Our Genomic Advocates

基因組醫學新力量

Nurturing Genomic Medicine Excellence for the Future

Encouraging more and more clinicians and researchers to engage in genomic medicine is a key priority in advancing precision medicine. Equally important is developing the next generation of professionals.

The stories you are about to discover showcase how HKGI's initiatives have galvanised medical professionals and young people to advance the horizons of genomic medicine, delivering meaningful patient outcomes while shaping the future of healthcare.

Three healthcare professionals will share new perspectives on diagnosis, treatment and prevention that genomic medicine brings to clinical practice. They are recipients of the "HKAM-HKGI Research Excellence Grants in Genomic Medicine" and "HKCP-HKGI Overseas Training Scholarship and Training Grant for Excellence in Genomic Medicine", which support their research and study in the clinical applications of genomic medicine.

Also spotlighted are three secondary and university students who participated in HKGI's internship and attachment programmes. They will share how these experiences allowed them to witness the immense potential of genomic medicine in clinical settings, and how these opportunities have inspired their future directions.

培育卓越人才 推動基因組醫學

鼓勵更多臨床及研究人員投身基因組醫學領域，是推動精準醫學發展的關鍵所在。與此同時，培育新一代專業人才同樣重要。

以下的專題故事展現基因組中心如何透過各項計劃，激勵醫療專業人士與年輕一代拓展基因組醫學疆界，不但為病人帶來實質療效，同時引領醫療服務的創新發展。

三位醫療專業人士將從「診斷、治療、預防」方面分享基因組醫學在臨床上帶來的新視野。他們分別獲得基因組中心與香港醫學專科學院共同設立的「基因組醫學卓越研究獎」，以及基因組中心與香港內科醫學院共同設立的「基因組醫學海外及本地進修獎學金」，支持他們深入研究基因組醫學的臨床應用。

另外，專題亦會介紹三位曾參與基因組中心實習及體驗計劃的中學生及大學生，他們將分享如何透過這些計劃親身見證基因組醫學在臨床上的無限潛力，以及這些經歷如何啟發他們的未來發展方向。

Young Mind Bright Aspiration
青年遠志 點亮前路

> This experience has solidified my desire to pursue genetics and genomics – as a career and as a way to change lives.
>
> 這段經歷更堅定了我將來投身遺傳學及基因組學的志向。不僅以此為業，更盼能以生命改變生命。

Natalie Wan 温曉潀 | Participant of HKGI 2025 Student Attachment Programme 香港基因組中心2025年學生體驗計劃參加者

Natalie has always been drawn to the world of life sciences. Her fascination with genomics was sparked after undertaking a course in genomic technologies for treating acute blood cancer. After that, she joined the Student Attachment Programme at Hong Kong Genome Institute (HKGI), receiving the opportunity to explore genomics in action alongside other students. Her journey reflects HKGI's commitment to nurturing talent and cultivating scientific curiosity at every age.

At HKGI, Natalie was able to put theory into practice, gaining hands-on experience with cutting-edge genomic technologies with guidance from HKGI's experts. She witnessed the applications of genomic medicine and learnt how this new field of modern medicine is having life-changing impacts. Together with other students, she immersed herself in a research project on nonsense-mediated mRNA decay, exploring how genetic variants might align with patient symptoms. As part of the Attachment Programme, Natalie had the invaluable opportunity to shadow healthcare professionals and observe the collaborative environment at HKGI and its partnering centre.

Natalie一直對生命科學領域充滿熱忱。她對基因組學的興趣源於一次因緣際會下，接觸有關運用基因組技術治療急性血癌的課程。其後，她參加了香港基因組中心（基因組中心）的學生體驗計劃，與其他學員共同探索基因組學的實際應用。她的學習歷程正正體現基因組中心致力培育各年齡層的人才、激發新一代對科學熱誠的堅定承諾。

在基因組中心，Natalie有機會在中心專家的指導下認識先進的基因組技術。她不但見證基因組醫學如何應用於臨床護理，亦了解到這個現代醫學的新興領域如何為生命帶來深遠的影響。她與其他學員一同了解「無義介導的mRNA降解」研究項目，探討基因變異與病人症狀的關聯。作為計劃的一部分，Natalie更獲得寶貴的機會，跟隨專業醫護人員實地視察，親身體驗基因組中心及其夥伴中心的協作環境。

The Human Side of Genomic Medicine

Genetic counselling is a crucial part of clinical practice in genomic medicine, and for Natalie, attending these sessions was nothing short of transformative. During one particular session, she observed a mother discussing test results for her 13-year-old son with a genetic disease. The counsellor patiently broke down complex information and procedures into simple terms, empowering the mother to understand her son's condition and make informed decisions about his care. That moment crystallised Natalie's ambition.

"I was struck by the mother's determination to ask many complex questions. She conducted thorough research and gathered a wealth of information through her own initiative," Natalie recalled. "She was actively seeking potential clinical trials so her son could receive the best possible treatment."

The mother's wish for more breakthroughs in genomics to help children with similar conditions also left a profound impression on Natalie. "It was such a powerful reminder of the importance of public awareness surrounding genetic diseases. This whole session was incredibly inspiring, heartwarming and admirable – a truly unforgettable experience for me," she said.

Looking to the Future

Alongside gaining a comprehensive perspective on genomic medicine and its clinical applications, Natalie believed her experiences at HKGI have solidified her decision to pursue genetics and genomics in future. Although her journey in genomic medicine has just begun, her passion exemplifies how nurturing young scientific minds today fosters healthcare innovations tomorrow.

