醫院管理局
HOSPITAL AUTHORITY

Hospital Authority

Strategic Service Framework for Genetic and Genomic Services

Contents

Foreword by Chairman

Advances in genetics and genomics, along with our increasing understanding of the human genome and the genetic basis of disease, have opened up exciting new avenues in healthcare to support early diagnosis, more targeted treatments and better disease prevention.

As the major provider of public healthcare services in Hong Kong, the Hospital Authority (HA) is expected to be at the local forefront of genetics and genomics in medicine. Yet, to achieve this there is more to do to develop and improve our services to ensure we do not continue to lag behind advances in the field, so that we could meet the modern healthcare needs of our patients and their families. Meanwhile, recognising the importance of genomics in contemporary medicine, the Government is driving for the development of genomic medicine in Hong Kong. Hence, the timing of this Framework has come at an important juncture, to set out the ambitions for our genetic and genomic services and the strategies we will pursue in the coming years to achieve them. It also provides the foundation for looking at how we could support the Government-led initiatives on genomic medicine.

Formulation of the Framework has involved the contribution of a great many people and I wish to express my sincere gratitude for the valuable time and inputs given by frontline healthcare professionals, our healthcare partners from the two local medical schools and the Department of Health Clinical Genetic Service, HA executives, as well as my fellow Board members.

I have every confidence that through the dedication, combined efforts and collaboration of us all in HA, the strategies laid out in this Framework will be turned into effective actions for improving our genetic and genomic services to meet the healthcare needs of our patients.

Professor John C Y LEONG
Chairman
Hospital Authority

Foreword by Chief Executive

The international trend in medicine is moving towards the increasing use of genetic and genomic information by healthcare professionals to deliver more personalised and tailored patient care, rather than the traditional “one-size-fits-all” approach.

In HA, we are taking care of a growing number of patients for whom information about their genetic makeup can help inform decisions on their treatment and management. To maximise the benefits, we have to adopt a more systematic approach in providing our genetic and genomic services, so that they are better organised, of high quality and accessible to patients in need. Crucially, this will require nurturing a workforce with the relevant skills and expertise to appropriately use genetics and genomics during the course of clinical care, and engaging patients and their families as partners in their care.

Building on our previous service initiatives, the Strategic Service Framework for Genetic and Genomic Services will serve as the overarching blueprint to guide and align our clinicians and executives in the development of HA’s genetic and genomic services in the coming years.

My sincere thanks go to all the staff and stakeholders who have contributed to developing the Framework. Bringing the Framework to fruition will require a phased approach and the concerted efforts from a range of different healthcare professionals. I look forward to working with you all to implement the Framework, so as to harness the advances of genetics and genomics technology for our patients.

Dr Tony KO
Chief Executive
Hospital Authority

Preface

The Strategic Service Framework for Genetic and Genomic Services will guide the development of HA's genetic and genomic services over the next five to ten years. It outlines the strategic directions we will pursue to build up the service model and system infrastructure, so as to address the service gaps and improve the quality of our genetic and genomic services.

Through the Framework we will focus on strengthening the governance and organisation of our clinical and laboratory services in this respect, to improve their coordination and effectiveness. A tiered approach will be adopted in service provision, so as to match the right level of care and expertise with patient needs. In parallel, steps will be taken to equip our staff members with the right knowledge and skills in genetics and genomics according to the level of service requirements and patient complexity. To keep pace with advancements in genetics and genomics and to facilitate timely access to services, we will also look at improving the financial arrangements that support their development and provision.

Formulation of the Framework has involved the participation of a wide range of stakeholders, including frontline healthcare staff, Head Office and Cluster executives, as well as representatives from the Department of Health Clinical Genetic Service, the University of Hong Kong, and the Chinese University of Hong Kong. Their views and aspirations have been instrumental in shaping the future service developments. In particular, our sincere appreciation goes to the members of the Taskforce and Working Groups for their guidance and expert advice, as well as to the Subgroup members for their in-depth deliberations. We would also like to extend our heartfelt thanks to everyone who provided suggestions and feedback on the draft Framework.

We look forward to your continuing support and participation in implementing the strategies of this Framework in the years ahead.

Dr Libby LEE
Director, Strategy & Planning Division
Hospital Authority Head Office

Dr K L CHUNG
Director, Quality & Safety Division
Hospital Authority Head Office

Executive Summary

Introduction

The Hospital Authority (HA) Strategic Service Framework for Genetic and Genomic Services (The Framework) is the overarching blueprint for guiding the planning and development of human genetic and genomic services in HA over the next five to ten years. It outlines the directions and strategies for building up the service model to address the existing issues and improve the service quality.

