以特定疾病為主題，建立本地及國際研究網絡，並與學術機構合作進行相關研究。中心於2021-22年與本地大學開展一系列研究合作，包括與香港科技大學合作，透過基因組學研究阿爾茨海默症、與香港中文大學合作研究糖尿病、與香港大學合作研究白血病，以及與香港兒童醫院合作研究兒童癌症。

## Genetics vs Genomics

### 遺傳學vs基因組學

While these two words look alike, they are not the same.

Genetics focuses on genes and heredity. It involves the study of specific and limited numbers of genes, or parts of the genes. It focuses on how genes are passed down from one generation to another, and how the differences in genes affect people.

Genomics looks at all the DNA of a person, i.e. the genome. Genome is like the body's instruction manual and contains all the information needed for a person to develop and grow. Studying the genome may help clinicians and scientists understand how genes interact with each other and with the environment and, how diseases are caused and be treated.

儘管這兩個詞彙看起來很相似，但涉及的範疇其實截然不同。

遺傳學專注於基因和遺傳，並針對特定及有限數量、或部分基因作研究，主要探討基因如何從一代傳給下一代，以及基因的差異如何影響人類。

基因組學則是研究一個人的所有DNA，即全部基因。

基因組就像人體的說明書，包含一個人所有發展和成長所需的訊息。研究基因組可幫助醫生和科學家了解基因與基因之間，和基因與環境之間如何互相影響，以及疾病的成因和治療方法。



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

### Keeping Local Standards Up to International Best Practices

HKGI has established global connections to keep the local industry abreast of the latest developments. The *Journal of Translational Genetics and Genomics* has invited our Chief Scientific Officer and some of his team members to be the chief journal editors for a special issue on genomics and precision health. This is an excellent way for HKGI to maintain an ongoing dialogue with investigators around the world, to exchange expertise and share it with local practitioners. These exchanges have promoted the development of genomics and precision health in Hong Kong. They will provide HKGI with more insights into the successful cases of other countries and regions that can benefit the ongoing work of HKGP.

In its first year of operation, HKGI had started to share its experience of launching HKGP with its overseas counterparts by publishing papers in well-recognised international journals. After conducting a literature review on similar international genomic projects, formulating main themes, and providing manuscript writing training to staff, HKGI succeeded in publishing its first research paper titled "Potentials and Challenges of Launching the Pilot Phase of Hong Kong Genome Project" in the *Journal of Translational Genetics and Genomics* in June 2022. Subsequent to the publication of this paper, HKGI staff were invited by Frontiers to submit a manuscript as expert contributors for the research topic "Equality, Diversity and Inclusive Research for Diverse Rare Disease Communities". This manuscript was then submitted to *Frontiers in Public Health* in August 2022 under the title of "Rare disease emerging as a global public health priority". A third paper on HKGP will be submitted in November 2022. Meanwhile, our Bioinformatics team also submitted a paper to share a tool with novel methodology for detecting structural variation in a high-performance manner.

### 確立標準 接軌國際

基因組中心致力建立國際網絡，以便本地業界緊貼行業的最新發展。中心的首席科學總監及科學團隊部分成員獲 *Journal of Translational Genetics and Genomics* 邀請，擔任其基因組學和精準醫療特刊的主編，讓基因組中心與世界各地的研究人員能保持緊密聯繫，並與本地專家交流和分享專業知識，推動香港基因組學及精準醫療的發展。基因組中心亦可更深入了解其他國家和地區的成功個案，促進基因組計劃的發展。

在營運的首年，基因組中心已積極在知名國際期刊上發表論文，藉此與海外同業分享推行基因組計劃的經驗。機構參考國際上其他相類似基因組計劃的文獻，並在確立主題和為同事提供寫作培訓後，於2022年6月成功在 *Journal of Translational Genetics and Genomics* 發表首篇研究論文——《推行香港基因組計劃先導階段的潛力與挑戰》(Potentials and Challenges of Launching the Pilot Phase of Hong Kong Genome Project)。其後，基因組中心獲Frontiers邀請以專家撰稿人身份就專題《罕見病社群的平等、多元性及包容性研究》(Equality, Diversity and Inclusive Research for Diverse Rare Disease Communities) 投稿，此論文隨後以《罕見病成為全球公共衛生優先議題》(Rare disease emerging as a global public health priority) 為題，於2022年8月呈交至 *Frontiers in Public Health*。而第三篇關於基因組計劃的論文則於2022年11月投稿。生物信息學團隊亦有發表論文，分享以高效能方式檢測結構變化的嶄新平台。

# Whole Genome Sequencing

## 全基因組測序

The “whole genome sequencing” technology enables us to understand more about the human genome. It involves three main steps:

### 1. Sequencing

This is the process of reading over 3 billion DNA bases in the human body one by one.

### 2. Data processing

This involves comparing the genome sequence with the “Human Reference Genome”, and detecting the differences which are known as “genetic variants”. There are millions of genetic variants in our body, most of which are not disease-causing.

### 3. Data Analysis

Data Analysis is the most complex and time-consuming step. It involves the identification of potential disease-causing variants, also called pathogenic variants, among millions of genetic variants. To reach a genetic diagnosis, it requires comprehensive analysis and in-depth research.

「全基因組測序」技術有助了解每人的基因組，主要包括三個步驟：

#### 1. 測序

測序是把人體內超過30億個DNA代碼逐一讀取，然後全部排列出來。

#### 2. 數據處理

數據處理是將基因組測序結果與「人類參照基因組」比對，找出基因序列上的不同，這些不同稱為「基因變異」。每人的身體均有數百萬個基因變異，大部分基因變異是不會致病的。

#### 3. 數據分析

數據分析是最複雜和耗時的步驟，涉及在數以百萬計的基因變異中，找出有機會導致疾病的基因變異。要作出遺傳病診斷，需要進行詳盡分析和深入研究。





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

### Looking Forward to More Collaborations

In the coming few years, HKGI will seek to accelerate genomic research by establishing a database and platform for new genomics technology and multi-omics studies. We will also improve analyses and interpretation of whole genome sequencing data by developing functional assays to characterise, annotate and interpret genes and variants. We will develop new genomic technologies, such as long-read sequencing, single-cell sequencing technologies as well as transcriptomics and epigenomics analyses, for clinical implementation. In addition to this, we will look for ways to engage industry partners to translate research findings into clinical use.

HKGI will also foster local and international genomic research collaboration by providing a rich database and flexible platform for researchers' exploration and discovery. We will provide support to researchers to curate the most appropriate dataset for individual research requests, ensuring the reliability and security of infrastructure, and facilitating innovative research algorithms to be run on the platform. Moreover, we will continue to collaborate with local and international academic institutes to conduct targeted research studies with specific disease themes.

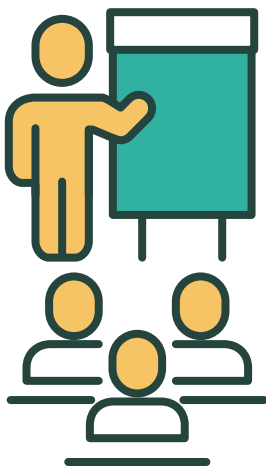
### 展望未來 共享成果

未來數年，基因組中心將為嶄新的基因組學技術和多組學研究建立數據庫和平台，致力加快基因組學研究；並將透過開發功能性分析，從而歸納、註釋及詮釋基因和基因變異，優化全基因組測序的數據分析和解讀；同時研發可供臨床應用的嶄新基因組技術，例如長序列測序技術、單細胞測序技術，以及轉錄組學和表觀遺傳學分析等。此外，基因組中心力爭與相關行業合作，務求將研究成果轉化至臨床應用。

基因組中心亦會提供完善的數據庫和靈活的平台，讓研究人員探索及研究當中的數據，促進本地與國際基因組研究的合作；機構亦會支援個別研究項目，協助研究人員整合最適合的數據組合，確保有關基礎設施安全可靠，並推動研發能在平台上應用的創新研究運算法。基因組中心將繼續與本地和國際學術機構合作，進行以特定疾病為主題的研究。



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



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

To succeed in our important mission, nurturing talents is an absolute must. The complexity and magnitude of genomic medicine necessitate a variety of skilled and competent professionals to maximise potential. In addition to scientists, clinical geneticists, genetic counsellors, and bioinformaticians, HKGI is in great need of genome curators, laboratory professionals, medical technologists, researchers, clinicians, nurses, and many others. Together, this dream team can make genomic discoveries, develop technologies and translate research findings into clinical applications.

### An Urgent Undertaking

While HKGI has rallied Hong Kong's leading genetic and genomic professionals to join HKGP, the genomic talent pool remains small. For this reason, one of HKGI's strategic foci is to nurture talents in genomic medicine. HKGI is committed to building a talent pool by enhancing the genetic and genomic knowledge of existing clinicians, nurses, and allied health professionals; promoting professional development with professional bodies; and attracting medical and nursing students to join the industry. It seeks to realise the Steering Committee's vision for HKGP, to "play a catalytic role in enhancing the genetic and genomic knowledge of healthcare professionals and attracting both local and international talent."

### Professional Development is the Foundation

To attain the strategic goal of enhancing genetic and genomic knowledge and professional development, HKGI implemented initiatives in 2021-22 to support continuing professional development programmes in genetics and genomics for clinicians, nurses, and allied health professionals, including genetic counsellors and bioinformaticians.

In the field of genetic counselling, many frontline medical practitioners providing genetic counselling services in research-funded programmes are currently trained on the job with or without certified qualifications. There is no accreditation body or programme in Hong Kong that certifies or registers locally or internationally trained genetic counsellors, nor is there

為了實現基因組中心的重要使命，培育人才是首要任務。基因組醫學博大精深，其複雜程度和規模之大，需要不同範疇的專家學者群策群力，方可發揮最大潛力。因此，推動香港的基因組醫學發展，除了需要科學家、臨床遺傳學家、遺傳輔導員及生物信息學家，基因組數據分析員、實驗室專業人員、醫務化驗師、研究員、醫生和護士，以及其他相關範疇的人才同樣不可或缺。這支陣容強大的夢之隊，定可為基因組學研究帶來突破、開發嶄新技術，並將研究成果轉化為臨床應用。

### 廣納人才 加強培訓

縱使基因組中心已吸納香港頂尖的遺傳學和基因組學專才加入基因組計劃，機構仍需招攬更多人才。有見及此，基因組中心的策略重點之一是培育基因組醫學人才，並致力加強現職醫生、護士及專職醫護人員的遺傳和基因組知識；與相關團體合作推動專業發展；並且吸引醫療和護理專業的學生加入，藉此擴大人才庫，實現督導委員會對基因組計劃的願景——促進醫療人員的遺傳和基因組知識，以及招攬本地和國際人才。

### 持續進修 發展專業

為實現強化遺傳學和基因組學知識及專業發展的策略目標，基因組中心於2021-22年度實施了一系列措施，開展遺傳學和基因組學方面的專業發展計劃，繼續支援醫生、護士和專職醫護人員的發展，當中包括遺傳輔導員及生物信息學家。

在遺傳輔導方面，不少於研究資助計劃中提供遺傳輔導服務的前線醫療人員均接受在職培訓，惟卻未有取得認證資格，皆因香港目前未有認可的機構或計劃向本地或國際培訓的遺傳輔導員頒發證書，亦未有專業團體推動行業倫理守則和專業標準。自成立以來，

a professional body to foster ethical conduct and professional excellence. Since its inception, HKGI has recruited a multi-disciplinary team of genetic counsellors, and trained them both in-house and through collaboration with the PCs to align practices. Preparatory work for setting up the Hong Kong Genetic Counselling Practice Consortium was completed in 2021-22 with formulation of a detailed proposal setting out the objectives of the Consortium, membership composition and work plan for the next two years to address the needs, challenges, and aspirations of developing the genetic counselling profession in Hong Kong. With the endorsement of the proposal by the HKGI Board and the HKSAR Government, we are in the process of discussing with relevant experts and stakeholders to create a scope of practice, establish a robust code of ethics, draft and implement a model of practice, and develop an accreditation system for the profession.

To promote the development of a professional network amongst bioinformaticians in Hong Kong, a collaborative communication network has been established for bioinformaticians at PCs and local universities with meetings held in 2021-22 to discuss ways to improve the data collection and sample analysing processes of HKGP. Tools were developed to facilitate regular sharing of knowledge and experience at the Hong Kong Children's Hospital and with bioinformaticians from the Department of Computer Science of The University of Hong Kong.

During the year, HKGI also started engaging staff of PCs, including clinical leads, referring clinicians, clinical geneticists, genetic counsellors, bioinformaticians, scientific officers and trainees, in multi-disciplinary team meetings of HKGP to enhance their knowledge and experience of applying genetics and genomics in their professional practices. Groundwork had also been laid to set up a network of genomics champions in different clinical specialties of public hospitals and engage them in promoting genomic education among colleagues of their respective clinical specialties.