基因組醫學 以人為本

遺傳輔導是基因組醫學在臨床實踐的重要一環，對Natalie來說，參與遺傳輔導的觀摩環節，更是一次意義非凡的啟發。在一次輔導過程中，她觀察到遺傳輔導員與一位母親討論其13歲、患有遺傳病的兒子的檢測結果。當時，輔導員耐心講解，深入淺出地解說複雜的資訊與醫療程序，讓這位母親能透徹理解兒子的病情，從而為他的護理作出明智的抉擇。這次觀察經歷，讓Natalie的志向變得更為清晰明確。

Natalie憶述道：「那位母親的求知精神讓我印象尤深。為了替兒子尋找最佳治療方案，她不僅進行了深入的資料搜集，還積極尋找有機會參與的臨床試驗，探索各種可能性。」

這位母親期盼基因組學能取得更多突破，以幫助更多類似病童。這份盼望，同樣使Natalie深受觸動。「這讓我真切體會到加強公眾認識遺傳病的重要性。整個輔導環節充滿真誠的互動，亦令我對遺傳輔導員的專業更加敬佩，絕對是一次難忘的經歷。」Natalie回憶道。

展望未來

在基因組中心的體驗不但讓Natalie對基因組醫學及相關臨床應用有更全面的認識，更堅定了她未來投身遺傳學和基因組學的志向。Natalie在基因組醫學的旅程才剛起步，而她對這個範疇的熱忱，充分體現出悉心啟迪科研青苗對成就明日創新醫療的重要。

From Learning to Leading 承傳學識 凝聚力量

> I was captivated by HKGI's detective-like work in genome curation. Learning how genomic medicine informs the diagnosis of rare diseases inspired me to establish a medical student association focusing on genetics and genomics, which acts as a platform for like-minded and passionate students to connect, learn, and advocate for those affected by rare conditions and other genetic disorders.
>
> 基因組中心在分析基因組數據方面如偵探般的工作深深吸引了我。了解到基因組醫學能為罕見病帶來診斷希望後，我決心創立一個專注於遺傳學與基因組學的醫學生組織，凝聚志同道合的同學交流知識，並為罕見病及其他遺傳病患者發聲。

Safari Chiu 趙敏玳
Medical Student at the Chinese University of Hong Kong
香港中文大學醫學院學生
HKGI 2023 Summer Intern
香港基因組中心2023年暑期實習生

Safari's fascination with genomics began back in secondary school when she first learnt that merely four simple letters – A, T, C, and G – underlie the complexity and uniqueness of all living organisms. From her medical studies, she understood that knowledge about clinical genetics and genomics is applicable and important in virtually all medical specialties in this era of personalised medicine. Thus, she was motivated to pursue an internship at HKGI in 2023, where she explored how genomic findings translated from bench to bedside. This aligns with HKGI's commitment in nurturing future healthcare practitioners, who understand both the science and human impact of genomic medicine.

When Textbooks Meet Clinical Application

This internship provided Safari a comprehensive exposure to genomic medicine, from hands-on lab experience to participation in genome curation meetings. A week-long shadowing at the Prince of Wales Hospital further allowed her to gain insight into complete patient journey in Hong Kong Genome Project (HKGP), from informed consent to sample collection, analysis, and results reporting. Moreover, through working on her group project on pharmacogenomics, she understood how precision medicine could advance treatment, not just for rare genetic diseases but also for common conditions. "This internship provided me the macroscopic view of how a large-scale population health project and biobank like HKGP is executed, as well as the microscopic view of how HKGP gives hope and treatment guidance for individual patients and their families," Safari reflected.

Bridging Science with Compassion

Inspired by the potential of genomic medicine and the dedication of HKGI experts to "orphan disease" advocacy, Safari co-founded the Hong Kong Medical Genetics and Genomics Student Society (HKGeneSoc) to create learning opportunities for medical students, such as hosting symposiums and clinical teaching sessions, as well as attending academic conferences in Chinese Mainland and overseas. Through this platform, Safari and her peers have also collaborated with NGOs and charities to support individuals and families navigating rare diseases, from public advocation on rare diseases in a podcast series to representing patients at clinics with visiting overseas experts, applying the patient-centred approach she observed during her time at HKGI.

Drawing wisdom from Hippocrates – "Cure sometimes, treat often, comfort always" – Safari viewed genomic medicine as a bridge between innovation and compassionate care. "While there may not always be a cure or even an answer for every case, we can reassure patients and families that we are allies on the same boat navigating the unknown waters with them. No matter how complex a case may be, always remember that we are treating people as a whole and not merely medical conditions," she concluded.

Future Visions

As Safari continues her journey to becoming a doctor, her experience at HKGI and understanding of genomic medicine continue to guide her approach to patient care and advocacy. Inspired by HKGI's vision of cultivating not only knowledgeable practitioners but also community leaders who extend its mission beyond their own practice, Safari is determined to carry these values forward.

Safari對基因組學的熱忱，始於中學時期。當時，她初次了解到僅僅A、T、C、G四個代表DNA鹼基的字母，便奠定了生物世界的複雜多樣與獨一無二。及至修讀醫科，她了解到在個人化醫療時代，遺傳學和基因組學可廣泛應用於所有醫學專科，並具重要意義。這份認知驅使她於2023年參加香港基因組中心的暑期實習計劃，親身探索基因組學的發現如何從實驗室科研轉化為臨床應用。而基因組中心正是透過這樣的實習體驗，致力培育下一代醫療專業人才，使他們不僅掌握扎實的科學知識，同時能體會基因組醫學對病人及其家庭的深遠意義。

臨床實踐 學以致用

這次實習為Safari提供了全面的基因組醫學視野，從實驗室的實際操作，到參與基因組數據分析的會議。其中，在威爾斯親王醫院為期一星期的實地跟隨觀察，更讓她進一步了解到香港基因組計劃(基因組計劃)中病人招募的完整流程，包括知情同意、收集樣本、分析以及結果報告。此外，透過參與藥物基因組學的小組研究項目，她亦了解到精準醫學如何推動治療方案的進步，其應用不僅限於罕見遺傳病，更能惠及常見疾病。Safari表示：「這次實習既讓我從宏觀角度了解到像基因組計劃等全港性大型計劃如何運作，同時亦從微觀角度見證這個計劃如何為每位病人及其家庭帶來希望並提供治療方向。」