According to the World Health Organisation, genetics is the study of heredity and the mechanism by which genetic factors are transmitted from one generation to the next; while genomics is the study of the structure and action of the genome (which is the complete set of genes), including how the different genes interact with each other and with the environment. The global trend of healthcare systems is moving towards integrating genetics and genomics (G/G) into mainstream clinical practices to facilitate personalised and precision medicine.

In view of rapid advances in the field of G/G worldwide that have transformed medical care, it is important for HA to enhance its G/G services and harness the potential of the relevant technologies for improving patients' health outcomes. In this regard, concerted efforts are required from frontline clinical and laboratory staff, as well as executives from the hospitals, Clusters and HA Head Office to work together towards achieving the following vision:

HA aims to provide structured and coordinated G/G services that are evidence-based and keeping pace with advances in G/G development, through professional staff with the relevant skills and expertise to meet patients' healthcare needs in a timely and equitable manner.

Planning Process

Formulation of the Framework commenced in late 2017. Under the policy direction and guidance of the Medical Services Development Committee (MSDC) and the Directors' Meeting, a Taskforce was established to oversee the formulation process. Working Groups were also set up to advise on the future governance and service development, as well as the specific areas of cancer G/G services, and prenatal and paediatric G/G services. Overall, a highly participative and broad engagement approach was adopted, with involvement of frontline healthcare staff and representatives from relevant Coordinating Committees (COC) and Central Committees (CC), as well as HA executives. In addition, as key providers of G/G services in Hong Kong, representatives from the Department of Health Clinical Genetic Service (DH CGS), the University of Hong Kong (HKU), and the Chinese University of Hong Kong (CUHK) also participated.

The formulation process included literature review to gather information about overseas experience and developments, as well as situation analysis of the local service landscape, particularly with regard to HA's existing G/G services. In addition, a comprehensive consultation exercise was carried out to identify the key issues and draw up the strategies for addressing them. This comprised a workshop carried out by an overseas expert, questionnaire survey of key specialties on G/G services in HA, hospital and site visits, and meetings with relevant COC/CCs and survey respondents.

The findings and proposed strategies were put forward to the Taskforce for formulating the Framework, and regular reports were made to the Directors' Meeting and MSDC for policy direction. Consultation on the draft Framework was conducted in June 2019. The responses and comments received were carefully considered and deliberated by the Taskforce. The refined Framework was subsequently submitted to the Directors' Meeting for endorsement, and then to MSDC for final approval.

Framework Strategies for HA's Genetic and Genomic Services

The Framework comprises five strategic directions for HA to improve its G/G services, covering service organisation, financial support, governance, talent and expertise, and performance monitoring, as follows:

Under each strategic direction, strategies have been formulated with reference to the identified key issues or opportunities for improvement in HA's G/G services.

1. Strengthen the Coordination and Collaboration of Different G/G Services to Enhance Service Quality and Accessibility

Opportunities for Improvement

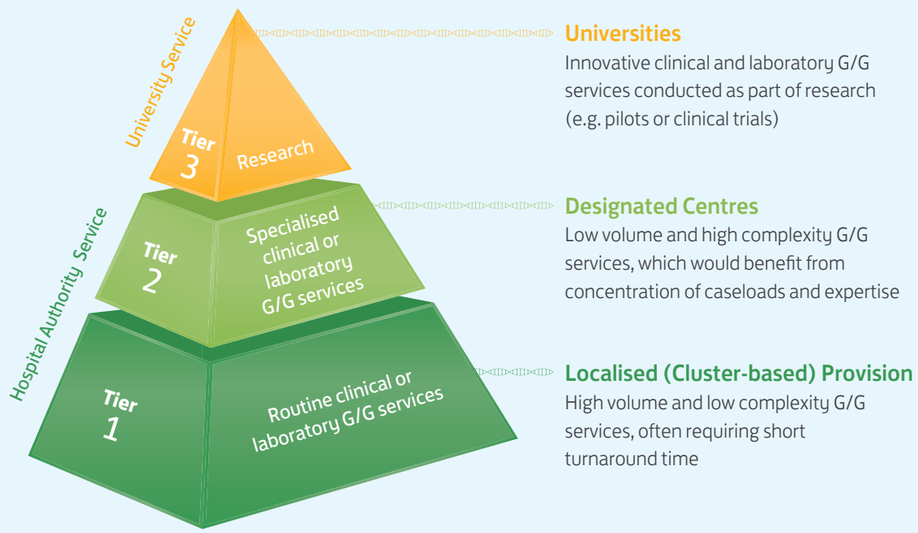
Due to the historical development of public clinical genetic service in Hong Kong, there is currently no dedicated clinical department providing G/G services in HA, while DH is the main service provider although it does not offer treatment for G/G disorders. In general, HA's G/G services have mostly developed at the local level and on an independent basis by individual clinicians or hospitals according to the interest and expertise of the respective staff and local needs, and have largely been driven through laboratory initiatives. The absence of a structured clinical genetic service in HA has resulted in significant service gaps, along with a mismatch between clinical needs and laboratory support. Moreover, there are very few standard protocols or test criteria for G/G services in HA and referral practices vary considerably, often relying on the informal links between individuals or departments, which give rise to inequitable access to the services.