基因組中心已成功招募一支跨專業遺傳輔導員團隊，並透過內部以及與夥伴中心合作提供培訓，從而統一實務層面。成立香港遺傳輔導專業發展聯席的籌備工作已於2021-22年完成，其中包括草擬組織宗旨、成員架構及未來兩年工作計劃的詳細建議，旨在應對社會需要及挑戰，實現香港發展遺傳輔導行業的願景。建議方案已獲得基因組中心董事局及香港特區政府的支持，而中心正與相關專家及持份者開展討論，以制訂實務範圍、建立完善的倫理守則、起草及實施實務模式，並且為行業制訂認證制度。

為促進香港生物信息學家之間的專業網絡發展，基因組中心為夥伴中心及本地大學的生物信息學家建立協作網絡，並於2021-22年舉行多次會議，討論如何改善基因組計劃數據收集及樣本分析的流程，包括開發相關工具，以便與香港兒童醫院及香港大學計算機科學系的生物信息學家定期交流知識和經驗。

過去一年，基因組中心開始鼓勵夥伴中心的職員，包括臨床醫生、轉介醫生、臨床遺傳學家、遺傳輔導員、生物信息學家、科學主任及實習醫護人員，參與基因組計劃的跨專業團隊會議，加強他們將遺傳學和基因組學應用至實務層面的知識和經驗。基因組中心亦於公立醫院各個臨床專科建立了基因組學倡議網絡，透過傑出人員及團隊的實務分享，樹立楷模，推廣基因組學的教育。



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

## Genetic Counsellors 遺傳輔導員

Genetic counselling refers to the process of helping patients and their families understand and adapt to the medical, psychological, and familial implications of genetic contributions to disease.

Being the bridge of communication between doctors and patients, genetic counsellors play a pivotal role in translating genomics into clinical practices. With training in genomics and psychology, they make sense of complex clinical and genomic data, explain to patients and their families how their health is affected, and what options they may have, in addition to offering emotional support and counselling.

遺傳輔導是指幫助病人及其家屬了解及適應由遺傳導致的疾病對其醫療、心理和家庭的影響。

作為醫生與病人之間的溝通橋樑，遺傳輔導員在將基因組學轉化為臨床實踐方面發揮着關鍵作用。透過基因組學和心理學方面的培訓，遺傳輔導員能理解複雜的臨床和基因組數據，向病人及其家屬解釋他們的健康如何受到影響，以及提供可選擇的方案，並提供情緒支援和輔導。



# Bioinformaticians

## 生物信息學家

Bioinformatics is about using computer technology to collect, store, analyse, annotate and share genetic and genomic data and information.

Bioinformaticians are specialists in computer science, statistics and biology, who are familiar with programming and coding, and turning them into effective analysis tools. They are trained to develop computational tools and analyse large data sets such as raw genomic data for clinical and research purposes.

生物信息學利用電腦技術去收集、儲存、分析、註釋和分享遺傳學和基因組數據及訊息。

生物信息學家是計算機科學、統計學和生物學方面的專家，不但熟悉編程和編碼，而且能將其轉化為有效的分析工具。他們經過培訓，可開發運算工具和分析大量基因數據，用於臨床和研究。



### Taking the First Steps with Professional Bodies

In addition to establishing the Hong Kong Genetic Counselling Practice Consortium, HKGI also enhanced its collaborative relationship with the Hong Kong Academy of Medicine and the Hong Kong Academy of Nursing in 2021-22 to promote the incorporation of genomic science and genomic medicine into their research, curriculum and training. The two institutions have now placed genomics at the top of their training agendas to cultivate talents and promote their professional growth. The constituent colleges of the Academy of Medicine are currently exploring the development of a training curriculum in genomics and exploring adding genomic medicine as a sub-specialty, following the examples of paediatrics and pathology. HKGI also collaborated with local universities, professional bodies, and international organisations during the year to keep the local sector abreast of the most recent breakthroughs and achievements in genomic medicine.

### 專業團體 攜手合作

除成立香港遺傳輔導專業發展聯席外，基因組中心亦於2021-22年加強與香港醫學專科學院及香港護理專科學院的合作，推動兩者將基因組科學及基因組醫學納入其研究、課程及培訓。目前，兩所院校已將基因組學列為培訓計劃的首要任務，從而培育人才並提倡專業發展。此外，香港醫學專科學院各分科學院正探索制訂基因組學的培訓課程，並仿效兒科和病理學專業，將基因組醫學納入作為附屬專科。基因組中心在過去一年積極與本地大學、專業團體及國際組織合作，讓本地業界了解基因組醫學的最新突破和成就。

## Nurture Talents in Genomic Medicine

### 培育基因組醫學人才

#### Looking Forward with Passion and a Strong Vision

The demand for genomics specialists is expected to increase as HKGI starts to implement the main phase of HKGP and advances the development and application of genomic medicine. In addition to continuing training all relevant professionals, bolstering the talent pipeline, and taking forward our work with the genetic counselling and bioinformatics professions, HKGI will seek to incorporate experiential learning into continuing education programmes.

To improve the genetic and genomic knowledge among tertiary students and encourage them to pursue careers in the field, HKGI has initiated discussions with local universities in 2021-22 to develop internship and enrichment programmes for undergraduate and postgraduate students in genomic medicine-related studies. The first internship programme of HKGI was implemented in the summer of 2022 with very encouraging positive feedback from both participating students and their mentors.

#### 滿懷熱誠 實現願景

隨着基因組計劃的主階段開始實施，加快基因組醫學的發展及應用，基因組中心預期對基因組學專家將有更大需求。中心不僅會持續培訓所有相關專業人員，加強人才儲備及促進遺傳輔導和生物信息學專業的工作，亦會將體驗式學習納入持續進修計劃。

為增進大學生在遺傳學和基因組學方面的知識，基因組中心於2021-22年與本地大學合作，為修讀基因組醫學相關課程的大學本科生和研究生開辦實習計劃和進修課程，希望鼓勵他們投身其中。機構已於2022年夏季開展首個實習計劃，參與的學生及他們導師均給予正面的意見。

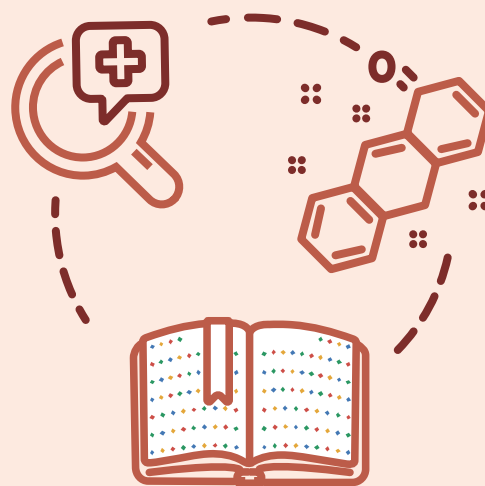
## Genome Curators 基因組數據分析員

The process of genome curation involves multiple steps that integrate large amount of genomic data with experimental evidence and literature across databases.

Genome curators are specialists who interpret genetic and genomic data to find out how the genetics is related to disease or drug response. They play a crucial role in identifying the disease-causing variants of the patients. With the study of genomics evolving rapidly, genome curators have to keep up to date with tremendous amount of literature and advancement in the field in order to identify useful information for interpreting the genome sequences.

基因組數據分析的過程涉及多個步驟，需將大量基因組數據與不同數據庫中的實驗證據及文獻互相整合。

基因組數據分析員是詮釋遺傳學和基因組數據的專家，能找出遺傳學與疾病或藥物反應的關係，在識別病人致病的基因變異上發揮着關鍵作用。隨着基因組學研究的迅速發展，基因組數據分析員必須追蹤該領域的大量文獻及了解領域的發展，以識別用於詮釋基因組序列的有用訊息。





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



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

As genomic medicine is a relatively new field, with a potentially profound impact for the healthcare system in Hong Kong, a core mission of HKGI is to instil a positive understanding among our stakeholders and the public. This includes in-depth explanation of different concepts relevant to genomics, such as what a genome is, how genomic science works, and the clinical applications of genomic medicine. In this regard, we seek to enhance genomic literacy and engagement, allowing the public to use genomics to make better personal and family health decisions, participate in policy discussions, and help propel the development of genomic medicine in Hong Kong.

Throughout the year, HKGI implemented different initiatives in raising public awareness and accomplished various milestones on the communication and publicity front. A wide range of authoritative and user-friendly information and publications on genomic medicine and HKGP were developed and shared with PCs for patient recruitment and on online channels. We also hosted a number of events to engage targeted stakeholders to deepen their understanding of genomic medicine and its benefits.

### Understanding Patients' Needs through Focus Groups

Tremendous efforts have been made in preparing for the launch of HKGP. Prior to developing the materials for the patient information package, we conducted a few focus group sessions in early 2021 to ensure that the content we create and the way we communicate with patients would be effective. Patients with undiagnosed diseases and hereditary cancers, families of patients, representatives of patient groups and many others were invited to offer their views. The ultimate goal was to help patients, medical and healthcare professionals, and the public understand the significance and benefits of genomics and genomic medicine, as well as to address questions, misperceptions and concerns. The sessions proved highly beneficial for public education and awareness-building, as reflected in the positive feedback received.

基因組醫學是嶄新領域，潛力巨大，足以對香港的醫療體系產生深遠的影響。基因組中心的核心使命是幫助相關持份者及公眾正確理解基因組醫學，包括深入解釋有關基因組學的各種概念，例如基因組是什麼、基因組科學的原理，以及基因組醫學的臨床應用。為此，基因組中心希望加強公眾對基因組學的認識及參與，令他們能夠為個人及家庭健康作出更佳決定，亦鼓勵他們加入政策討論，繼而推動香港基因組醫學的發展。

過去一年，基因組中心透過不同形式和宣傳渠道加強公眾的認知，在傳訊及推廣工作亦達到多項里程碑，其中包括編製了一系列既具權威性又顯淺易明的公眾教育刊物，深入淺出地講解基因組醫學的基本知識及簡介基因組計劃的背景目的，提供予夥伴中心作招募病人之用，並在網上分享了相關內容。機構亦舉辦了不同的活動，加深相關持份者對基因組醫學及其效益的了解。

### 接觸病人 了解所需

早在基因組計劃推出前，基因組中心已全力籌備，包括於2021年初舉辦了多次聚焦小組會議，確保病人資料冊的內容及與病人溝通的方式能發揮其成效。患有未能確診病症的病人、與遺傳有關的癌症病人、病人家屬、病人組織代表及其他相關人士均獲邀出席並發表意見，最終目標是協助病人、醫護人員和公眾了解基因組學及基因組醫學的重要性及效益，釋除他們的誤解和疑慮。與會人士亦給予了正面的回應，足以證明這些討論有助促進公眾教育及提高市民的認知。



## Strengthening Awareness and Knowledge with Corporate Publications

To facilitate participant recruitment, a series of publicity materials catered to the needs and interests of various stakeholders were produced. Apart from developing the HKGP information package comprising leaflets, booklets, posters and souvenir items in mid-2021, we customised different versions for adults and children to clearly explain the Project details, including the eligibility and rights of the participants, while answering common questions. These communications materials also play a crucial role in bringing our work closer to the general public.

The *HKGI Strategic Plan 2022-25*, which outlines our strategic foci, strategies, priorities and key actions for the coming three years, was developed as a key communications tool to promote understanding of the purposes and objectives of HKGI and HKGP. The printed version was distributed to various specific stakeholders such as the Department of Health, Hospital Authority, PCs, professional bodies, public libraries, and the Panel on Health Services of the Legislative Council. An electronic flipbook was also created and published on our website for ease of access by key stakeholders and members of the public.



## 出版刊物 宣傳教育

基因組中心編製了一系列切合不同持份者需要的宣傳刊物，協助招募基因組計劃的參加者。2021年年中，中心製作了包含單張、小冊子、海報及紀念品的資料冊，亦分別為成人及兒童設計不同版本的資料，清楚說明計劃的詳情，包括參加者資格及須知，並解答常見問題，令公眾更了解基因組中心的工作。

此外，基因組中心亦制訂了《2022-25年策略計劃》，概述未來三年的策略重點、具體策略、工作優次及主要方向，讓公眾了解基因組中心和基因組計劃的宗旨和目標。策略計劃乃傳達機構發展方向的重要媒介，其印刷本已分發予各相關持份者，如衛生署、醫院管理局、夥伴中心、專業團體、公共圖書館及立法會衛生事務委員會等，而電子書版本則已刊登於基因組中心的網站，供主要持份者及公眾人士查閱。



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

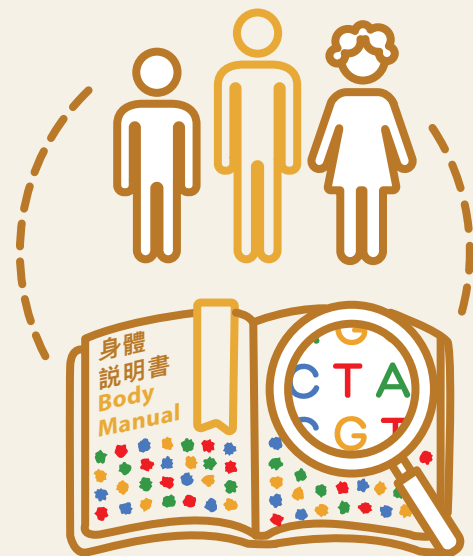
# Genome 基因組

“Genome” refers to the entire set of genetic materials in a living thing. For every human being, the genome includes over 20,000 genes. Each of us has our own unique genome, which can be thought of as an operation manual for our body. It provides instructions that determine the physical characteristics of our body such as skin colour, height and potential risks of developing different diseases.