科研為媒 醫者仁心

有見基因組醫學潛力無限，並受基因組中心專家對罕見病的關注所啟發，Safari與一眾志同道合的同學成立「香港遺傳學學生協會」，為醫學生創造更多學習機會，如舉辦專題研討會、臨床教學環節，以及參與內地與海外的學術會議等。透過這個平台，他們與非政府組織及慈善團體合作，支援罕見病患者和家庭。從製作網上電台節目提升公眾認知，到陪同病人會見訪港的海外專家，Safari在這些行動中處處實踐着從基因組中心實習時所學到的「以病人為本」理念。

Safari從希波克拉底的智慧箴言「時而治癒，時常醫治，時刻安慰」中得到啟發，視基因組醫學為創新思維與人文關懷之間的橋樑。她總結道：「縱使並非每一個案都能夠找到治癒方法或答案，我們仍可讓病人與家屬感受到，我們是與他們同行的夥伴，一起探索未知領域。無論病情多複雜，我們都需時刻銘記，我們治療的是一個完整的人，而非單純疾病。」她總結道。

實踐理想 未來可期

如今Safari正逐步實現成為醫生的夢想，而她於基因組中心的學習及對基因組醫學的深刻理解，將繼續指引她向前邁進，實踐病人照護和倡導工作。基因組中心不但致力培育學識淵博的醫護人員，更期望他們將使命延伸至專業以外，成為惠澤社群的領袖。Safari亦深受啟發，決心身體力行，將這份信念延續下去。

Our Genomic Advocates
基因組醫學新力量

Making a Difference with Genomic Medicine

基因組醫學 惠及病人

> The HKGI internship guided my career path towards becoming a clinician and making a difference for patients with undiagnosed diseases.
>
> 在基因組中心實習的經歷為我的職涯指明方向，促使我投身臨床工作，致力協助未能確診病症的病人。

Dr Ng Hoi Chak 吳鎧澤

Resident
駐院醫生

HKGI 2022 Summer Intern
香港基因組中心2022年暑期實習生

Dr Ng's journey into genomic medicine began during his fourth year of medical school when he encountered a baby suffering from an unexplained condition. Despite exhaustive investigations, no diagnosis emerged, until genomic testing supplied the pivotal answer that made targeted treatment possible. The family's relief deeply moved Dr Ng, and the episode convinced him of the power of genomic medicine to solve clinical mysteries.

Motivated by a desire to supplement what he learnt during his Master of Public Health, Dr Ng joined HKGI's internship in the summer of 2022 to explore the interplay between genomics, clinical medicine, and population health. The HKGI internship fostered his development into a clinician who not only understands genomic medicine but also its broader implications for healthcare.

吳醫生接觸基因組醫學的契機，始於他在醫學院四年級時，遇到一名罹患不明病症的嬰兒。即使經過詳盡檢查，醫療團隊仍然無法確診，直至基因組測序結果揭示病因，才得以為其制訂個人化治療方案。測序結果不但使嬰兒家屬如釋重負，更深深觸動了當時仍是醫科學生的吳醫生，讓他體會到基因組醫學在破解臨床確診疑難方面的龐大潛力。

為了將公共衛生碩士課程所學付諸實踐，並探索更深層次的知識，吳醫生於2022年夏天參加了基因組中心的實習計劃，深入鑽研基因組學、臨床醫學與人口健康三者之間的關係。這次實習，不僅加深了吳醫生對基因組醫學的認識，更助他了解到相關技術和臨床應用對整體醫療體系的深遠影響，為他從醫之路奠定了穩固基石。

A Deep Dive into Genomics

Through participation in HKGI research, Dr Ng gained deeper insight into HKGI's broader public health mission, understanding how HKGI's Southern Chinese database addresses the significant burden of undiagnosed diseases by filling critical knowledge gaps in predominantly European-based genomic data.

Through learning how PCSK9 inhibitors, a new cholesterol lowering drug, were developed from studies of the rare genetic condition familial hypercholesterolemia, Dr Ng discovered that research on rare diseases can unlock benefits for the wider population.

Dr Ng's HKGI experience encompassed far more than technical skills. In addition to mastering laboratory techniques, genomic data analysis, and variant interpretation, he learnt about genetic counselling principles, explaining uncertainty to patients, and managing family expectations. He came to appreciate that genomic testing is iterative: today's "no answer" can lay the groundwork for tomorrow's breakthrough. "I find it deeply rewarding when I successfully help patients and their families navigate the evolving process of genomic testing," Dr Ng said.

Effective scientific communication also became a central part of his training. "I learnt to explain scientific concepts to patients from diverse backgrounds." Dr Ng also assisted experts in developing advocacy materials for HKGP, translating complex genomic concepts for stakeholders with varying levels of understanding. In his clinical practice, he applies these skills to empower patients, offering clarity as they navigate the complexities of genomic testing.

Looking to the Future

As a clinician, Dr Ng actively identifies patients for HKGP and bridges genomics with routine patient care. His journey exemplifies HKGI's success in cultivating professionals who apply genomics technically, communicate it effectively, and understand its broader implications, bringing genomics and medicine together to transform healthcare. By continuing to integrate genomics into everyday practice, Dr Ng is helping to ensure that tomorrow's patients will encounter fewer diagnostic dead ends and far more targeted solutions.