Strategies

In order for HA's G/G services to develop and move forward, it is crucial to improve the service organisation by strengthening their coordination and collaboration so that the services are more structured and systematic. Strategies include:

- Developing structured and coordinated G/G services by organising different services based on a tiered approach. The services will be organised into three tiers according to service complexity and expertise requirement, as shown in the following diagram. Tier 1 consists of routine services provided in local hospitals through a Cluster-based approach. Tier 2 comprises specialised services which will be provided at designated centres to concentrate the caseloads and expertise. Tier 3 involves innovative services that will be provided mainly in the teaching hospitals by the universities as part of research.

Structured Genetic and Genomic Services based on a Tiered Approach



Universities
Innovative clinical and laboratory G/G services conducted as part of research (e.g. pilots or clinical trials)

Designated Centres
Low volume and high complexity G/G services, which would benefit from concentration of caseloads and expertise

Localised (Cluster-based) Provision
High volume and low complexity G/G services, often requiring short turnaround time

- Establishing collaborative G/G service networks according to a programme-based hub-and-spoke service model. The "hubs" are Tier 2 designated centres providing specialised services, while the "spokes" are Tier 1 routine services provided locally. The different networks could be grouped under the following three themes: (i) Paediatric Programmes that cover networks for prenatal, newborn and paediatric G/G services; (ii) Disease / Condition-based Programmes, such as networks for cancer G/G services; and (iii) Organ-specific Programmes, such as cardiac G/G service networks.

- Developing a G/G Service Directory to facilitate standardised service provision and information sharing. The electronic Directory will provide up-to-date reference on where HA funded G/G clinical services and laboratory tests are available, as well as the access criteria.

2. Provide Timely Financial Support for G/G Service Provision and Development to Help Keep Pace with G/G Advancements

Opportunities for Improvement

Largely due to the independent development of G/G tests across HA, as well as unavailability of certain tests as standard service, there are variable funding arrangements for G/G tests received by patients. The funding mechanism can include HA funding, self-financed items (SFI), funding by universities, and sponsorship by pharmaceutical companies, which is complicating how tests are provided and their accessibility. In addition, some HA laboratories may cross-charge other HA hospitals and departments for tests that have been referred to them, mainly because they are developing and providing the tests using their baseline budgets without additional resources. An underlying reason is the long lead time in the annual planning cycle for getting the resources, which cannot keep up with the rapid G/G development.

Strategies

The development of structured and accessible G/G services in HA requires appropriate planning and allocation of financial resources. To tie in with the fast pace of G/G development and the reengineered service model, the strategies include:

- Establishing a designated fund to help expedite the introduction of new G/G tests into HA. The funding will be allocated to the relevant departments on a time-limited basis to pilot and evaluate new tests. Recurrent funding, if needed, could subsequently be sought through the annual planning process.

- Developing programme-based funding to support the collaborative G/G service model. The funding will be distributed among the clinical departments and laboratory services involved in a specific G/G programme for meeting their operational needs, including to designated centres for providing specialised services across HA. Steps will also be taken to provide and fund the relevant biomarker tests for drugs in the HA Drug Formulary.

3. Enhance the Governance of G/G Services for Better Coordination

Opportunities for Improvement

Over the years, CC(Genetic Services) has been the only platform for coordinating HA's G/G services. Despite its cross-specialty membership, coordination of the specialties and hospitals providing G/G services as well as between the clinical and laboratory services is suboptimal. As a result, the G/G tests and technologies are not introduced into HA in a systematic or coherent manner, and they are often not in sync with the overall clinical need. At the same time, there is no mechanism for evaluating when to transpose research-based services into HA. This means well-evidenced services or tests from the academic sector are not transferred into mainstream clinical practice in a timely manner, which could otherwise have benefitted a wider number of patients. All these have contributed to HA's G/G services falling behind international developments.

Strategies

Robust governance is essential for HA's G/G services to ensure the service provision and development is carried out in an effective manner with transparency and accountability. This includes the governance in determining which innovations or new technologies should be introduced and how to introduce them. Strategies include:

- Strengthening the governance structure and process for overseeing G/G service provision and development. A newly established Steering Group on Genetic and Genomic Services in HA, chaired by the Chief Executive, will steer the overall development with regard to the strategies laid out in this Framework. Under the guidance of the Steering Group, CC(Genetic Services) will oversee the service development and coordination. In addition, various Expert Panels for clinical and laboratory work streams as well as for the different programmes in the new service model will be set up under CC(Genetic Services) to provide advice, and will include representatives from the universities and DH CGS as appropriate.