If we compare the genomes of two people who are not related by blood, 99.9% of their genomes will be the same and only 0.1% of them will be different. However small this 0.1% might look like, it is significant enough to represent millions of differences in the two DNA sequences, accounting for each person’s unique physical characteristics.

「基因組」是指生物體內的「所有遺傳物質」，而人類的基因組內有超過兩萬個基因。每人的基因組都是獨一無二的，猶如「身體的說明書」，決定了每人擁有的不同特徵，例如膚色、高度及患上不同疾病的潛在風險等等。

若比較兩位沒有血緣關係的人，兩者的基因組有99.9%相同，不同之處只佔0.1%；然而，這看似微細的0.1%，卻已包含數以百萬計基因排序上的不同，足以為每人造就不同的身體特徵及功能結構。



### Bolstering Communications with Multimedia and Web Presence

To expand the reach of our communications and highlight the significance of HKGI’s work, a number of videos were developed and released in Q3 2021, which were then circulated to PCs for patient recruitment. These include over 10 patient case videos, as well as patient recruitment videos and animations tailored for both adults and children.

### 善用網絡 深入社區

為接觸廣大市民，宣傳基因組中心工作的重要性和重要性，中心於2021年第三季製作及發布了超過10條影片，協助夥伴中心招募病人，其中包括病人分享，以及專為招募成人和兒童參加者而設計之影片及動畫。

The HKGI website serves as an imperative and informative tool for public education and engagement, as it offers user-friendly information about HKGI, HKGP, patient recruitment, genomics and genomic medicine, as well as other genome projects worldwide. The corporate website was augmented with lively graphics and a compendium of videos and project materials produced for targeted stakeholders and the public to learn more about the subject. Since the launch of the Traditional Chinese website in mid-2021, and subsequently the English and Simplified Chinese versions, the website has been regularly updated with latest news on our work, project details, videos, events, educational materials and corporate publications.

基因組中心的網站是公眾教育和互動的重要平台，提供了有關基因組中心、基因組計劃、病人招募、基因組學、基因組醫學乃至世界各地基因組計劃的資料。網站載有生動的圖像，以及為目標持份者和公眾製作的影片和計劃資料，有助加深對基因組醫學的認識。基因組中心於2021年年中率先推出繁體中文版本的網站，隨後陸續推出英文及簡體中文版本，並定期更新以提供最新消息、基因組計劃詳情、影片、活動、教育資訊和機構刊物等。

## > 3 billion 30億個 DNA Bases 代碼

Deoxyribonucleic acid, or DNA in short, is a substance that exists in our human cells. Its structure consists of two long spiral chains with four types of bases, which can be thought of like English letters. They are adenine (A), thymine (T), cytosine (C), and guanine (G), the basic units of DNA.

There are over 3 billion DNA bases in the human genome. The sequence, or order, of these DNA bases determines the genetic information and instructions for our body's physical features and functions.

This is similar to how we communicate in English by using the 26 letters in the alphabet to form words and sentences to convey different meanings.

DNA是Deoxyribonucleic Acid (去氧核糖核酸)的縮寫，存在於人體細胞之中。它的結構由兩螺旋長鏈組成，長鏈上有四種像英文字母一樣的「代碼」，分別是A (adenine)、T (thymine)、C (cytosine)和G (guanine)。

人體內的基因組有超過30億個DNA代碼，這些代碼的序列或順序，決定人體身體特徵和功能上的遺傳指令和訊息。

一如在英語中，我們運用26個英文字母組成不同詞彙及句子，以表達各種意思。



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

# > 20,000 個 Genes 基因

A gene is a segment of DNA that serves as the basic unit of instruction for building our body's observable traits and functions.

The DNA bases are linked together in a chain to form a DNA strand. Only certain sections of the chain contain genetic instructions to build our body's observable traits and functions.

By analogy, a gene is like a complete sentence or paragraph in the English language which conveys particular meaning.

基因是遺傳指令的基本單位，影響着每個人的身體結構和功能。

四種DNA代碼A、T、C和G環環相扣，形成DNA長鏈。整條長鏈中只有一部分盛載着特定的遺傳指令，那便是「基因」。

若以英語作比喻，基因便有如一段完整的句子或文章段落，用來表達一個完整的訊息。



## Engaging Stakeholders and Furthering the Reach

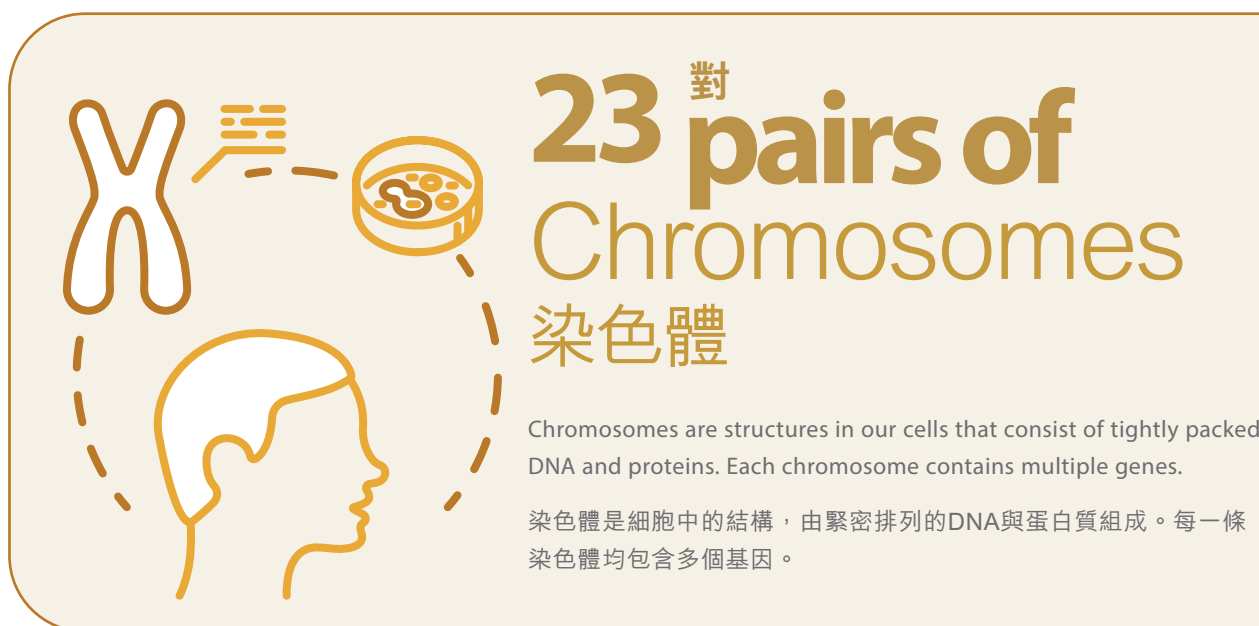
A proactive approach has been adopted to build rapport with targeted stakeholders and the media to keep them abreast of our latest development. In Q3 2021, we hosted a visit for the former Secretary for Food and Health to HKGI with a tour to one of our PCs, the Hong Kong Children's Hospital, to learn about updates on HKGI and HKGP. For Legislative Council members, we engaged with them through a meeting with the Panel on Health Services, in which we briefed them on HKGI's progress and answered members' questions in relation to HKGP. To further extend our reach and spread key messages to the public, a media briefing was organised to introduce the background of HKGI and HKGP, and to clarify misperceptions on the Project. Through extensive and positive news coverage by top-tier media outlets, we were able to enhance public awareness of genomic medicine and to promote its development in Hong Kong.

Moreover, HKGI has maintained close communication with targeted organisations including the Hong Kong Academy of Medicine and the Hong Kong Society of Genetic Counselling. Through engagements with these groups, we hope to better promote awareness of genomic medicine and its benefits to professional bodies, in addition to the general public.

## 多管齊下 接觸持份者

基因組中心一直主動與持份者及媒體建立良好關係，讓他們了解中心的最新發展。前食物及衛生局局長於2021年第三季到訪，並參觀了其中一間夥伴中心香港兒童醫院，了解基因組中心及基因組計劃的工作進展。基因組中心團隊亦透過參與立法會衛生事務委員會的會議，與議員進行交流，向他們簡述機構的發展，並解答有關基因組計劃的問題。為進一步向公眾推廣重要信息，基因組中心舉辦了首個傳媒簡報會，介紹機構及基因組計劃的背景，釋除公眾對計劃的疑慮。藉着媒體廣泛而正面的報道，基因組中心成功提高公眾對基因組醫學的認識，促進基因組醫學在香港的發展。

此外，基因組中心與不同專業機構保持緊密聯繫，包括香港醫學專科學院及香港遺傳諮詢學會，希望藉此提高專業團體乃至公眾對基因組醫學及其效益的認識。



**23 對 Chromosomes**  
**染色體**

Chromosomes are structures in our cells that consist of tightly packed DNA and proteins. Each chromosome contains multiple genes.

染色體是細胞中的結構，由緊密排列的DNA與蛋白質組成。每一條染色體均包含多個基因。



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

### Looking Forward to More Engagement

Our efforts have been met with positive reception from the public, the industry and stakeholders. We will continue to improve understanding of genomic medicine and its significance in the coming year. One of our key activities will be the production of expert videos featuring esteemed HKGI Board Members, who will share their inspiring journey in the research and development of genetics and genomics, and the vast potential of genomic medicine in shaping the future of healthcare.

To strengthen online presence and maximise effectiveness of our public education initiatives, various platforms including YouTube and Google are being explored. Efforts will also be made to explore broadcasting the videos at PCs and public hospitals for enhanced exposure. This will assist patient recruitment and promote awareness of genomic medicine.

Last but not least, we will further expand our engagement with patient groups and professional bodies through hosting meetings or visits. We will also participate in industry events and continue to work closely with the Health Bureau and targeted stakeholders to foster better understanding of and support for HKGI's work.

### 多方合作 積極參與

基因組中心努力不懈，獲得公眾、業界及持份者的認同，來年將繼續致力加強公眾對基因組醫學及其重要性的了解，其中一項重點計劃是董事局權威學者的專訪系列，由他們透過影片分享在遺傳學和基因組學方面的研發歷程，介紹基因組醫學對塑造醫療行業前景的巨大潛力。

為加強網上曝光及提升公眾教育活動的成效，基因組中心正探索YouTube及Google等平台及推廣途徑，同時爭取在夥伴中心及公立醫院播放影片，協助招募病人及推廣基因組醫學。

最後，基因組中心將會進一步加強與病人組織和專業團體的聯繫，例如舉辦會議或參觀活動，亦會參與業界活動，並與醫務衛生局及相關持份者保持緊密合作，讓他們深入了解及繼續支持基因組中心的工作。



**Operate with Excellence**  
**卓越營運**



## Operate with Excellence 卓越營運

When HKGI was founded, we immediately established essential procedures, controls, and guidelines to govern our operations. Although HKGI is a newly established organisation, we have operated with the greatest levels of accountability, transparency, effectiveness, and integrity, right from the start. Along with the meticulousness and professionalism for the HKGP as well as clinical and research work, our pursuit of operation excellence is instrumental in maintaining public trust, which is vital for HKGI as a publicly funded organisation.

### Securing Our Future with Talent Acquisition

As a start-up, it is critical to set up infrastructure, protocols, and systems for effective human resources management. In this regard, we have implemented comprehensive processes, including recruitment, compensation and benefits, payroll procedures, performance management and other essential policies. An employee handbook was published to clearly lay out our organisational principles.

To date, HKGI has over 60 staff members housed under scientific, bioinformatics and administration branches. Given the novelty of genomic medicine in Hong Kong, staff recruitment is our top priority but has been challenging, especially for the scientific and bioinformatics branches. This was due to the limited pool of relevant professionals and experts in genomic medicine who possess the required knowledge, skill set and experience in the market.

To address this challenge, we are on the lookout for fresh talent by reaching out to students in related fields and providing training and internship opportunities, in addition to exploring different recruitment channels through industry platforms and referrals. This will help us recruit additional staff to be engaged in specialised roles with the expansion of HKGP in scope. We also launched our first summer internship programme in July 2022 which provided valuable work experience for seven interns.