深耕基因組學 洞察生命意義

實習期間，透過參與基因組中心的研究項目，吳醫生對基因組中心在公共衛生領域的願景有了更深刻的領會。現時國際間的基因組數據以歐洲人為主，基因組中心建立以華南人口為基礎的基因組數據庫，致力填補這個缺口，有望為未能找到病因的患者帶來希望。

他亦從了解PCSK9抑制劑（一種新型降膽固醇藥物）的研究中獲得啟發，這項原本針對罕見遺傳病「家族性高膽固醇血症」的藥物研究，最終竟催生出治療常見高膽固醇問題的新療法。吳醫生因此領悟到，罕見病研究的成果同樣能為普羅大眾帶來福祉。

吳醫生在基因組中心實習所得，遠不止於技術層面的知識。他不僅掌握了實驗室技術，亦學習到如何分析基因組數據及詮釋基因變異，更學會遺傳輔導的基本原則，即如何向病人解釋病症的不確定性，並妥善管理家屬的期望。他亦體會到基因組測序技術是一個不斷演進的過程，今日暫無答案的疑難，或者會為未來的突破埋下伏筆。他表示：「能夠陪伴病人與家屬逐步走過基因組檢測的各個階段，令我感受到這份工作的非凡意義。」

有效的溝通技巧亦是他實習中不可或缺的一環。「我學會如何向不同背景的病人，講解深奧的科學概念。」實習期間，吳醫生協助基因組中心為基因組計劃撰寫推廣資料，學習如何將晦澀的基因組學知識，轉化為不同知識背景的持份者都能明白的內容。如今在臨床工作中，他善用這些技巧，在病人面對複雜的基因組測序流程時，為他們提供清晰指引，讓他們更有信心地應對。

開創未來

吳醫生的職業歷程，是基因組中心致力培育人才的最佳證明。在臨床上，他不僅積極識別有需要的病人並轉介他們參與基因組計劃，更致力將基因組學的知識融入日常護理。他的實踐體現了醫療專業人員的核心特質：既能精準運用基因組學技術，又能有效傳達相關知識，並切實應用，為大眾健康帶來深遠影響。吳醫生將繼續結合基因組學與醫學，期望助病人擺脫診斷困境，開創個人化精準治療的新未來。

Genomic Medicine Unlocks New Horizons in Kidney Disease Diagnosis and Treatment

基因組醫學 開拓腎病診治新視野

> The scope of genomic medicine goes far beyond diagnosing rare genetic diseases. It has enormous potential for treating common conditions such as kidney disease.
>
> 基因組醫學的應用遠不止於診斷罕見遺傳病，對診治腎病等常見疾病同樣深具潛力。

Dr Becky Ma 馬銘遙醫生

Specialist in Nephrology
腎病科專科醫生

Awardee of the 2023/24 HKCP-HKGI Overseas Training Scholarship and Training Grant for Excellence in Genomic Medicine
2023/24年度香港內科醫學院 — 香港基因組中心「基因組醫學卓越海外及本地培訓獎學金及助學金」得主

Awardee of the 2024/25 HKAM-HKGI Research Excellence Grants in Genomic Medicine
2024/25年度香港醫學專科學院 — 香港基因組中心「基因組醫學卓越研究獎」得獎學者

Kidney disease affects one in every 10 adults in Hong Kong, making it one of the most common chronic diseases. According to local data, approximately 12% of new end-stage kidney failure cases are attributed to genetic or unknown causes. “Genomic medicine helps us uncover the deeper origins of disease,” explains Dr Becky Ma.

腎臟疾病是香港常見的慢性疾病之一，每十位香港成人中就有一位患有慢性腎病。本地數據指出，末期腎衰竭的新症中，約有12%由遺傳或不明病因所致。馬銘遙醫生解釋道：「基因組醫學有助我們找出更深層次的疾病根源。」

A Lifesaver as Precision Diagnosis Reveals Disease Origins

While current kidney disease diagnosis primarily involves clinical assessment, blood and urine tests, and tissue biopsy, whole genome sequencing (WGS) has unlocked crucial diagnostic support. “Through WGS, we can identify the specific genetic variants causing kidney disease, especially important for conditions where similar clinical presentations have entirely different underlying causes,” Dr Ma noted.

The doctor recalled a case of a patient with stage-five kidney failure of unknown cause. When the patient was admitted, tissue biopsy was impossible because the kidneys had already severely atrophied. Through WGS, the patient was diagnosed with the rare hereditary condition Nephronophthisis, enabling targeted treatment. Understanding this disease led doctors to also identify the associated risk of eye complications, allowing for timely detection and treatment of retinal disease.

The Formidable Protection of Family Health Through Early Prevention and Monitoring

For families suffering from familial kidney diseases, WGS enables comprehensive prevention strategies. “Genomic sequencing helps us establish clear connections between genetic variants and kidney disease, allowing us to offer preventive monitoring for high-risk family members and intervene early to slow disease progression,” Dr Ma explained.

揭示病源 生命轉機

現時腎病的診斷方法以臨床評估、血液與尿液化驗以及活組織檢查為主，而全基因組測序可突破現有診症限制，提供關鍵支援。馬醫生指出：「透過全基因組測序，我們可以找出引致腎病的特定基因變異，對於那些臨床表徵相似但成因不同的腎病，這一點攸關重要。」

馬醫生憶述一宗病例，該名患者患上第五期腎衰竭卻病因不明，入院時由於兩邊腎臟已嚴重萎縮，無法抽取腎組織化驗。而透過全基因組測序，患者最終確診罕見遺傳病「腎消耗病」(Nephronophthisis)，得以展開針對性的治療。同時，因了解此病引致的眼部併發症風險，臨床團隊及時轉介患者治療視網膜病變，使患者得到全面診治。

及早預防監測 守護家人健康

對患有家族遺傳性腎病的家庭而言，全基因組測序技術可助他們制訂周全的預防性策略。馬醫生解釋道：「基因組測序有助我們釐清基因變異與腎病之間的明確關聯，從而為高風險家庭成員提供預防性監測，並及早介入，以延緩病情。」

She used Polycystic Kidney Disease as an example, a relatively common hereditary condition affecting approximately one in every 1,000 to 2,500 individuals. According to Dr Ma, WGS can diagnose this condition before symptoms appear. "Early diagnosis means we can immediately prescribe targeted medications such as Tolvaptan to slow cyst growth and preserve kidney function," she explained. "We can implement early prevention measures for complications such as hypertension and kidney infections, significantly improving long-term outcomes."