- Establishing a central mechanism for assessing and prioritising the introduction of new G/G services in a timely and systematic manner. To be developed under the revamped governance structure, the mechanism will cover all G/G services that are proposed to be included as HA standard service and which require additional resources. It will also cover the transfer of G/G services across the different service tiers, including the translation of research-based services into HA service.

4. Nurture a Skilled and Competent G/G Workforce in HA

Opportunities for Improvement

As far as specialised G/G expertise is concerned, there is at present no "Clinical Geneticist", "Genetic Counsellor", or "Bioinformatician" establishment in HA. Most of the expertise is vested in HKU, CUHK and DH CGS. Besides, the role and duties of staff involved in HA's G/G services are not well defined and there are different interpretations among departments on what they should involve. It is also unclear who should perform certain tasks, such as genetic counselling or the ordering of tests. As a result, there is variability across HA on what staff are involved in G/G services and to what extent. In addition, there is inadequate genetic literacy among healthcare staff. Many of them may not be able to recognise conditions with a genetic basis.

Strategies

Fundamental to the provision of HA's G/G services in alignment with the reengineered service model is a workforce with the right mix of skills and competencies. The priority is to strengthen the workforce for delivering the different tiers of G/G services. Strategies include:

- Setting out the competency requirements and building up relevant expertise for the delivery of advanced G/G services. This includes defining the qualifications, professional competencies and associated training for specialised roles.

- Taking steps to raise the G/G literacy of healthcare staff for enhanced awareness and capabilities. The aim is to facilitate the healthcare staff in managing common G/G cases and knowing when to appropriately refer to specialised G/G services.

5. Promote Performance Monitoring for Continuous Quality Improvement

Opportunities for Improvement

At present there is no systematic monitoring of HA's G/G services in terms of quality and performance. Service monitoring is mostly performed at the department level in the absence of standardised HA-wide indicators.

Strategies

Systematic monitoring of clinical and laboratory G/G services is important to help drive improvements. Strategies include:

- Identifying key domains and developing indicators for evaluating and monitoring patient outcomes.

- Enhancing data collection, including standardisation of data capture and alignment of measuring tools.

Key Enablers

Implementing the strategies will require key enablers, in particular Information Technology (IT), to support the service development. For instance, IT system infrastructure will be crucial in enabling the workflow, communication and coordination of different clinical specialties and pathology disciplines. Specifically, IT will be deployed in the establishment of the electronic G/G Service Directory, and in providing system support for the collection, storage, analysis and sharing of high volumes of G/G data in HA.

Implementation and Monitoring

Successful implementation of the Framework requires the concerted efforts of various stakeholders and input of different resources. It will be led by the Steering Group on Genetic and Genomic Services in HA, which will report to the Directors' Meeting and MSDC, as appropriate, on the development of G/G services in HA. While the overall directions and strategies are laid out in this Framework, the operational details for the implementation will be worked out by the key stakeholders under the guidance of the Steering Group. Some of the strategies do not require additional resources, while others will incur resources, which could be sought through the annual planning process.

The implementation will be monitored at different levels, including the existing mechanism of Annual Plan programme monitoring; progress review of the operational plan for the Framework to be conducted by CC(Genetic Services), with guidance from the Steering Group on Genetic and Genomic Services in HA; and the development of HA-wide quality indicators on clinical and laboratory G/G services.

Conclusion

Rapid advances in the field of G/G internationally have transformed our understanding of the role our genes play in health and disease. Keeping in pace with the development and adopting the relevant technologies to guide healthcare decisions and treatment is the way forward for healthcare systems worldwide. As the blueprint for HA's G/G services, the Framework sets out the directions for HA to work towards in this regard, thereby contributing to the overall development of G/G in Hong Kong. The strategies will not only help address the service needs of today, but also lay the foundations for HA to leverage on the huge potential of G/G innovations and advancements to benefit patient care in the years to come.