基因組中心成立後，隨即制訂各項必要的程序、措施及政策作為機構運作的基礎。縱然基因組中心是新成立的機構，其運作仍貫徹最高標準的問責制、透明度、效率及誠信，不僅奉行一絲不苟的專業精神推行基因組計劃及開展臨床研究，亦追求卓越營運，此乃維持公眾信任的關鍵，這一點對政府資助機構而言尤為重要。

### 吸納人才 共創未來

作為初創機構，建立基礎設施、規程守則及系統對達致有效的人力資源管理至關重要。為此，基因組中心實施全方位的流程，包括招聘、薪酬福利、支薪程序、績效管理等基本政策，並且在僱員手冊中清楚列明機構的原則。

目前，基因組中心的科學、生物信息學及行政部門共有逾60名員工。鑒於基因組醫學在香港屬相對嶄新的領域，人才招聘乃基因組中心的首要任務，而且充滿挑戰，物色科學及生物信息學的人才更為困難，原因是市場上缺乏具備相應知識、技能及經驗的基因組醫學專才及專家。

面對這個挑戰，基因組中心積極探索不同的招聘途徑，除了業內平台及推薦等方式，亦接觸相關學科的學生，向他們提供培訓及實習機會。此舉有助招攬更多擔任專業崗位的員工，迎合基因組計劃服務範圍日益擴大的需要。基因組中心於2022年7月推出首個暑期實習計劃，七名實習生從中獲得寶貴的工作經驗。

## Driving Excellence with Continuous Learning

As part of our commitment to nurturing talents, we are developing appropriate career paths for young people and experienced professionals through tailor-made on-the-job training. A culture of continuous learning creates momentum for new staff members and inspires future generations to drive the development of genomic medicine.

Throughout the year, we organised a wide range of training workshops and programmes on various topics including integrity management, root cause analysis, and cybersecurity. Industry experts and veterans were also invited to present lectures for staff members to equip with domain knowledge in various fields.

## Safeguarding the Health of Our Staff

HKGI sees staff as the most important asset. Various preventive measures were implemented to safeguard the health and safety of employees during the pandemic. These included strengthened infection control and enhanced cleaning and ventilation of the office area, adoption of staggered lunch hours, provision of COVID test kits and implementation of testing on alternate workdays, as well as close supervision of social distancing measures. A work group has also been set up to closely monitor the pandemic situation and ensure timely response if assistance is needed from staff members.

## 持續學習 追求卓越

基因組中心致力培育人才，精心規劃在職培訓內容，為年輕人及具經驗的專業人員規劃合適的事業方向；同時營造鼓勵持續學習的文化，啟發新加入的年輕員工推動基因組醫學的發展。

此外，基因組中心於年內就多個主題舉辦一系列的培訓研討會及課程，內容涵蓋誠信管理、根源分析法及網絡安全等，邀請相關行業專家及資深人士作分享，鞏固員工在不同領域的專業知識。

## 關懷同事 保障健康

人才是基因組中心最為寶貴的資產，在疫情肆虐期間，基因組中心落實多項防疫措施，保障同事的健康及安全，當中包括加強辦公室的防疫工作、提升清潔及通風水平、分開午膳時間、提供新冠病毒快速抗原測試包，並且每隔一個工作天檢測一次，亦嚴謹執行社交距離措施。基因組中心更成立工作小組，密切監察疫情，確保同事有需要時能即時得到協助。



## Operate with Excellence

### 卓越營運

#### Setting Up Collaborative Workplace

Our work is vital to the future of Hong Kong, and we have established a base to reflect this. Before HKGI's office was set up, our teams operated out of satellite offices across Hong Kong Science Park. Specific consideration had been given to the design and set-up of our new office, given its multifunctional nature, including a full-scale laboratory, as well as working, collaboration, and meeting areas for all of our teams. We designed and optimised the new office area to enhance productivity and efficiency. While areas are zoned out for different teams, there is ample open space to encourage discussion, collaboration and exchange of ideas. The whole office is also kitted out with facilities and furniture that meet with the occupational health and safety requirements for the benefit of our staff.

#### Building Robust IT Infrastructure

Information Technology (IT) has played a crucial role along our journey of starting from scratch to commencing full operations. In addition to the IT infrastructure for our bioinformatics and laboratory work, we have established robust IT systems for the office to support operations. We had the first system set up while the teams operated out of several satellite offices in Hong Kong Science Park, and the second one for the new office when all the teams moved in August 2021. The system and data migration was challenging but with detailed planning and thorough preparation, we enjoyed a seamless transition in our new office. We have since then continued to further strengthen the network and architecture to accommodate the increasing number of staff members and increased workload for enhancing productivity and efficiency.

Furthermore, comprehensive documentation for proper IT control and maintenance has been compiled. The manual includes an array of procedures pertaining to day-to-day work and operations, which has been updated regularly to keep our staff abreast of the latest measures.

With data security paramount, we formulated strict IT and cybersecurity policies with the management and internal audit team to ensure best practices were adopted and implemented throughout the organisation. To further enhance IT security, the "BitLocker Drive Encryption" function has also been enabled on all laptops for data protection when the devices have to be taken out of office.

#### 加強協作 各展所長

基因組中心的工作對香港的未來影響深遠。基因組中心在辦公室正式投入運作前，於香港科學園內不同大樓設立了臨時分部。鑒於新的辦公室需要滿足不同功能，基因組中心對新辦公室的設計與佈置加入了針對性的考量，包括設立功能齊備的實驗室，亦提供可以容納所有團隊辦公、協作和舉行會議的空間，藉此提升生產力和效率。辦公室不但劃分了不同區域供不同團隊使用，更有足夠的空間讓同事進行討論、協作和交流。辦公室的設施與設備均符合職安健的要求，令員工能夠在安全舒適的環境下盡展所長。

#### 完善系統 提升效率

無論是起步階段還是全面投入運作後，資訊科技同樣不可或缺。除生物信息學及實驗室工作所需的基礎設施外，基因組中心亦在辦公室建立完善的資訊科技系統，令運作更暢順，當中包括在香港科學園各個臨時分部已經建立的首套系統，以及其後於2021年8月遷入新辦公室時建立的第二套系統。系統和數據轉移並非易事，但有賴周密的規劃和充分準備，在遷入新辦公室時成功達致無縫銜接。此後，基因組中心繼續提升網絡及架構，應付員工人數及工作量不斷增加的需要，從而提升生產力和工作效率。

此外，基因組中心亦就管制及維持資訊科技系統編製了周詳完善的手冊，內容包括日常工作及營運的各項程序，並定期更新內容，確保員工第一時間獲得最新措施的資訊。

基因組中心非常重視數據安全，故此制訂了嚴密的資訊科技及網絡安全政策，管理層及內部審核團隊亦積極參與其中，確保中心採用及貫徹最佳的作業流程。為進一步提升資訊科技保安，資訊科技團隊在所有筆記型電腦啟用了「BitLocker磁碟機加密」功能，務求電腦離開辦公室範圍後亦獲得數據保護。



## Empowering Operations Through Finance and Procurement

We have put in place stringent financial controls to ensure that our resources are used prudently and effectively. All expenditures are managed in a cost-efficient manner and payment authorities are governed by relevant policies and guidelines endorsed by the Finance and Administrative Committee (FAC). Our financial results are also overseen by the FAC and the Audit and Risk Committee.

We also have robust Guidelines and Procedures on Procurement, setting out the delegated authority for approval and prescribed procedures for conducting procurement of goods and services for HKGI to safeguard fairness and integrity of the procurement process. Over 70 quotation and tender exercises for provision of hardware, software and services, including the purchase of sequencing and bioinformatics services, have been conducted during the year.

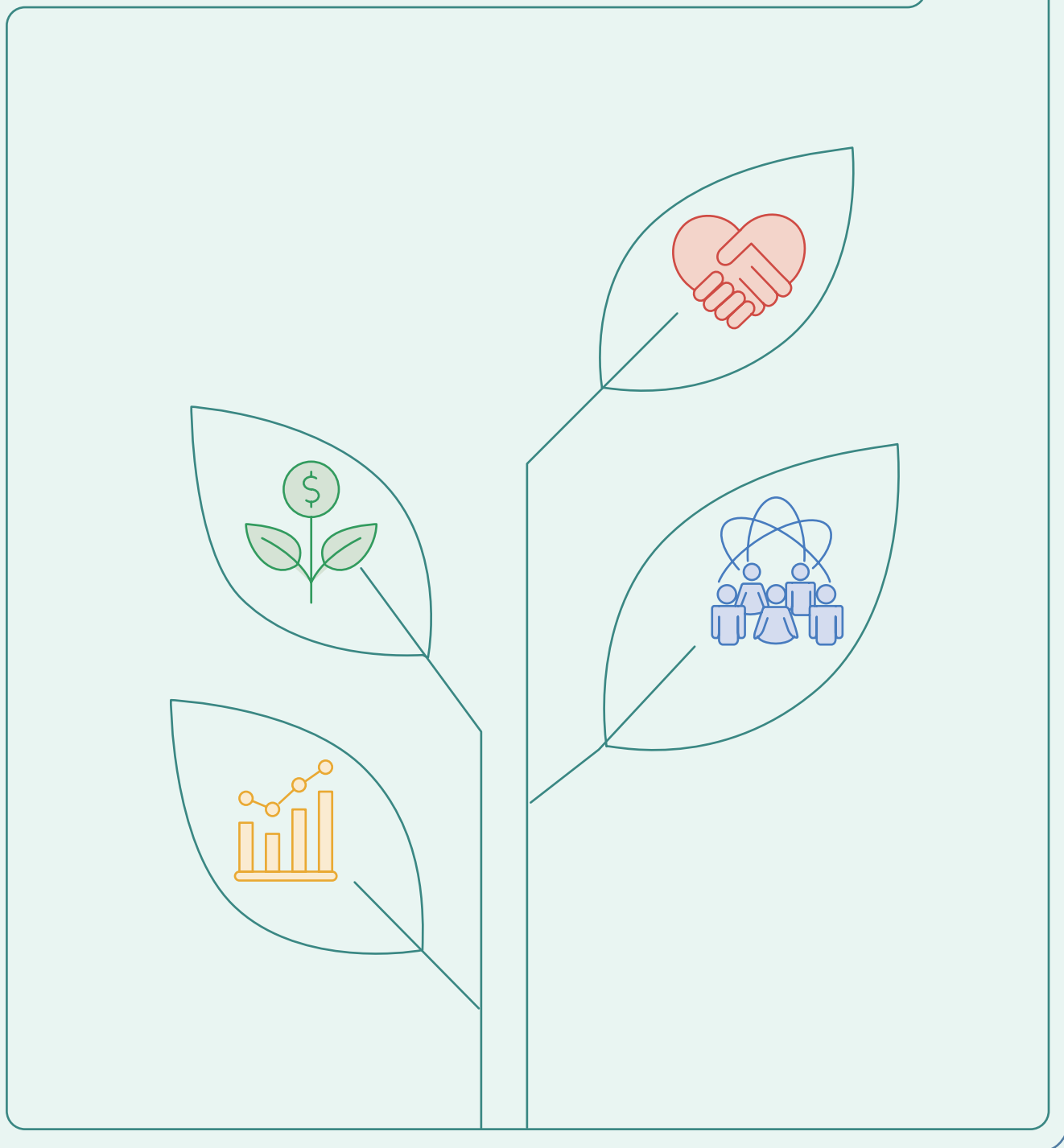
## 審慎理財 穩健營運

基因組中心制訂了嚴謹的財務管理守則，以確保審慎有效地運用資源。所有開支均按照成本效益原則進行管理，而付款權限則根據財務及行政委員會認可的政策及指引規管，機構的財務狀況亦由財務及行政委員會，以及審計及風險委員會予以監督。

另外，基因組中心制訂了完善的採購指引及程序，列明貨品及服務採購的審批授權與既定程序，使採購流程符合公平誠信的原則。年內，基因組中心就硬件、軟件及服務展開了逾70項報價及招標工作，涵蓋採購測序及生物信息學服務等事宜。

# Corporate Governance

## 企業管治



# Principles and Practices

## 原則與實務

A robust corporate governance system centred around the Board of Directors has been put into place to govern the operations of HKGI and the implementation of HKGP, observing the core corporate governance principles of accountability, transparency, fairness, and responsibility.



### Accountability

The Board of Directors of HKGI is accountable to various stakeholders, including the HKSAR Government, participants of HKGP, the general public, as well as a wide range of community groups. HKGI maintains close communication with the HKSAR Government. There are three public officers serving on its Board, namely, the Under Secretary for Health, the Deputy Secretary for Health, and the Deputy Director of Health. The HKGI Chairperson and the Chief Executive Officer (CEO) of HKGI regularly meet with the government officials to discuss issues relating to the work of HKGI. The CEO of HKGI attends meetings of the Legislative Council (LegCo) together with government officials to brief LegCo Councillors on the plans, policies and operations of HKGI, and answer their questions as needed.