她以多囊性腎病為例子，這種遺傳性腎病相對常見，約每1,000至2,500人中就有一名患者。馬醫生表示，全基因組測序可在病徵出現前診斷病情。她解釋道：「及早診斷意味着我們可以立即處方特效藥物如Tolvaptan，延緩囊腫的生長速度，並保存腎臟功能。我們可及早採取預防措施，應對高血壓和腎臟感染等併發症，這將大幅改善患者的長遠預後。」

Advancing Local Genomic Medicine Research is a Gamechanger

本地研究 開創新局

Precise diagnosis requires robust genomic data, and population-specific data is indispensable. "Different ethnic groups have distinct genetic profiles, so Western research findings don't always apply to our local patients. For this reason, building a local genome database is essential for advancing precision medicine," Dr Ma emphasised. "The Hong Kong Genome Project provides genomic data specific to the Southern Chinese population, revealing unique genetic variants that help us better serve local patients' needs."

精準診斷需要穩健的基因組數據，而特定人口的數據更是不可或缺。馬醫生強調：「不同族群的基因圖譜各異，西方研究成果未必完全適用於本地患者。因此，建立本地基因組數據庫是推動精準醫學發展的重要一環。基因組計劃正正為我們提供聚焦華南地區人口的基因組數據，揭示本地特有的基因變異，有助我們針對本地患者所需。」

Dr Ma said the practical experience she gained at HKGI was transformative: "I've seen firsthand how genomic data translates into clinical applications. Through my research into genetic associations in kidney transplant medications, I've witnessed the genuine hope that genomic medicine brings to kidney disease patients. I believe that by advancing WGS capabilities, we are building a foundation for precision medicine that will benefit not only kidney transplants but also potentially extend to other fields and complex disease treatments."

馬醫生分享她在基因組中心積累的寶貴經驗：「我親身見證基因組數據如何轉化為臨床應用。透過積極研究腎臟移植藥物與基因的關聯，我看到基因組醫學為腎病患者帶來真正的希望。我堅信推動全基因組測序技術的發展，能夠為精準醫學奠下穩固基礎，其效益不僅限於腎臟移植，更有望擴展至其他領域及複雜疾病的治療。」

Precision Medication Improves Kidney Transplant Success
精準用藥提升腎臟移植成功率

For patients with end-stage kidney disease, transplantation offers the best hope. However, demand far exceeds supply. More than 2,000 patients in Hong Kong are currently awaiting kidney transplants, with fewer than 100 donors becoming available each year. Dr Ma's research focuses on personalised medication regimens that reduce rejection and side effects, thereby improving transplant outcomes.

According to Dr Ma, post-transplant patients require long-term use of the anti-rejection drug Tacrolimus (FK506). Current standard dosage has been developed primarily based on research data from Western populations, but Hong Kong patients exhibit significant genetic differences. Getting the balance right is critical, as insufficient drug levels result in rejection whereas excessive levels cause side effects such as hand tremors and diabetes.

WGS can identify the key genes that affect drug metabolism, enabling clinicians to predict individual patient responses and provide optimal dosage from the start. Dr Ma's research promises to provide evidence-based prescribing guidelines for kidney transplant medications, advancing precision medicine in organ transplantation and improving outcomes for every valuable transplant opportunity.

對於末期腎病患者來說，腎臟移植是最佳選擇。然而，全港目前有超過2,000名患者輪候腎臟移植，而每年可進行移植的捐贈者卻不足百人。馬醫生的研究聚焦於為腎臟移植患者提供個人化用藥方案，旨在減少排斥反應和副作用，從而提升器官移植的成效。

馬醫生指出，移植患者術後需要長期服用抗排斥藥物Tacrolimus（FK506），現行標準劑量主要基於西方人口的研究數據，但本港患者存在明顯的基因差異。用藥關鍵在於精確控制藥物濃度，濃度不足會導致排斥反應；過高則引發手震、糖尿病等副作用。

全基因組測序可識別影響藥物代謝的關鍵基因，有助預測個別患者的藥物反應，繼而從治療初始階段便可處方最佳劑量。馬醫生的研究可望為腎臟移植藥物提供具實證支持的處方指引，促進器官移植方面的精準醫學發展，並提升每次寶貴移植機會的成效。

Our Genomic Advocates
基因組醫學新力量

Genomic Medicine Transforms Childhood Myopia Care in Hong Kong

基因組醫學改寫港童近視照護模式

> Genomic medicine presents tremendous potential in childhood myopia research. By identifying specific risk genes, we can provide more precise prevention and management strategies for children.
>
> 基因組醫學在兒童近視研究中展現龐大潛力。透過識別特定的風險基因，我們可為兒童制訂更精準的近視預防及管理策略。

Professor Guy Chen
陳理佳教授

Specialist in Ophthalmology
眼科專科醫生

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2023/24年度香港醫學專科學院 — 香港基因組中心「基因組醫學卓越研究獎」得獎學者

Hong Kong has a high incidence of myopia. Prior to the pandemic, myopia rates among six-year-old children were approximately 12% to 15%. However, after the pandemic, the figures more than doubled to 25%, meaning that one in four Primary 1 students has myopia. "Myopia is a multifactorial disease involving both genetic and environmental factors," Professor Guy Chen explained. "Particularly for young children with myopia, genetic factors may play a more decisive role."

香港屬於近視高發病率地區，新冠疫情爆發前，六歲兒童的近視率約為12%至15%。但疫情過後，數字卻飆升一倍多至25%，即每四名小一學童中就有一人患有近視。陳理佳教授解釋道：「近視是遺傳與環境因素共同作用下的多因素疾病。對幼齡近視兒童而言，遺傳因素可能發揮更決定性的作用。」

Myopia primarily follows two inheritance patterns: single-gene and polygenic. "Single-gene inheritance can be dominant or recessive. When parents don't have myopia but a child develops severe myopia, this could be due to recessive inheritance. However, most myopia cases are a result of polygenic inheritance, where dozens or even hundreds of genes combine with environmental factors to cause myopia," said Professor Chen.