摘要

引言

為了規劃和發展未來五至十年的遺傳及基因組服務，醫院管理局（醫管局）制訂了《遺傳及基因組服務策略》作為策略性藍本，當中闡述服務模式的發展方向和策略，務求應對目前服務上的不足，以提升服務質素。

根據世界衞生組織的定義，遺傳學是研究基因和遺傳因子由一代傳到下一代的科學；而基因組學主要研究基因組（即一套完整的基因）的結構和作用，包括不同基因之間及基因與環境之間，如何產生相互作用。現時全球醫療體系都趨向將遺傳學及基因組學納入主流臨床應用，以提供個人化和精準的治療。

鑑於遺傳及基因組學在全球迅速發展，徹底改變了傳統的醫療方法，而且能夠達至更有效、更到位的治理，所以醫管局有必要提升這方面的服務發展，並採用相關具潛力的科技，改善病人的醫療成效。前線醫護人員及醫院、聯網和總辦事處的行政人員，必須同心協力，才可以實現以下願景：

醫管局旨在為市民提供協調及有系統的遺傳及基因組服務：以實證為本、隨着有關科學的發展步伐，並配合專業人員的技能，適時及公平地為有需要的病人提供治療。

規劃過程

醫管局於2017年末正式開展本服務策略。在「醫療服務發展委員會」及「總監會議」的政策發展指導下，我們成立了專責小組，負責監督整個服務策略的制訂過程。此外，我們亦成立了不同的工作小組，分別就未來的管治架構、服務發展，以及有關癌症、產前和兒科的遺傳及基因組服務提供意見。為了集思廣益，我們邀請了前線醫護人員、相關統籌委員會和中央委員會的代表，以及醫管局行政人員參與服務策略的制訂過程。另外，衞生署醫學遺傳科、香港大學和香港中文大學作為現時三大提供遺傳及基因組服務的機構亦參與其中，一同刻劃未來服務發展的藍圖。

制訂過程除了透過閱覽文獻以參考海外相關經驗和發展外，我們亦就本地情況進行了深度的分析，特別探討醫管局現有的遺傳及基因組服務缺口。同時，我們透過廣泛的諮詢工作，包括舉辦由海外專家主持的工作坊、向醫管局轄下提供遺傳及基因組服務的主要專科進行問卷調查、到訪醫院和相關機構，以及與有關的統籌委員會／中央委員會成員和問卷受訪者會面，從而找出我們需要面對的主要挑戰，以制訂應對及發展策略。

專責小組在仔細討論其分析結果和擬議策略後，需定期向「總監會議」及「醫療服務發展委員會」匯報，以確立遺傳及基因組服務的發展方向。我們於2019年6月就本服務策略的初稿進行了諮詢，並在專責小組詳細分析和討論所得的意見後，將優化的服務策略呈交「總監會議」審批，再由「醫療服務發展委員會」討論及通過。

醫管局遺傳及基因組服務策略

本服務策略訂立了以下五大策略方向，分別就服務組織、財政資助、管治、人才和專長培訓，以及服務監察各方面，提出改善醫管局遺傳及基因組服務的方針：

我們就上述策略方向制訂了相應的謀略，希望在可能的範疇內，改善醫管局遺傳及基因組服務方面主要的問題。

1. 加強各種遺傳及基因組服務的協調與協作，以提升服務質素和便捷度

可改善的範疇

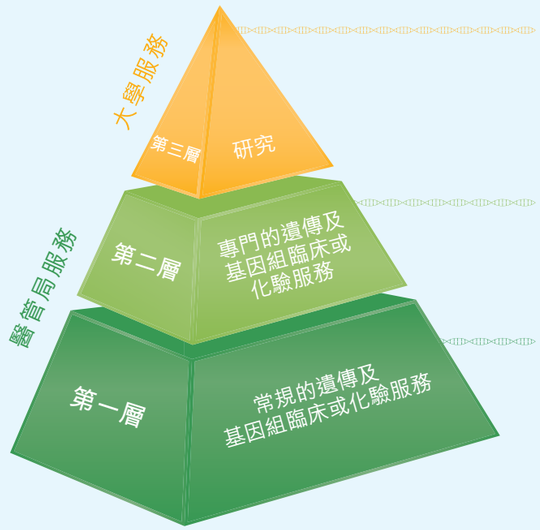
基於本港公共醫學遺傳科發展的歷史因素，衞生署設有醫學遺傳科，現時是遺傳及基因組服務的主要提供者，當中包括遺傳病輔導服務和基因測試，而醫管局則負責處理轉介個案，提供部分的化驗服務和一切相關的治療，但並未有設立專責部門提供遺傳及基因組服務。一般來説，醫管局的相關服務發展主要是基於個別醫生和醫院的臨床需要，並大部份由化驗工作主導。局內因沒有架構完整的醫學遺傳科，有關服務明顯不足，臨床需要和化驗室支援也時有出現不協調的情況。此外，局內尚未有相關完整的標準規程或測試準則，基因測試及專科治療的轉介做法亦各有不同，往往只依賴個別人員或部門間的非正式聯繫，令病人難以公平地獲得所需服務。