In accordance with the Memorandum of Administrative Arrangements (MAA) signed with the HKSAR Government, HKGI shall, as soon as practicable and in any case, not later than six months after the expiry of a financial year, furnish a report on the activities of HKGI, a copy of the statement of accounts of HKGI, and the auditor's report for that year to the Permanent Secretary for Health (PSH). In this respect, an annual report, with the approval of the Board of Directors, will be published for each financial year. HKGI is also required to submit an Annual Plan cum Draft Estimates of Income and Expenditure for the coming financial year to PSH in the first quarter of each year, outlining the work it will carry out to achieve its strategic goals during the year.

HKGI recognises the importance of risk management as a systematic tool for identifying, analysing, assessing, and treating all types of risks attached to its activities and resources. It has adopted an integrated Enterprise Risk Management framework to provide a holistic view of the enterprise risks facing the organisation. Reporting to the Board, the Audit and Risk Committee receives and

我們已建立以董事局為中心的健全企業管治制度，以管理基因組中心的運作及基因組計劃的實施，並遵守問責、透明、公平及責任的核心企業管治原則。

### 問責

基因組中心董事局對不同持份者負責，包括特區政府、基因組計劃參加者、公眾，以及社會上不同類型的團體。基因組中心與特區政府保持緊密溝通，共有三名公職人員參與董事局的工作，分別是醫務衛生局副局長、醫務衛生局副秘書長及衛生署副署長。基因組中心主席及行政總裁定期與政府官員會面，討論與基因組中心工作的相關事宜。此外，基因組中心行政總裁會與政府官員一同出席立法會會議，向立法會議員介紹基因組中心的計劃、政策及運作，並適時回答問題。

根據與特區政府簽署的行政安排備忘錄，基因組中心須在切實可行的情況下，不遲於財政年度屆滿後六個月，盡快向醫務衛生局常任秘書長提交有關基因組中心的活動報告、基因組中心的賬目報表副本及年度核數師報告。經董事局批准後，基因組中心將就上述資料刊發各財政年度的年度報告。基因組中心亦須於每年第一季度向常任秘書長提交下一財政年度的年度計劃及收支預算草案，概述其在年內為實現策略目標而將開展的工作。

基因組中心明白風險管理作為系統工具，在識別、分析、評估及處理與活動及資源相關的各類風險中的重要性，並已採用一套綜合企業風險管理框架，以全面了解機構面臨的企業風險。審計及風險委員會代表董事局接收及審議有關主要企業風險及相關緩解策略

## Principles and Practices 原則與實務

considers internal audit reports on key enterprise-wide risks and the related mitigation strategies. It also monitors HKGI's financial and administrative control processes to ensure the safeguarding of resources and operational efficiency. This is achieved by reviewing HKGI's overall management and control framework, measures for mitigating significant risks in its key business processes, and through the external audit reports.



### Transparency

Adequate degree of transparency and disclosure of information about the organisation are important pillars of a good corporate governance system. HKGI has established various means and channels to enhance transparency, disclose information, and report progress of work to its stakeholders. The *HKGI Strategic Plan 2022-25*, as an overarching document for guiding all aspects of HKGI's development and planning in the coming three years, was approved by the Board of Directors at its meeting held in September 2021, and subsequently accepted by PSH for publication and promulgation to various stakeholders. The strategic priorities of the Annual Plans 2022-23, 2023-24 and 2024-25 are guided by the strategic goals, directions, and strategies set out in the HKGI three-year *Strategic Plan* to channel resources to specific programmes for translating these strategies into actions.

For transparency and openness, the HKGI website provides up-to-date and comprehensive corporate information about HKGI in the form of texts and videos. HKGI also maintains close dialogue with the media and other key stakeholders, particularly the patient groups and relevant professional bodies. It issues press releases and holds media briefings to inform the media and the public on all current issues and important matters relating to its work.

The six functional committees submit progress reports to the Board of Directors on a regular basis, informing the Board of the major deliberations and decisions made by individual committees. The management also submits to the Board regular reports on the implementation progress of HKGP as well as the progress of carrying out the programmes and initiatives set out in the Annual Plans.

的年度報告，並通過仔細審核基因組中心整體管理及控制框架、降低主要業務流程中重大風險的措施的報告以及透過外部審計報告，監察基因組中心的財務及行政控制流程，確保資源運用得宜及保障營運效率。

### 透明

良好企業管治制度的重要支柱，包括高度透明及充分披露機構的資料。基因組中心已建立多種方式及渠道向持份者提高透明度、披露充足資料及報告工作進度。作為指導基因組中心未來三年各方面發展及規劃的總體文件，《2022-25年策略計劃》已於2021年9月舉行的董事局會議上得到通過，隨後獲常任秘書長同意發表並頒布予各持份者。2022-23、2023-24及2024-25年度計劃的策略優先事項以三年策略計劃所載的策略目標、方向及制訂的策略為指引，將資源用於具體計劃，使有關策略轉化為行動。

為保持透明及公開，基因組中心網站以文字及影片形式提供有關基因組中心最新及最全面的信息。基因組中心亦與傳媒及其他主要持份者，尤其是病人組織及相關專業團體保持緊密聯繫，並發布新聞稿及舉行傳媒簡報會，向傳媒及公眾通報所有當前問題及與其工作有關的重要事項。

六個專責委員會定期向董事局提交進度報告，通報個別委員會的主要審議及決定。管理層亦向董事局提交定期報告，匯報基因組計劃的進度和年度計劃中所載方案及措施的進展。



## Fairness

HKGI embraces the principle of fairness and strives to treat all its stakeholders equally and ethically. It has involved the medical and legal experts as well as patient advocate serving on its Ethics Advisory Committee in the development and implementation of an ethically sound system for seeking patients' informed consent to participate in HKGP. Before the commencement of patient recruitment for the Project in July 2021, HKGI had successfully obtained ethical approval from the relevant Institutional Review Boards of its Partnering Centres (PCs).



## Responsibility

The Board of Directors accepts full responsibility for the powers that it is given and the authority that it exercises. It is responsible for overseeing and monitoring the management of HKGI's operations, its activities and performance. In this respect, it acknowledges its responsibility for establishing and ensuring the effectiveness of HKGI's internal control system, which is designed to provide reasonable assurance regarding the achievement of the objectives in the categories of effectiveness and efficiency of operations; reliability of internal and external reporting; and compliance with applicable laws, regulations and internal policies/guidelines. This responsibility is delegated to the CEO in daily operations.

## 公平

基因組中心奉行公平原則，也致力以平等及合乎倫理的方式對待所有持份者。倫理諮詢委員會成員包括醫學及法律專家，以及病人代表，共同制訂及實施合乎倫理的健全制度，尋求病人知情同意參與基因組計劃。於2021年7月開始為基因組計劃招募病人前，基因組中心已成功從夥伴中心的相關倫理審查委員會獲得批准。

## 責任

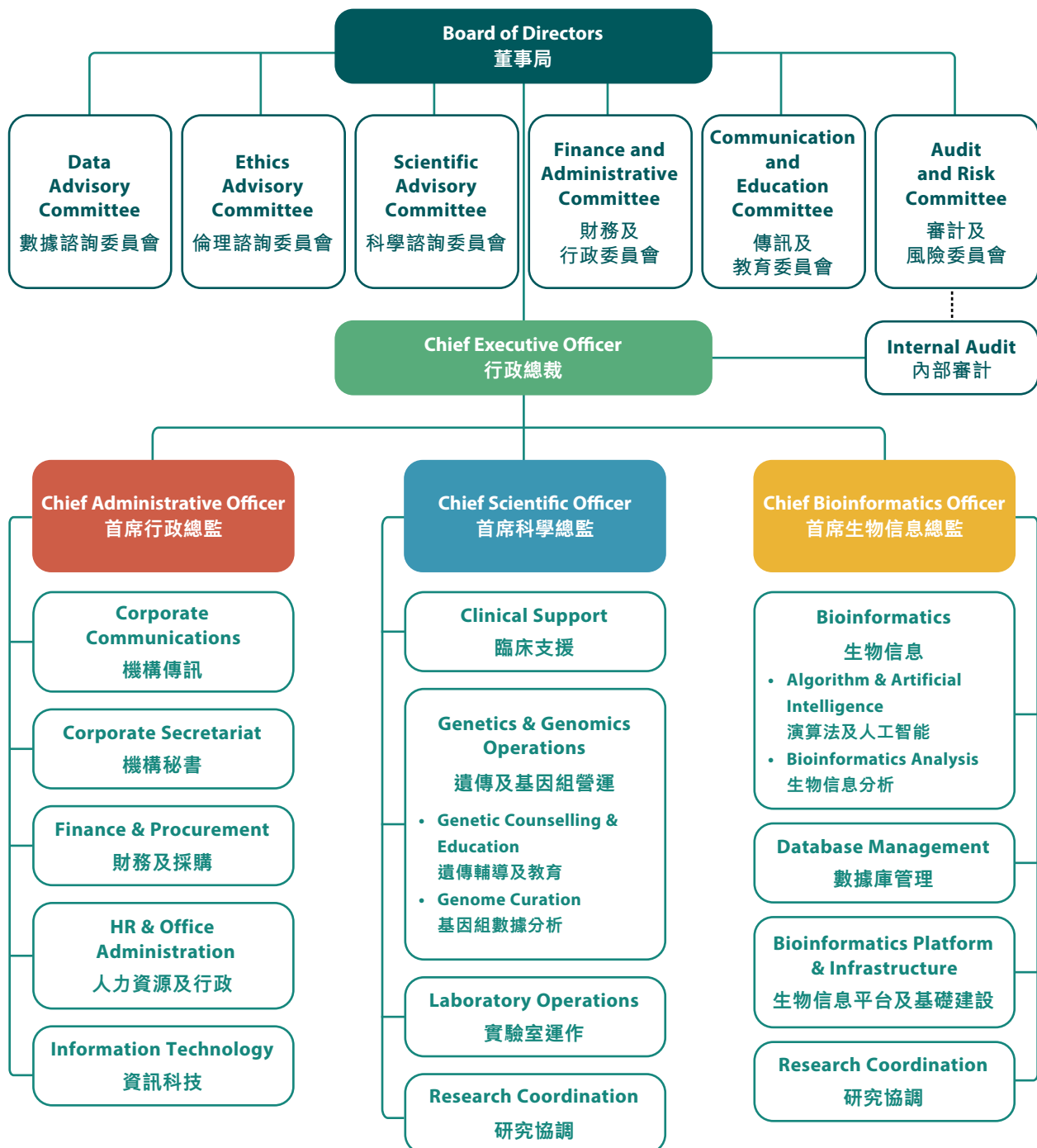
董事局對其獲賦予的權力及行使的職權承擔全部責任。董事局負責監督及監察基因組中心的營運、活動及業績的管理。就此而言，董事局有責任建立及確保基因組中心的內部控制系統行之有效，確保合理地實現營運效能及效率的目標；內部及外部匯報的可靠性；並且遵守適用的法律、規例及內部政策／指引。相關的責任，在日常營運中會委派予行政總裁。



## Governance Structure 管治架構

HKGI has set up an effective corporate governance structure comprising the Board of Directors and six functional committees to provide policy directions and implementation guidance to the executive management.

基因組中心已建立有效的企業管治架構，由董事局及六個專責委員會組成，向管理團隊提供政策方向及實務指引。



## Board

### Board Functions

HKGI is a company limited by guarantee, established and wholly owned by the HKSAR Government to accelerate the development of genomic medicine in Hong Kong. It is accountable to the HKSAR Government through the Secretary for Health. According to Section 14 of the Articles of Association of HKGI registered under Company Ordinance (Cap. 622), the operations and affairs of the Institute are managed by the Board of Directors, who may exercise all the powers of the Institute. Therefore, the Board of Directors is the highest authority in the governance structure of HKGI.

### Board Diversity

Membership of the HKGI Board of Directors comprises 16 non-public officers and three public officers, engaging experts from different sectors, including biomedical scientists, medical ethicists, data scientists, bioinformaticians, genetic counsellors, accountants, and public educationalists, etc., in promoting the development of genomic medicine in Hong Kong.