近視主要循單一基因與多基因兩種模式遺傳。陳教授表示：「單一基因遺傳有顯性或隱性之分。當父母雙方皆無近視，子女卻出現重度近視，便可能是隱性遺傳造成。但大部分近視個案源於多基因遺傳，即由數十至數百個基因與環境因素共同影響所致。」

Major Health Risks Associated with High Myopia

In addition to severely affecting vision, high myopia could trigger other serious eye diseases. "The risk of high myopia patients having retinal detachment is 100 times higher than normal people," Professor Chen said. Other common complications include macular degeneration, glaucoma, and early-onset cataracts. "High myopia patients may develop cataracts between 40 and 50 years old, 10 to 20 years earlier than average," Professor Chen added.

These complications also impact work and quality of life. Once the eyeball elongates to become myopic, it is essentially irreversible. Even after laser vision correction surgery, the risks associated with high myopia remain.

高度近視 健康隱憂

高度近視除了嚴重影響視力外，更可能引發其他嚴重眼疾。陳教授指出：「高度近視患者發生視網膜剝離的風險比常人高出100倍。」其他常見的併發症包括黃斑病變、青光眼及早發性白內障。陳教授補充：「高度近視患者可能在40至50歲之間就出現白內障，比平均發病年齡提早10至20年。」

這些併發症不僅嚴重影響視力，也會影響工作和生活質素。陳教授強調，眼球一旦拉長導致成為近視眼，基本上就無法逆轉。即使患者接受了激光矯正視力手術，也無法消除高度近視帶來的相關疾病風險。

Making Childhood Myopia a Thing of the Past

Professor Chen and his team are dedicated to researching genes related to childhood myopia, collecting genetic data from nearly 40,000 Hong Kong children. Through genome-wide association study (GWAS), they discovered four new genes associated with childhood myopia. "These childhood myopia genes were never found in adult myopia research. Each gene variant increases myopia risk by 1.2 to 1.5 times. If a child carries all four gene variants, their myopia risk increases by more than three times."

兒童近視 畫下句點

陳教授與其團隊致力研究兒童近視相關基因，並收集了近4萬名港童的基因數據。他們以全基因組關聯分析發現了四個與兒童近視相關的新基因。陳教授說：「成人近視研究從未發現這些兒童近視基因，每個基因變異都令近視風險增加1.2至1.5倍。若一名兒童同時攜帶四種基因變異，其近視風險將增加三倍以上。」

The research also found that children with high genetic risk scores and who engage in prolonged close-up work have a myopia risk increase of four to five times. “These findings enable us to formulate personalised myopia care strategies,” Professor Chen said. “High-risk children’s daily cumulative reading time should not exceed three hours, while low-risk children should not exceed four hours.”

該研究同時發現，遺傳風險評分高的兒童如果長時間進行近距離工作，其近視風險將增加四至五倍。陳教授表示：「這些發現讓我們能夠制訂個人化的近視管理方案。例如，高風險兒童每日累積閱讀的時間不應超過三小時，而低風險兒童則不應超過四小時。」

New Directions in Childhood Myopia Prevention Transform Care

For treatment, besides wearing appropriate glasses, atropine eye drops can effectively slow myopia progression. Some children even experienced myopia reversal after combining Atropine eye drops with red light therapy, according to another study conducted by Professor Chen’s team: “Some children experienced complete reversal from 100 degrees of myopia after six months of treatment, no longer needing glasses, and their eye axis also shortens.”

Professor Chen emphasised the importance of establishing local genomic databases, as different ethnic groups have varying genetic characteristics, and Western research findings may not necessarily apply to local patients. Having a local database offers valuable reference for precision medicine across specialties. “By analysing local children’s genetic data, we can more accurately identify myopia risk genes and provide more precise prevention strategies for Hong Kong children. Genomic medicine enables us to identify high-risk children early. We can adjust reading habits, implement personalised treatment plans, and reduce the likelihood of high myopia,” he said.

防範近視 照護新方

治療方面，除了配戴合適的眼鏡外，使用阿托品（Atropine）眼藥水亦有效控制近視加深。陳教授團隊另一項研究指出，部分兒童在結合阿托品眼藥水與紅光治療後，甚至可以逆轉近視情況：「部分兒童接受六個月治療後，100度近視完全逆轉，不再需要配戴眼鏡，眼軸長度亦隨之縮短。」

陳教授強調建立本地基因組數據庫的重要性，因為不同族群的遺傳特徵存在差異，西方研究成果未必適用於本地患者。建立本地數據庫能為不同專科發展精準醫學提供寶貴的參考價值。他表示：「透過分析本地兒童的基因數據，我們能更準確地識別近視風險基因，為香港學童提供更精準的預防策略。有賴基因組醫學，我們能及早識別高風險兒童，進而調整他們的閱讀習慣、實施個人化治療計劃，從而降低高度近視發生的可能性。」

Childhood Myopia and Genomic Analysis
兒童近視及基因組分析

As myopia often begins during school age when the eyeball is still developing, conducting paediatric myopia research reveals not only the disease’s origins but also its developmental trajectory.