策略

要推動醫管局遺傳及基因組服務的發展，必須透過加強專科之間的協調與協作，令服務架構更完面、服務安排更有系統。有關策略包括：

- 將各項服務按照三個層級來組織，以建立協調和有系統的遺傳及基因組服務。如圖所示，按服務的複雜程度、服務量和專才的需求，把服務分為三個層級。第一層包括以聯網為本、由地方醫院提供的常規服務；第二層是在指定中心提供的專門服務，以便集中處理複雜的個案和匯聚專才；第三層是創新服務，主要由大學在教學醫院提供，作為研究項目。

遺傳及基因組服務的分層結構



大學

創新的遺傳及基因組臨床及化驗服務，作為醫療研究的一部份（例如先導計劃或臨床試驗）

指定中心

量少但複雜的遺傳及基因組服務，需要集中個案和匯聚專才

地區化服務（聯網為本）

量多而較簡單的遺傳及基因組服務，處理時間一般較短

- 以「軸輻式」的服務模式，因應遺傳病項目而建立不同的遺傳及基因組服務網絡。「軸」是指在第二層提供專門服務的指定中心，「輻」是指在第一層提供常規服務的地方醫院。軸輻服務相連，關係密切，目的是為有需要的病人提供適切、適時的服務。不同的協作式網絡可按下列三個主題分類：(i) 兒童醫療項目，包括產前檢測、為初生嬰兒及病童提供遺傳及基因組服務的網絡；(ii) 疾病或病況為本的醫療項目，如與癌症有關的遺傳及基因組服務網絡；和 (iii) 特定器官的遺傳病項目，如有關心臟病的遺傳及基因組服務網絡。

- 建立醫管局遺傳及基因組服務名冊，方便提供標準化的服務和分享相關資料。名冊會以電子形式向醫護人員提供最新資訊，包括醫管局恆常的相關臨床服務、檢測服務點和使用準則。

2. 改善財政及撥款安排，以配合遺傳及基因組服務的發展步伐

可改善的範疇

由於公營醫院的遺傳及基因組化驗服務各有發展，加上部份檢測項目不屬於標準服務，所以病人接受檢測的經費安排也有所不同，當中包括由醫管局資助、病人自費項目、大學撥款，以及由藥廠贊助，令檢測安排變得複雜及影響服務的便捷度。此外，部份化驗室在處理由醫管局轉下其他醫院或部門轉介的基因測試時，往往因為沒有額外資源而需要動用基本經常性撥款來提供服務，又或者會向有關醫院及部門徵收所需費用。這可能是由於透過周年工作計劃申請撥款到獲配資源一般需時，令服務難以跟上遺傳及基因組學的迅速發展。

策略

醫管局要發展有系統和便捷的遺傳及基因組服務，必須適當地規劃和分配財政資源。為配合遺傳及基因組學的迅速發展及服務重組的需要，相關策略包括：

- 設立特定撥款機制，以助醫管局加快引入新的遺傳及基因組測試。撥款將以設定時限的方式分配予相關部門，以便試行和評估新的檢測方法。如有恆常服務的需要，相關部門可於日後透過周年工作計劃申請經常性撥款。

- 為遺傳及基因組服務網絡所組成的主題醫療項目提供「項目為本」的撥款，在財政上支援新的協作服務模式。撥款將會分配予參與特定項目的臨床和化驗部門（包括提供專門的遺傳及基因組服務的指定中心），以應付運作的需要。此外，我們亦會就《醫管局藥物名冊》內的有關藥物，安排和資助相應的生物標記測試。

3. 提升遺傳及基因組服務的管治，以增進協調

可改善的範疇

多年來，「遺傳服務中央委員會」是協調醫管局各遺傳及基因組服務的唯一平台。現時委員會由跨專科成員組成，可是服務協調上仍有待改善，尤其是牽涉不同專科、醫院、臨床和化驗服務之間的工作。新的遺傳及基因組測試和科技亦因此未能有系統或有條理地引進醫管局，更無法配合整體的臨床需要。與此同時，目前沒有機制評估何時適宜將大學研究的醫療服務轉移成為醫管局內的恆常服務，以致在學術界經實證的研究或測試未能適時引進主流臨床應用，無法讓更多病人受惠。凡此種種，均令醫管局的遺傳及基因組服務落後於國際發展。

策略

健全的管治對醫管局發展遺傳及基因組服務尤為重要，特別是確保有效地提供服務、增加其透明度和問責性等方面，當中涉及制定如何引入新科技或創新項目的機制。相關策略如下：

- 強化管治架構和程序，以監督遺傳及基因組方面的服務和發展。最近醫管局成立了「遺傳及基因組服務督導委員會」，由行政總裁擔任主席，將監督落實此服務策略的整體發展。在督導委員會的引領下，「遺傳服務中央委員會」將負責發展和協調相關服務。此外，「遺傳服務中央委員會」將會設立多個專家小組，分別就臨床和化驗工作及新服務模式下的各個主題醫療項目，提供意見。