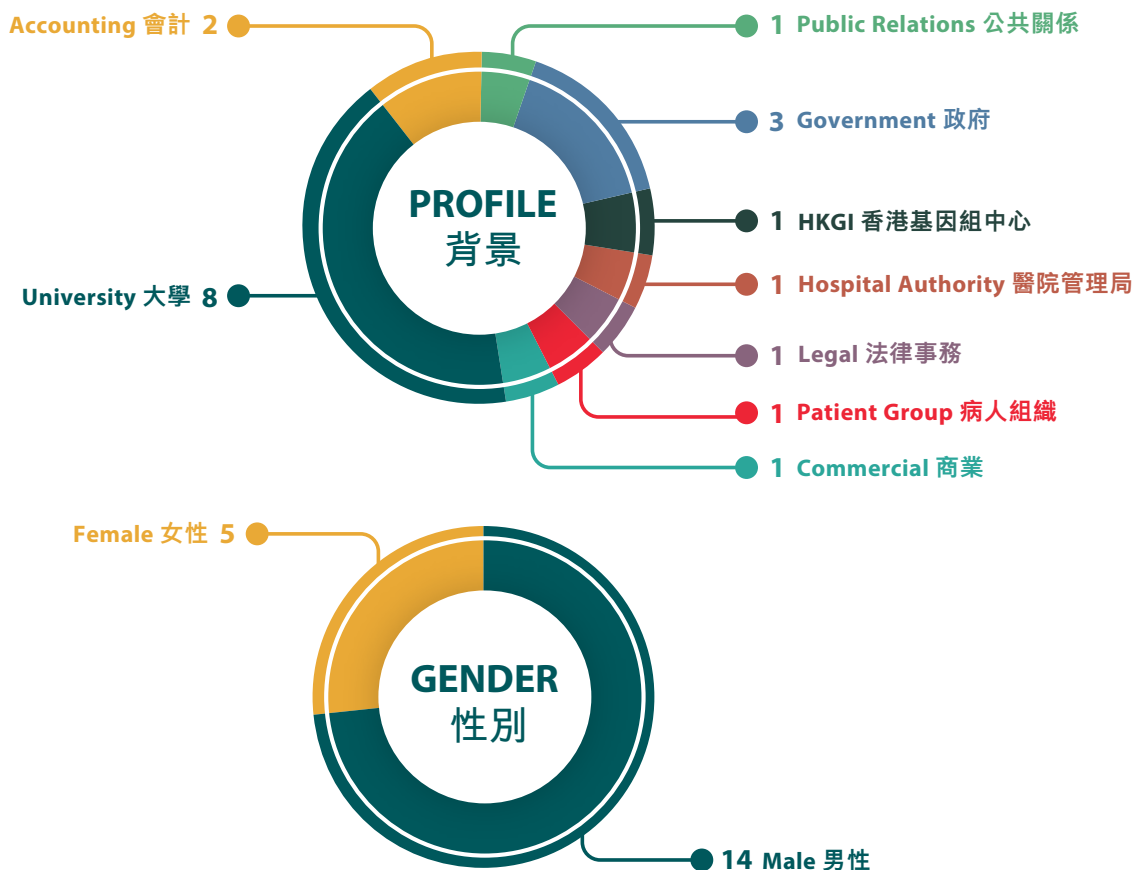
## 董事局

### 董事局職能

基因組中心是一家由特區政府成立並全資擁有的擔保有限公司，旨在促進香港基因組醫學的發展，透過醫務衛生局局長向香港特區政府負責。根據《公司條例》(第622章)註冊的基因組中心的《組織章程細則》第14條，基因組中心的運作及事務由董事局管理，董事局可對基因組中心行使所有權力。因此，董事局是基因組中心管治架構中的最高權力機構。

### 董事局多元化

基因組中心董事局成員由16名非公職人員及三名公職人員組成，當中委任來自不同領域的專家，包括生物醫學科學家、醫學倫理學家、數據科學家、生物信息學家、遺傳輔導員、會計師及公共教育學家等，促進香港基因組醫學的發展。



## Governance Structure 管治架構

### Board Meetings

Since its inception in November 2020 to June 2022, the Board of Directors had held 10 meetings with 71 papers (including four circulation papers) disseminated to facilitate deliberation and decision. Issues discussed included Standing Orders of the Board, MAA between the HKSAR Government and HKGI, MAA between HKGI and PCs, provision of bioinformatics and sequencing services, communication and education programmes, lease agreement of the HKGI office cum laboratory, ethics review application for HKGP, Audited Financial Statements and Financial Reports, implementation progress of HKGP, *Strategic Plan 2022-25*, Annual Plans and Draft Estimates for 2021-22 and 2022-23, Data Privacy Assessment, new initiatives of HKGI, proposal for establishment of the Hong Kong Genetic Counselling Practice Consortium, and progress reports of functional committees. Attendance rate of Board members at these 10 meetings is shown below:

### 董事局會議

董事局自2020年11月成立，直至2022年6月，共舉行10次會議，發布71份文件（包括4份傳閱文件），以供審議和決策。討論的議題包括董事局議事規則、特區政府與基因組中心簽訂的行政安排備忘錄、基因組中心與夥伴中心簽訂的行政安排備忘錄、提供生物信息學及測序服務、傳訊及教育計劃、基因組中心辦公室及實驗室的租賃協議、基因組計劃的倫理審查申請、經審計的財務報表及財務報告、基因組計劃的實施進度、《2022-25年策略計劃》、2021-22年及2022-23年的年度計劃及預算草案、數據私隱評估、基因組中心的新舉措、設立香港遺傳輔導專業發展聯席(Hong Kong Genetic Counselling Practice Consortium)的建議及各專責委員會的進展報告等。董事局成員於該10次會議的出席率如下：

Members 成員		Attendance Rate (%) 出席率(%)
Chairperson: 主席	Mr Philip TSAI Wing-chung 蔡永忠先生, BBS, JP	100
Deputy Chairperson: 副主席	Professor Raymond LIANG Hin-suen 梁憲孫教授, SBS, JP	100
Non-official Directors: 非官方董事	Dr LO Su-vui 羅思偉醫生	100
	Dr Derrick AU Kit-sing 區結成醫生	80
	Mr Ray CHAN Chin-ching 陳展程先生	90
	Professor CHAN Wai-yee 陳偉儀教授	90
	Ms Ivy CHEUNG Wing-han 張穎嫻女士	90
	Dr CHUNG Kin-lai 鍾健禮醫生	80
	Professor Nancy IP Yuk-yu 葉玉如教授, SBS, BBS, MH, JP	80
	Professor LAU Chak-sing 劉澤星教授, BBS, JP	60
	Dr Shawn LEUNG Shui-on 梁瑞安博士	90
	Dr Isabella LIU Fang-chun 劉芳君博士	90
	Professor Dennis LO Yuk-ming 盧煜明教授, SBS, JP	70
	Professor Alfonso NGAN Hing-wan 顏慶雲教授	90
	Mr Tim PANG Hung-cheong 彭鴻昌先生	90
Professor YIU Siu-ming 姚兆明教授	100	
Official Directors: 官方董事	Dr CHUI Tak-yi 徐德義醫生, JP	100
	Ms Shirley KWAN Yu-pik 關如璧女士	100
	Dr Teresa LI Mun-pik 李敏碧醫生, JP	100

## Committees

For optimal performance of its roles and exercise of powers, the Board of Directors of HKGI has formed six functional committees, namely, the Data Advisory Committee, Ethics Advisory Committee, Scientific Advisory Committee, Finance and Administrative Committee, Communication and Education Committee, and Audit and Risk Committee.

### Data Advisory Committee

#### Membership

Convenor:	Professor YIU Siu-ming
Non-official Members:	Professor Andrew CHAN Man-lok Professor CHAN Ting-fung Dr Chris CHAN Tsun-leung Dr CHEUNG Ngai-tseung Dr Lucas HUI Chi-kwong Professor JIANG Pei-yong Professor Terrence LAU Chi-kong Dr Shawn LEUNG Shui-on Dr Isabella LIU Fang-chun Professor LUO Qiong Professor Ian WONG Chi-kei Professor YANG Wan-ling
Official Member:	Representative from the Health Bureau

#### Terms of Reference

1. To advise on the overall architecture for storing and accessing data for HKGP.
2. To review and approve protocols related to the data access and transfer of HKGP.
3. To advise on the set up and operation of the genome database.
4. To advise on data related issues surrounding genomic medicine, as requested by the Board.

#### Focus of Work

Since its inception, the Data Advisory Committee had conducted five meetings to discuss and advise on the strategies and implementation initiatives relating to the establishment of HKGI's bioinformatics platform services in support of HKGP. These included the provision of bioinformatics services for HKGI, its data access protocol for the Clinical Frontend, and the bioinformatics platform's data and security standards, data privacy assessment as well as disaster recovery plan. The Committee also received updates on the progress of sample analysis for HKGP participants, HKGI's initiatives to standardise its Cloud Infrastructure and the development of an inhouse Bioinformatics Analysis Browser.

## 委員會

為充分發揮最大作用及行使職權，基因組中心董事局成立了六個專責委員會，分別是數據諮詢委員會、倫理諮詢委員會、科學諮詢委員會、財務及行政委員會、傳訊及教育委員會以及審計及風險委員會。

### 數據諮詢委員會

#### 成員

召集人：	姚兆明教授
非官方成員：	陳文樂教授 陳廷峰教授 陳俊良博士 張毅翔醫生 許志光博士 江培勇教授 劉智剛教授 梁瑞安博士 劉芳君博士 羅瓊教授 黃志基教授 楊萬嶺教授
官方成員：	醫務衛生局代表

#### 職權範圍

1. 就基因組計劃整體的數據儲存及讀取權限提供意見。
2. 審視及批准與基因組計劃數據讀取及轉移有關的規程。
3. 就設立基因組數據庫及其運作提供意見。
4. 按董事局要求，就與基因組醫學數據相關的議題提供意見。

#### 工作重點

自成立以來，數據諮詢委員會舉行了五次會議，就基因組中心設立生物信息平台以支援基因組計劃的策略及執行細節進行討論，並提出意見。其中包括基因組中心所需的生物信息服務、臨床資訊管理平台存取數據的規程、生物信息平台的數據及安全標準、數據私隱評估以及事故復原計劃。委員會亦審視了有關基因組計劃樣本分析的進展、基因組中心標準化雲端基礎設施的舉措，以及生物信息學分析開發瀏覽器的最新資料。

## Governance Structure 管治架構

### Ethics Advisory Committee

#### Membership

Convenor:	Dr Derrick AU Kit-sing
Non-official Members:	Dr Josephine CHONG Shuk-ching Dr Calvin HO Wai-loon Professor Stephen LAM Tak-sum Professor LEUNG Suet-yi Mr Tim PANG Hung-cheong Dr Mary TANG Hoi-yin Mr Stephen WONG Kai-yi Mr James YIP Shiu-kwong
Official Members:	Representative from the Health Bureau Representative from the Department of Health

#### Terms of Reference

1. To provide ethical oversight for HKGP.
2. To consider, review and approve the ethics protocol of HKGP with reference to local and international practices.
3. To advise on patient consent protocol and arrangement.
4. To identify, define, examine and respond to ethical issues in HKGP to ensure its delivery is in the interests of the participants and the public.
5. To advise the Board on ethical issues related to genomic medicine, as requested by the Board.

#### Focus of Work

Since its inception in January 2021, the Ethics Advisory Committee met five times to discuss and oversee the ethical issues involved in the implementation of HKGP. It advised on the ethics protocols of HKGP, including patient consent protocol and arrangement and contents of the adult consent form, consent form for parents/legal guardians signing on behalf of minors, and consultee consent form for participants with mental incapacity. It also considered and endorsed HKGI's Ethics Review Application for submission to the Institutional Review Boards of PCs, the withdrawal procedures of HKGP, and HKGI's proposed approach to address the ethical issues arising from secondary findings of whole genome sequencing.

### 倫理諮詢委員會

#### 成員

召集人：	區結成醫生
非官方成員：	莊淑貞醫生 何維倫博士 林德深教授 梁雪兒教授 彭鴻昌先生 唐海燕醫生 黃繼兒先生 葉兆光先生
官方成員：	醫務衛生局代表 衛生署代表

#### 職權範圍

1. 監督實施基因組計劃的倫理問題。
2. 參考本地及國際慣例以考慮、審視及批准基因組計劃的倫理規程。
3. 就獲取病人知情同意的規程及相關安排提供意見。
4. 識別、界定、審查及回應實施基因組計劃所涉及的倫理問題，確保有關處理符合參加者及公眾的利益。
5. 按董事局要求，就基因組醫學的相關倫理問題提供意見。

#### 工作重點

自2021年1月成立以來，倫理諮詢委員會舉行了五次會議，以討論及監察實施基因組計劃所涉及的倫理問題。委員會就基因組計劃的倫理規程提供意見，包括獲取病人知情同意的規程及相關安排、成人同意書、父母／法定監護人代表未成年人士簽署的同意書，以及由代決人代表無自決能力之成年參加者簽署的同意書。委員會亦審議及批准了基因組中心提交予夥伴中心研究倫理委員會的倫理審查申請、退出基因組計劃的程序，以及基因組中心如何處理進行全基因組測序時，次要發現所引起的倫理問題。



## Scientific Advisory Committee

### Membership

Convenor:	Professor Dennis LO Yuk-ming, SBS, JP
Non-official Members:	Professor Godfrey CHAN Chi-fung Dr Gladys KWAN Wai-man Professor LAU Chak-sing, BBS, JP Dr Danny LEUNG Chi-yeu Professor LEUNG Tak-yeung Dr Edmond MA Shiu-kwan Professor Tony MOK Shu-kam, BBS Professor SHAM Pak-chung, JP Professor Michael YANG Mengsu
Official Members:	Representative from the Health Bureau Representative from the Department of Health

### Terms of Reference

1. To consider, advise and approve the clinical, laboratory and research protocols of HKGP.
2. To advise the Board on the latest science and technologies relevant to the effective implementation of HKGP.
3. To determine the research priorities of the main phase of HKGP.
4. To advise the Board on the scientific issues in genetics and genomics, as requested by the Board, with a view to promoting genomic medicine in Hong Kong.