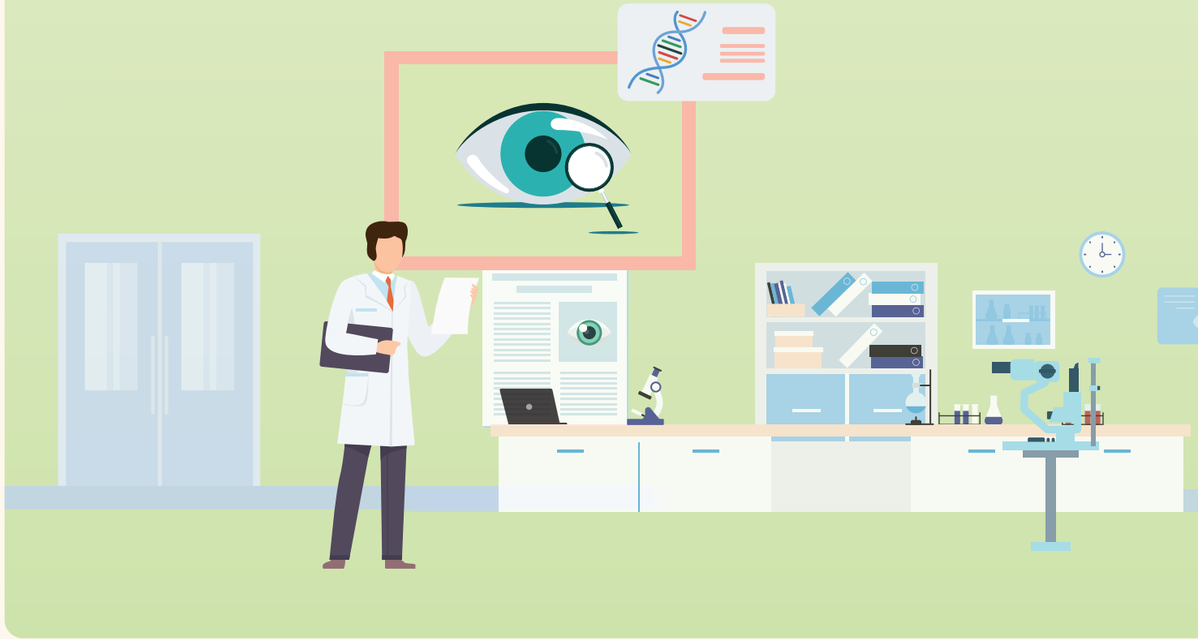
Professor Chen’s research studies both the genetic and environmental factors of childhood myopia. His research team invites primary school students and their parents to participate in detailed ophthalmological examinations, including vision tests, eye pressure, and fundus examinations, while collecting oral mucosa or blood samples to gather genetic data from nearly 40,000 Hong Kong children.

With support from the HKAM-HKGI Research Excellence Grants in Genomic Medicine, Professor Chen’s team conducted genomic study on approximately 6,000 children, with the goal of discovering new childhood myopia-related genes and developing genetic risk scoring models to identify high-risk children early. In doing so, doctors will be able to implement targeted prevention and management measures, reducing myopia’s impact on the next generation.

由於近視通常在學齡期開始出現，此時眼球仍處於發育階段，因此進行兒童近視研究不僅能揭示病源，更能掌握其發展軌跡。

陳教授的研究旨在探討兒童近視的遺傳與環境因素。他的研究團隊邀請小學生及其家長參與詳盡的眼科檢查，包括視力測試、眼壓測量及眼底檢查，並採集口腔黏膜或血液樣本，至今已收集近四萬名港童的基因數據。

在香港醫學專科學院－香港基因組中心「基因組醫學卓越研究獎」的支持下，陳教授的團隊對當中約6,000名兒童進行全基因組研究，期望透過發現新的兒童近視相關基因，開發遺傳風險評分模型，以助盡早識別高風險兒童，並實施針對性的預防及管理措施，降低近視對下一代的影響。



Our Genomic Advocates
基因組醫學新力量

Genomic Medicine Unlocks New Frontiers in Psychiatric Treatment

基因組醫學 啟發精神科治療新方向

> **Genomic medicine is opening new possibilities for clinical psychiatry. Through whole genome sequencing, we can better understand the genetic factors that contribute to mental illness, with the hope of providing personalised diagnosis and medication strategies to patients while reducing medication side effects.**
>
> **基因組醫學為臨床精神科帶來嶄新可能。透過全基因組測序，我們能更深入了解導致精神疾病的遺傳因素，希望將來為病人提供個人化診斷及用藥方案，並降低藥物可能引起的副作用。**

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Mental Illness and the Genetic Connection

The causes of mental illness have long been an important area of medical research. As Dr Fong Chun-ho introduced, "The vast majority of mental illnesses relate to both genetic and environmental factors. These conditions are typically influenced by multiple genes rather than a single gene."

精神疾病 基因關聯

精神疾病的成因一直是醫學研究的重要領域。方醫生指出：「絕大部份精神疾病均與遺傳及環境因素息息相關。從遺傳角度而言，這些疾病通常受多個基因共同影響，而非單一基因所引致。」

Through genome-wide association study (GWAS), researchers have identified an increasing number of genetic variants associated with mental disorders. "For example, schizophrenia maybe connected to genes that affect the immune system and neurotransmission, while bipolar disorder is associated with genes that control how brain cells communicate. Understanding these genetic variants helps us predict disease mechanisms and enriches our knowledge of these conditions," explained Dr Fong.

Genomic Breakthroughs in Psychiatric Diagnosis

The rapid development of genomic medicine in recent years has the potential to open new pathways for the diagnoses of mental disorders. "Genetic screening allows us to identify high-risk population before symptoms appear," Dr Fong said. "We now know that chromosomal and genetic conditions such as Fragile X syndrome, Down syndrome, and DiGeorge syndrome are associated with increased risks of autism, Alzheimer's disease, and schizophrenia respectively. These discoveries enable us to conduct disease screening, diagnoses, and treatments more efficiently."

This is only the start of realising the potential of genomic medicine. Dr Fong added, "Leveraging advancements in medical technologies, scientists have already been assessing the combined impact of hundreds of genetic variants alongside environmental risk factors to develop screening models for mental illnesses. In time, the establishment of polygenic risk scores will enable more accurate prediction of risks of developing mental disorders, facilitating early intervention or even prevention."