- 設立中央機制，對新的遺傳及基因組服務項目作出評估，和訂立推行的優先次序，務求能適時和有系統地引入新項服務。重整後的管治架構將主導此機制，審核範圍包涵所有申請納入醫管局標準服務並需要額外資源的遺傳及基因組服務，以及在三層架構中不同服務由一層轉移至另一層的安排（包括把大學醫療研究項目轉型為醫管局恆常服務）。

4. 培育醫管局內具備遺傳及基因組學技能的專業工作團隊

可改善的範疇

就遺傳及基因組服務而言，醫管局並未設有「臨床遺傳學家」(Clinical Geneticist) 、「遺傳諮詢顧問」(Genetic Counsellor) 或「生物信息學家」(Bioinformatician) 的人手編制，現時這些專家均由香港大學、香港中文大學和衞生署醫學遺傳科所聘用。另外，局內參與提供遺傳及基因組服務的人員並無特定的分工與職責，而不同部門對相關工作範疇也有不同的理解，個別工作該由誰來處理（例如進行遺傳病輔導或安排基因測試）亦沒有清晰的釐定。因此，各部門和醫院對於遺傳及基因組服務提供者及其負責的範疇，均有不同的做法。除此之外，一般醫護人員普遍上對遺傳學的認知不足，很多時候未能識別與遺傳病有關的病徵。

策略

具備適當技能的工作團隊是體現嶄新服務模式中不可或缺的一環。首要工作就是根據不同層級的遺傳及基因組服務，持續強化醫護團隊，策略包括：

- 訂定職能要求和培育相關專才，以提供先進的遺傳及基因組服務，包括釐定資歷要求、專業技能和專科人員所需的培訓。

- 致力提升醫護人員在遺傳及基因組方面的認知和技能，以便處理有關疾病的常見個案，並適切地轉介個案到指定中心接受專門的診治。

5. 加強服務指標監察，以持續提升服務質素

可改善的範疇

在質素和服務表現方面，醫管局現時未有對遺傳及基因組服務進行有系統的監察。由於缺乏局內通用的指標，有關服務的監察主要限於個別部門的層面。

策略

為改善遺傳及基因組的臨床和化驗服務，醫管局須設立有系統的監察機制，有關策略包括：

- 選定主要監管範疇，釐訂表現指標，以評估和監察醫療成效。
- 加強收集數據，包括統一擷取數據的工作及使用劃一的量度工具。

主要配套

要落實上述策略有賴不同的支援配套，其中資訊科技擔當着關鍵的角色。除了可促進不同臨床專科和病理學科的工作流程、通訊及協作外，我們亦須借助資訊科技建立電子化的遺傳及基因組服務名冊。資訊科技更可用以收集、儲存、分析和分享醫管局內有關遺傳及基因組的大量數據。

推行及監察

要成功推行本服務策略，我們需要各持份者的共同努力和好好運用各種資源。醫管局「遺傳及基因組服務督導委員會」將領導有關的執行工作，並適時向「總監會議」和「醫療服務發展委員會」匯報遺傳及基因組的服務發展。督導委員會將按着本服務策略的發展方向，引領各主要持份者落實執行細節。當中有些策略在推行時並不需要額外的資源，其他策略則可按需要，透過周年工作計劃的機制申請額外撥款。

我們會在不同的層面監察服務策略推行的進度和成效，包括透過現有的周年工作計劃監察機制、「遺傳服務中央委員會」在督導委員會的指引下跟進服務策略的實施進度，以及遺傳及基因組臨床和化驗服務所制訂的質素指標。

總結

遺傳及基因組學在國際間迅速發展，讓我們能夠掌握基因對健康和疾病的影響。現時全球醫療系統均緊貼這方面的發展步伐，趨向引入相關科技以進行適當、精準的醫療評估和治理。醫管局制訂了本服務策略作為發展遺傳及基因組服務的藍圖，並規劃了未來的發展方向，從而為本港遺傳及基因組學的整體發展跨出一步。這些策略不僅有助醫管局應對當前的服務需要，也為長遠發展奠下基礎，而憑着遺傳及基因組學的巨大應用潛力，加上革新的醫療方法，必定能令更多病人受惠。

Introduction

Setting the Scene for Development of the Strategic Service Framework for Genetic and Genomic Services

Genetics is the study of heredity and the mechanism by which genetic factors are transmitted from one generation to the next[1]. Genes in the living cells carry the code for directing the characteristics and biological functions of an individual. The genetic codes are held in the deoxyribonucleic acid (DNA) molecules, which are made up of strings of four chemical bases arranged in pairs in a double helix structure. The order or sequence of these bases determines the meaning of a genetic message. All genes are made up of stretches of these bases, arranged in different order or sequence and in different lengths. Each person has two copies of each gene, one inherited from each parent.