### Focus of Work

Since its establishment, the Scientific Advisory Committee met five times to discuss and consider matters relating to the planning and implementation of the pilot and main phases of HKGP, and the scientific issues involved. These included the research protocol for HKGP, the types of clinical information required for the pilot phase of HKGP (i.e. undiagnosed disorders and hereditary cancers), principles of main findings reporting, multi-disciplinary team meeting arrangements, the proposed approaches for implementing the main phase of HKGP, clinical research projects under the theme of "Genomics and Precision Health", and the data sharing and publication policy of HKGI.

## 科學諮詢委員會

### 成員

召集人：	盧煜明教授, SBS, JP
非官方成員：	陳志峰教授 關慧敏醫生 劉澤星教授, BBS, JP 梁子宇教授 梁德楊教授 馬紹鈞醫生 莫樹錦教授, BBS 沈伯松教授, JP 楊夢甦教授
官方成員：	醫務衛生局代表 衛生署代表

### 職權範圍

1. 考慮、建議及批准基因組計劃的臨床、實驗室及研究規程。
2. 就有效實施基因組計劃所需的相關最新科技，向董事局提出建議。
3. 決定基因組計劃主階段的研究重點。
4. 按董事局要求，在遺傳學及基因組學方面的科學問題上提供意見，以促進基因組醫學在香港的發展。

### 工作重點

自成立以來，科學諮詢委員會舉行了五次會議，商討及考慮基因組計劃先導階段和主階段的規劃和實施事宜，以及所涉及的科學問題。其中包括基因組計劃的研究規程、基因組計劃先導階段所需臨床資料涵蓋的種類（即未能確診病症及與遺傳有關的癌症）、報告主要研究結果的原則、跨專業團隊會議的安排、實施基因組計劃主階段的建議方法、在「基因組學及精準醫學」主題下的臨床研究項目，以及基因組中心的數據共享及出版政策。

## Governance Structure 管治架構

### Finance and Administrative Committee

#### Membership

Convenor:	Ms Ivy CHEUNG Wing-han
Non-official Members:	Mr Andrew FUNG Hau-chung, BBS, JP Mr LAI Kam-tong Ms Adelaide YU Hoi-man
Official Member:	Representative from the Health Bureau

#### Terms of Reference

1. To advise on the overall policies and procedures relating to financial, human resources and administrative matters of the HKGI.
2. To review and oversee the annual plan, budget and financial statements of the HKGI.
3. To review and make recommendations on the HKGI's organisation structure and level of staff compensation and benefits.
4. To advise pertaining to administrative matters, including procurement, legal, insurance on the HKGI's corporate services.
5. To consider any other finance and administrative matters of the HKGI.

#### Focus of Work

Since its inception, the Finance and Administrative Committee conducted a total of seven meetings to ensure proper stewardship and effective use of financial and manpower resources, and to review various finance and administration related matters. The Committee considered and endorsed HKGI's Standing Orders, staff structure and remuneration, employee handbook, appointment of service providers for publicity and promotion services, bioinformatics platform and sequencing services; guidelines and policies on procurement, delegation of authority, records management and information technology; and Estimates of Income and Expenditure for 2021-22 and 2022-23. It also received quarterly updates on HKGI's staff recruitment and organisation structure, financial reports, and expenditure summary of the PCs.

### 財務及行政委員會

#### 成員

召集人：	張穎嫻女士
非官方成員：	馮孝忠先生, BBS, JP 黎鑑棠先生 俞海珉女士
官方成員：	醫務衛生局代表

#### 職權範圍

1. 就基因組中心有關財務、人力資源及行政事宜的整體政策及程序提供意見。
2. 檢視及監督基因組中心的年度計劃、預算及財務報表。
3. 檢視基因組中心的組織架構，以及員工薪酬和福利水平，並提出建議。
4. 就基因組中心企業服務相關的採購、法律及保險等涉及行政事宜提供意見。
5. 審視基因組中心任何其他財務及行政事宜。

#### 工作重點

自成立以來，財務及行政委員會共舉行七次會議，以確保妥善管理及有效運用財務和人力資源，並審視各項和財務及行政相關的事宜。委員會審議及批准了基因組中心的議事規則；員工架構及薪酬水平；僱員手冊；宣傳推廣服務、生物信息平台及測序服務供應商的聘約；採購、授權、檔案管理及資訊科技的指引及政策；以及2021-22年及2022-23年的收支預算。委員會亦審視了有關基因組中心員工招聘及組織架構、財務報告及夥伴中心開支摘要的季度更新資料。

## Communication and Education Committee

### Membership

Convenor:	Mr Ray CHAN Chin-ching
Non-official Members:	Mr Stephen CHUNG Chun-kit Dr Wendy LAM Wing-tak Mr Tim PANG Hung-cheong Ms Leona WONG Nga-lai
Official Member:	Representative from the Health Bureau

### Terms of Reference

1. To advise on the overall strategy and value proposition of HKGP relating to publicity and education matters of the HKGI.
2. To make recommendations to the Board of Directors' of HKGI of the appointment of the publicity and social media consultant(s).
3. To review and oversee the HKGI's branding, communications, publicity activities and key messages delivered to the public, including the awareness, clinical benefits and data privacy issues of HKGP.
4. To review and oversee a dedicated website and social media platforms with creative design in the promotion and public education on HKGP.
5. To consider any other publicity and education matters of HKGI.

### Focus of Work

The Communication and Education Committee had held six meetings since its formation to advise HKGI on its publicity and education initiatives. Major matters the Committee deliberated included the findings of the focus groups conducted with potential HKGP participants and the branding proposal for HKGI and HKGP. The Committee also discussed the creative concepts, promotional copy and applications of a wide range of printed and multimedia publicity materials for public education and patient recruitment. They included the HKGP information booklet, leaflet, poster and souvenirs for patients, as well as over a dozen introductory videos and animations that promote HKGP, HKGI and the significance of genomic medicine. In addition, the Committee provided advice on HKGI's website design and strategies on strengthening its online presence. It also endorsed HKGI's "2022-23 Publicity Plan" and received operational updates on HKGI's corporate communication projects covering corporate publications, media activities and engagement events.

## 傳訊及教育委員會

### 成員

召集人：	陳展程先生
非官方成員：	鍾振傑先生 藍詠德博士 彭鴻昌先生 黃雅麗女士
官方成員：	醫務衛生局代表

### 職權範圍

1. 就基因組計劃與基因組中心宣傳及教育事宜有關的整體策略及價值定位，提供意見。
2. 就聘任宣傳及社交媒體顧問，向基因組中心董事局提出建議。
3. 檢視及監察基因組中心的品牌推廣、傳訊、宣傳活動及向公眾傳遞的主要信息，包括基因組計劃的認知度、臨床效益及資料私隱事宜。
4. 檢視及監察基因組計劃的創意設計、網站及社交媒體平台，以進行宣傳及公眾教育。
5. 審視基因組中心任何其他宣傳及教育事宜。

### 工作重點

傳訊及教育委員會自成立以來舉行了六次會議，就基因組中心的宣傳及公眾教育工作提供建議。委員會曾討論的主要事項包括：針對基因組計劃潛在參加者所進行的聚焦小組調研結果，以及有關基因組中心及基因組計劃品牌定位的提案。此外，委員會亦就基因組中心為公眾教育及招募病人而製作的一系列宣傳刊物及多媒體影片進行討論，包括其中的創作概念、宣傳文案及推廣應用。該等資訊包括為病人提供的基因組計劃小冊子、傳單、海報及紀念品，以及逾十部影片及動畫，以宣傳基因組計劃、基因組中心的工作及基因組醫學的重要性。委員亦就基因組中心的網站設計及其網絡宣傳策略提出建議。除上列所述，委員會亦審閱了基因組中心「2022-23年宣傳計劃」，並聽取了基因組中心在企業傳訊方面的工作匯報，涵蓋刊物出版、媒體活動及廣泛接觸各界的活動。

## Governance Structure 管治架構

### Audit and Risk Committee

#### Membership

Convenor:	Dr Isabella LIU Fang-chun
Non-official Members:	Dr KAM Pok-man, BBS Mrs Lesley WONG CHUI Yue-chue, SBS
Official Member:	Representative from the Health Bureau

#### Terms of Reference

1. To review and monitor the overall effectiveness of the HKGI's internal control procedures and risk management systems and make recommendations to the HKGI as and when necessary.
2. To make recommendations to the Board of the appointment, re-appointment and removal of the external auditor.
3. To review the findings of the external auditor and oversee the implementation of their recommendations.
4. To consider the findings of major investigations of internal control matters as delegated by the Board or on its own initiative.
5. To consider any other audit matters of the HKGI.

#### Focus of Work

Since its inception, the Audit and Risk Committee had conducted five regular meetings with some other businesses transacted by circulation. The Committee exercised active oversight of the internal audit function of HKGI, considered matters related to the audit of HKGI's financial statements, and oversaw the effectiveness of risk management and internal controls at HKGI. In regard to HKGI's internal audit function, the Committee considered and approved HKGI's Internal Audit Charter, the internal audit plans for 2021-22 and 2022-23, and received progress reports on audit results of HKGI's operations, including its human resources management and controls, information technology general security and controls, procurement as well as fixed assets management. Jointly with the Finance and Administrative Committee, the Committee reviewed and endorsed HKGI's audited financial statements for 2020-21 and 2021-22 prepared by the external auditor. For risk management, the Committee considered and approved HKGI's enterprise risk management framework and its 2022-23 Enterprise Risk Management Plan. It also oversaw implementation of HKGI's enterprise risk management systems, focusing on the planned mitigation actions for the top ten risks identified.

### 審計及風險委員會

#### 成員

召集人：	劉芳君博士
非官方成員：	甘博文博士, BBS 黃徐玉娟女士, SBS
官方成員：	醫務衛生局代表

#### 職權範圍

1. 檢視及監察基因組中心內部控制程序及風險管理系統的整體成效，並在必要時向基因組中心提出建議。
2. 就任命、重新任命及罷免外聘核數師向董事局提出建議。
3. 檢視外聘核數師提交的帳目，並監督其建議實施情況。
4. 按董事局授權或主動審議有關內部控制事宜的主要調查結果。
5. 審視基因組中心任何其他審計事宜。

#### 工作重點

自成立以來，審計及風險委員會舉行了五次定期會議，部分其他事務以書面傳閱方式處理。委員會積極監督基因組中心的內部審計職能，考慮與基因組中心財務報表審計有關的事宜，並監督基因組中心風險管理及內部控制的成效。就基因組中心內部審計職能而言，委員會審議及批准了基因組中心的內部審計章程、2021-22年及2022-23年的內部審計計劃，並接收有關基因組中心業務營運的審計結果進度報告，包括人力資源管理及控制、資訊科技的一般安全及控制、採購，以及固定資產管理。委員會與財務及行政委員會共同審視及批准了由外聘核數師編製的基因組中心2020-21年及2021-22年經審計財務報表。就風險管理而言，委員會審議及通過了基因組中心的企業風險管理框架及2022-23年企業風險管理計劃。此外，委員會亦監督基因組中心實施企業風險管理制度，主要集中於十大風險的緩解行動。

## Executive Management

The executives are charged by the Board of Directors with the responsibility of managing and administering the day-to-day business and operations of HKGI. To ensure that the management can discharge duties in an effective and efficient manner, the Board of Directors has set out clear delegated authority, directions, policies, and guidelines for the executives. Regular reports on the progress of agreed performance targets were presented to the Board.