透過全基因組關聯分析，研究人員已發現越來越多與精神疾病相關的基因變異。方醫生解釋道：「舉例而言，思覺失調有可能與影響免疫系統及神經傳導的基因有關；躁鬱症則與控制腦細胞間通訊的基因相關。了解這些基因特性及變異，有助我們推測發病機制並增加對疾病的認知。」

醫學進步 診斷突破

基因組醫學有望於未來為精神疾病診斷另闢新徑。方醫生指出：「基因篩查有助我們在症狀出現前，識別出高風險組群。目前，我們已知的染色體與遺傳疾病，如脆性X綜合症、唐氏綜合症和迪喬治症候群，分別與自閉症、阿茲海默症和思覺失調的風險增加有關。這些發現有助我們更有效地篩查病患、作出診斷及提供治療。」

這僅僅是發揮基因組醫學潛力的起步。方醫生續指：「隨着醫學技術日新月異，科學家正努力透過綜合數百種基因變異的影響，配合環境風險考量，從而為精神疾病開發篩查模型。未來，透過建立多基因風險評分，我們有望能夠更準確預測患者罹患精神疾病的風險，實現早期干預，甚至預防發病。」

Personalised Medicine Becoming Reality

With its valuable insights into the genetic foundations of mental health disorders, genomic medicine is also driving new momentum in disease treatment and patient care. Pharmacogenomics, which studies how genes affect drug responses, has become an essential tool in transforming future psychiatric medication strategies, allowing doctors to predict both medication effectiveness and potential adverse reactions based on a patient's genetic profile. Dr Fong explained, "For example, before prescribing the mood stabiliser Carbamazepine, we must conduct genetic screening for the HLA-B*1502 variant. Patients carrying this gene face high risks of developing severe, potentially fatal skin reactions like Stevens-Johnson syndrome when taking this medication, with symptoms including widespread skin peeling and tissue death."

Dr Fong believes that as this new sphere of modern science and medicine continues to flourish, such approach of personalised medicine can be further expanded. The goal of pharmacological research in genomic medicine is to analyse patients' genetic profiles to predict their responses to different psychiatric medications, thereby optimising treatment outcomes while minimising side effects. It is anticipated that in the near future, applications of genomic medicine will further enhance the precision of psychiatric treatments and inspire research and development of new drugs, bringing benefits to patients.

"Although mental illnesses are often associated with environmental and social factors such as childhood trauma and stress, and we still have much to learn about the relationship between genetics and mental health, genomic medicine holds tremendous promise," Dr Fong concluded. "It will enable more effective diagnoses and treatments for psychiatric patients, ultimately enhancing their quality of life. This is the belief that drives me as a clinician and researcher."

個人化用藥 落地應用

基因組醫學助我們了解精神疾病與遺傳的關連，為臨床治療和護理帶來嶄新啟示。就此而言，研究基因如何影響藥物反應的「藥物基因組學」便是革新精神科用藥的關鍵工具，讓醫生能夠根據患者的基因圖譜，預測藥物效用和潛在副作用。方醫生舉例說：「譬如在處方情緒穩定劑卡馬西平（Carbamazepine）前，我們必須為患者進行HLA-B*1502基因篩查。這是由於攜帶此基因的患者服用此藥物後，有較高機率出現嚴重皮膚過敏反應，包括誘發可能致命的史蒂芬－約翰遜症候群，出現廣泛性皮膚脫落及壞死等。」

方醫生深信隨着基因組醫學迅速發展和普及，這種個人化的用藥方式定將進一步擴展。基因組醫學的藥物研究志在透過分析患者的基因組特徵以預測病人對不同精神科藥物的反應，期望藉此提升治療成效及減少副作用。基因組醫學的應用不僅有望於不久的將來提高精神科治療的精準度，同時有助啟發新藥研發，為病人帶來裨益。

方醫生總結道：「儘管精神疾病受童年創傷、壓力等環境和社會因素影響，但病患與基因遺傳之間的關係仍然是重要的醫學領域，充滿未知和可能，有待我們研究探索。我深信基因組醫學蘊藏巨大潛力，隨着這個專業不斷發展，將可實現精準診斷和個人化治療，提升患者的生活質素。作為醫生和研究人員，這也是我努力向前的信念和動力。」

Genomics Research on Tardive Dyskinesia and Antipsychotic Medications
基因變異及抗精神病藥物與遲發性運動障礙的研究

Dr Fong's research focuses on tardive dyskinesia (TD) caused by antipsychotic medications. TD is a serious and often irreversible medication side effect affecting approximately 10% – 20% of antipsychotic drug users. Symptoms include uncontrolled movements of muscles in the mouth, tongue, or other body parts. The pathological mechanisms of this condition remain incompletely understood.

Dr Fong's research explores the association between IL-10 genetic variations and the risk of developing TD. IL-10 is an important anti-inflammatory cytokine that helps regulate the immune and antioxidant systems. Through this research project, Dr Fong aims to examine three specific genetic variants of IL-10, and compare the differences between 160 patients and non-patients to gain a deeper understanding on the relationship between IL-10 and the mechanisms underlying TD.

For patients who need antipsychotic medications, tailored prescriptions could alleviate symptoms, reduce the risk of side effects, and enhance medication adherence, thereby improving their quality of life and facilitating progress towards recovery. This research is anticipated to deepen our understanding of applications of genomic medicine in mental health care, from predicting disease risk to enhancing diagnostic accuracy and ultimately enabling personalised treatment plans.

方醫生的研究聚焦抗精神病藥物引起的遲發性運動障礙。遲發性運動障礙是一種嚴重且通常不可逆轉的藥物副作用，約有10%–20%的抗精神病藥物使用者有機會出現此病症。症狀包括口、舌或身體其他部位的肌肉不受控制地蠕動。目前醫學發展仍未完全釐清此病症的病理機制。

方醫生的研究旨在探討IL-10基因變異與遲發性運動障礙風險的關聯性。IL-10是重要的抗炎細胞因子，有助調節免疫系統及抗氧化機制。他希望透過檢驗IL-10的三種特定基因變異，比較160名患者與非患者的差異，從而了解更多IL-10和遲發性運動障礙致病機制的關係。

對於需要使用抗精神病藥物的患者而言，精準用藥能有效改善症狀，降低出現副作用的風險，並提升藥物依從性，助他們改善生活質素，踏上康復之路。藉着這項研究，我們有望加深認識基因組醫學在精神健康領域的應用，從預測疾病風險到精準診斷，以及實現個人化治療，讓病人受惠。