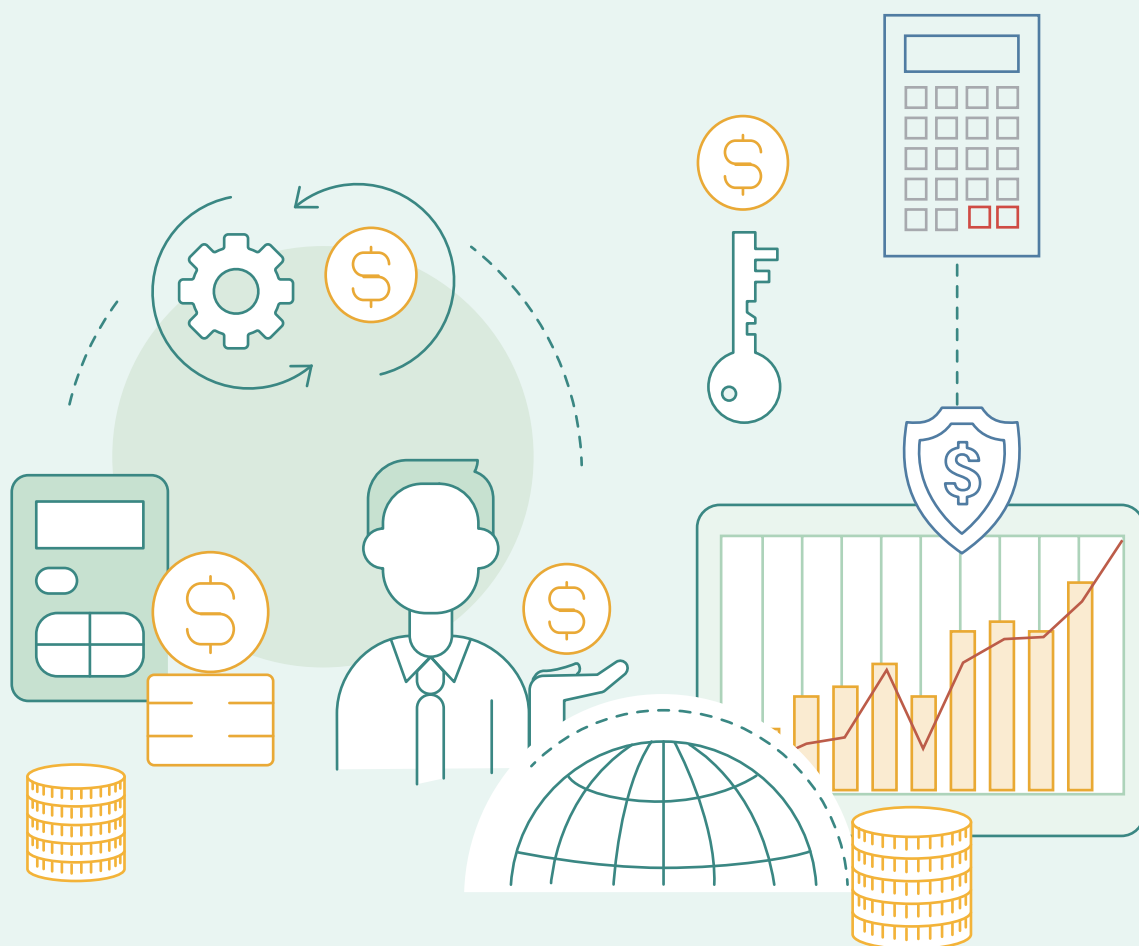
## 管理團隊

董事局委派主管級人員負責掌管和管理基因組中心的日常業務及營運。為確保管理層能夠有效且高效率履行職責，董事局已為行政人員制訂清晰的授權、策略方向、政策及指引。管理層會定期向董事局提交議定目標的進度報告。



# Financial Report

## 財務報告



The Hong Kong Genome Institute (HKGI) is a company incorporated in Hong Kong limited by guarantee and wholly-owned by the HKSAR Government. HKGI entered into a Memorandum of Administrative Arrangements (MAA) with the HKSAR Government in May 2021. The MAA provides the framework for the relationship between the HKSAR Government and HKGI, and sets out the responsibilities of both parties.

The principal activity of HKGI is to implement the Hong Kong Genome Project (HKGP), which 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. In February 2021, HKGI entered into Memorandum of Arrangements with each of the three Partnering Centres (PCs) operated by the Hospital Authority at the Hong Kong Children's Hospital, The Chinese University of Hong Kong at the Prince of Wales Hospital and The University of Hong Kong at the Queen Mary Hospital to provide HKGI with clinical support for the implementation of HKGP.

For the financial year 2021-22, the highlights were as follows:

- (a) Recurrent subvention of HK\$109,866,000 was received for the funding of personnel emolument and other operating charges. In addition, HK\$9,512,853 of the recurrent subvention for the PCs included in deferred income in previous year was recognised as income which corresponded with the related expenditure of the PCs during the financial year.
- (b) Non-recurrent subvention of HK\$34,184,217 was received for the bioinformatics services and sequencing services, of which HK\$15,779,492 was recognised as income during the financial year which matched with expense payments to the service providers. The balance of non-recurrent subvention was recorded as deferred income in the statement of financial position.
- (c) Capital subvention of HK\$9,934,747 was received for the procurement of laboratory equipment and peripheral items, and HK\$218,981 was recognised as income which represented the depreciation charge on those non-current assets during the financial year. The balance of capital subvention was recorded as deferred income in the statement of financial position.

香港基因組中心(基因組中心)為一家於香港註冊成立的擔保有限公司，由特區政府全資擁有。基因組中心於2021年5月與特區政府訂立《行政安排備忘錄》，為特區政府與基因組中心之間的關係提供框架，並載列雙方的職責。

基因組中心的主要工作為推行「香港基因組計劃」(基因組計劃)，該計劃為建立本地人口的基因組數據庫、人才庫，以及基因組測序設施和規程的催化劑項目。於2021年2月，基因組中心與醫院管理局／香港兒童醫院、香港中文大學／威爾斯親王醫院及香港大學／瑪麗醫院的三家夥伴中心訂立《安排備忘錄》，為基因組中心推行基因組計劃提供臨床支援。

2021-22財政年度概要如下：

- (a) 就支付人員薪酬及其他營運費用收取經常性補助109,866,000港元。此外，上年度計入遞延收入的9,512,853港元的夥伴中心經常性補助於本年度確認為收入，與財政年度內的夥伴中心相關開支一致。
- (b) 就生物信息學服務及測序服務收取非經常性補助34,184,217港元。其中15,779,492港元於本財政年度確認為收入，與向服務供應商支付的費用相符。非經常性補助結餘金額於財務狀況表入賬為遞延收入。
- (c) 就採購實驗室設備及周邊設備收取資本補助9,934,747港元，其中218,981港元已確認為收入，為該等非流動資產於本財政年度的折舊費用。資本補助結餘金額於財務狀況表入賬為遞延收入。

- (d) After netting off the expenditure items and depreciation charges, the surplus and total comprehensive income for the year ended 31 March 2022 was HK\$42,336,139.
- (e) As at 31 March 2022, the non-current assets of property, plant and equipment and rights-of-use assets were HK\$34,916,727 and HK\$14,071,944 respectively. The net current assets included inventories of HK\$6,032,532, bank balances of HK\$34,032,716 and payables and accruals of HK\$9,750,287. The accumulated fund was HK\$58,311,548.

The financial statements of HKGI for the year ended 31 March 2022 had been prepared in accordance with Hong Kong Financial Reporting Standards issued by the Hong Kong Institute of Certified Public Accountants, accounting principles generally accepted in Hong Kong and the Companies Ordinance (Cap. 622). They had been approved by the Board of Directors of HKGI on 20 June 2022 and audited by the independent auditors, Ernst & Young with an unqualified audit opinion. An extract of the Statement of Income and Expenditure and Other Comprehensive Income and the Statement of Financial Position are set out on pages 111-112.

Notes:

The financial information relating to the year ended 31 March 2022 and the period from 7 May 2020 (date of incorporation) to 31 March 2021 included on pages 111-112 to this annual report are not the Company's statutory annual financial statements for the year/period. Further information relating to those statutory financial statements required to be disclosed in accordance with section 436 of the Companies Ordinance (Cap. 622) is as follows:

The Company has delivered those financial statements to the Registrar of Companies as required by section 662(3) of, and Part 3 of Schedule 6 to, the Companies Ordinance (Cap. 622).

The Company's auditor, Ernst & Young, has reported on those financial statements. The auditor's reports were unqualified; did not include a reference to any matters to which the auditor drew attention by way of emphasis without qualifying its reports; and did not contain a statement under sections 406(2), 407(2) or (3) of the Companies Ordinance (Cap. 622).

- (d) 經扣除開支項目及折舊費用後，截至2022年3月31日止年度的盈餘及全面收益總額為42,336,139港元。
- (e) 於2022年3月31日，非流動資產內物業、廠房及設備和使用權資產分別為34,916,727港元及14,071,944港元。流動資產淨額包括存貨6,032,532港元、銀行結餘34,032,716港元，以及應付款項和應計費用9,750,287港元。累計資金為58,311,548港元。

基因組中心截至2022年3月31日止年度的財務報表乃根據香港會計師公會頒佈的香港財務報告準則、香港公認會計原則及《公司條例》(第622章)編製。該等財務報表已於2022年6月20日獲基因組中心董事局批准，並經由獨立核數師安永會計師事務所審核，及獲發無保留審計意見書。收支及其他全面收益表，以及財務狀況表的摘錄載於第111至112頁。

附註：

本年報第111至112頁截至2022年3月31日止年度及由2020年5月7日(註冊成立日期)至2021年3月31日的財務資料，並非本公司於該年度/期間的法定財務報表。有關該等法定財務報表須根據《公司條例》(第622章)第436條作進一步披露的資料如下：

本公司已根據《公司條例》(第622章)第662(3)條及附表6第3部份的規定，向公司註冊處處長遞交財務報表。

本公司的核數師安永會計師事務所已就財務報表作出匯報。該核數師報告並無保留意見；並不包括核數師在不作保留意見的情況下，以強調方式提述需予注意的任何事宜；亦無載有按《公司條例》(第622章)第406(2)、407(2)或(3)條所指的陳述。

# Statement of Income and Expenditure and Other Comprehensive Income 收支及其他全面收益表

For the year ended 31 March 2022

截至2022年3月31日止年度

		Year ended 31 March 2022  截至2022年 3月31日止年度	Period from 7 May 2020 (date of incorporation) to 31 March 2021  2020年5月7日(註冊成 立日期)至2021年 3月31日
		HK\$ 港元	HK\$ 港元
<b>INCOME</b>	<b>收入</b>		
Recurrent subvention	經常性補助	109,866,000	99,953,000
Less: Amount deferred	減：遞延金額	–	(75,000,000)
Add: Release of deferred income	加：遞延收益攤銷	9,512,853	–
		119,378,853	24,953,000
Non-recurrent subvention	非經常性補助	15,779,492	–
Capital subvention	資金補助	218,981	–
		135,377,326	24,953,000
Bank interest income	銀行利息收入	14,523	986
Total income	收入總額	135,391,849	24,953,986
<b>EXPENDITURE</b>	<b>開支</b>		
Recurrent expenditure	經常性開支		
Personnel emoluments	人員薪酬	(37,352,792)	(3,381,715)
Partnering Centres expenses	夥伴中心開支	(9,512,853)	–
Other operating charges	其他營運費用	(18,539,292)	(5,568,252)
		(65,404,937)	(8,949,967)
Depreciation	折舊		
Property, plant and equipment	物業、廠房及設備	(5,588,535)	(28,610)
Right-of-use asset	使用權資產	(5,096,455)	–
Loss on derecognition of lease liability	租賃負債終止確認虧損	(587,599)	–
Finance cost on lease liability	租賃負債的融資成本	(598,692)	–
Non-recurrent expenditure	非經常性開支	(15,779,492)	–
Total expenditure	開支總額	(93,055,710)	(8,978,577)
<b>SURPLUS AND TOTAL COMPREHENSIVE INCOME FOR THE YEAR/PERIOD</b>	<b>年內／期內盈餘及全面 收益總額</b>	42,336,139	15,975,409

# Statement of Financial Position

## 財務狀況表

As at 31 March 2022

於2022年3月31日

		2022 HK\$ 港元	2021 HK\$ 港元
<b>NON-CURRENT ASSETS</b>	<b>非流動資產</b>		
Property, plant and equipment	物業、廠房及設備	34,916,727	915,846
Right-of-use asset	使用權資產	14,071,944	-
Prepayments	預付款項	3,935,123	-
Total non-current assets	非流動資產總額	52,923,794	915,846
<b>CURRENT ASSETS</b>	<b>流動資產</b>		
Inventories	存貨	6,032,532	-
Prepayments and deposits	預付款及按金	72,823,421	75,938,263
Bank balances	銀行結餘	34,032,716	16,292,605
Total current assets	流動資產總額	112,888,669	92,230,868
<b>CURRENT LIABILITIES</b>	<b>流動負債</b>		
Other payables and accruals	其他應付款項及應計費用	9,750,287	2,171,305
Deferred income – Partnering Centres	遞延收入－夥伴中心	65,487,147	75,000,000
Deferred income – Non-recurrent subvention	遞延收入－非經常性補助	18,404,725	-
Deferred income – Capital subvention	遞延收入－資本補助	1,898,186	-
Total current liabilities	流動負債總額	95,540,345	77,171,305
<b>NET CURRENT ASSETS</b>	<b>流動資產淨額</b>	17,348,324	15,059,563
<b>TOTAL ASSETS LESS CURRENT LIABILITIES</b>	<b>資產總額減流動負債</b>	70,272,118	15,975,409
<b>NON-CURRENT LIABILITIES</b>	<b>非流動負債</b>		
Deferred income – Capital subvention	遞延收入－資本補助	7,817,580	-
Provision for reinstatement costs	修復成本撥備	4,142,990	-
Total non-current liabilities	非流動負債總額	11,960,570	-
Net assets	資產淨額	58,311,548	15,975,409
<b>FUNDS</b>	<b>資金</b>		
Accumulated fund	累計資金	58,311,548	15,975,409





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