An individual's complete set of DNA, and thus all of the genes, is known as a genome. **Genomics** is the study of the structure and action of the genome, including how the different genes interact with each other and with the environment. According to a full sequence of the entire human genome completed and published in April 2003 by the Human Genome Project[2], which was an international collaborative research programme, a human genome contains at least 20,000 genes formed by over three billion pairs of bases. The different genes are located in specific parts of a human's 23 pairs of chromosomes.

The main difference between genetics and genomics is that genetics scrutinises the functioning and composition of the single gene whereas genomics addresses all genes and their inter-relationships in order to identify their combined influence on an individual[3]. In genomics, enormous amounts of DNA-sequence data are analysed using high performing computing and mathematic techniques known as bioinformatics to find variations that affect health, disease and response to drugs[4].

Despite the difference, genetics and genomics are both concerned with using genetic information to diagnose, treat, prevent and cure illnesses that have a genetic component. Many conditions or diseases are related to mutations in some of the genes. The mutations can be inherited (i.e. hereditary) or happen during a person's lifetime (i.e. acquired) contributed by environmental or lifestyle factors. Examples of inherited genetic disorders include cystic fibrosis, sickle cell disease

1 World Health Organisation. *Human Genomics in Global Health.* www.who.int/genomics
2 National Human Genome Research Institute. *An Overview of the Human Genome Project*. www.genome.gov/human-genome-project
3 World Health Organisation. *WHO Definitions of Genetics and Genomics*. www.who.int/genomics/geneticsVSgenomics
4 The Jackson Laboratory. *Genetics vs. Genomics*. www.jax.org/personalized-medicine/precision-medicine-and-you

and Huntington disease. Diseases that are caused by a combination of genetic and environmental factors rather than a single genetic defect and have been studied in the field of genomics include cancer, diabetes and heart disease[5].

Since the identification and mapping of the entire human genome, the field of genetics and genomics (G/G) in healthcare and medical sciences have developed in leaps and bounds. It has also transformed our understanding of the human genome and the association between variations in genes with health and disease. Correspondingly, G/G is increasingly being applied to support early diagnosis and more precise identification of the underlying causes of disease, help determine which individuals are at risk of developing an illness, as well as predict how specific patients will respond to particular interventions in order to direct targeted treatments and avoid those which are unnecessary or may have undesirable effects.

As a result, genetic services which have historically played a key role in identifying uncommon chromosome and single gene disorders in children and during pregnancy are shifting to encompass a much broader scope. For example, genetic services now cover multi-gene as well as single gene diseases, chromosomal abnormalities and disorders, etc., with applications that are relevant across a range of different clinical specialties and increasingly covering adult patients. Moreover, the global trend is towards greater integration of G/G into mainstream clinical practice to facilitate personalised and precision medicine. That is to say, rather than grouping patients into broad categories of disease and using a "one-size-fits-all" or "trial-and-error" approach to treatment, patient care can now be tailored and fine-tuned based on an individual's unique genomic and biological makeup.

Locally, recognising the importance of G/G in contemporary medicine and scientific research, the Government is driving the development of genomic medicine in Hong Kong through the establishment of a Steering Committee on Genomic Medicine in November 2017. Currently, the Hospital Authority (HA) has very limited clinical G/G services, although its laboratories have managed to provide quite a number of G/G tests for hereditary diseases. On the other hand, the Department of Health (DH) is the main provider of public clinical and laboratory G/G services for constitutional genetic disorders, but it does not offer treatment. Most of the G/G expertise is also vested in the University of Hong Kong (HKU) and the Chinese University of Hong Kong (CUHK), but their services are mostly research-based and cater only for a limited number of patients. As the major public healthcare provider for the bulk of patients, HA is facing a growing demand for subsidised G/G services. It is important for HA to develop its G/G services to meet the gaps as well as harness the potential of the relevant technologies for improving patients' clinical outcomes. Hence, the Strategic Service Framework for Genetic and Genomic Services has been formulated to serve as the blueprint to guide the planning and development of HA's G/G services.

5 National Human Genome Research Institute. *Genetics vs. Genomics Fact Sheet*. www.genome.gov/about-genomics/fact-sheets

DNA

- Deoxyribonucleic acid (DNA) is the hereditary material in humans, found in most cells of the body
- Information in DNA is stored as a code made up of four chemical bases: adenine (A), guanine (G), cytosine (C), and thymine (T)
- Most DNA is stored in the nucleus of a cell, but a small amount can also be found in the mitochondria - small structures in cells that convert food to energy

3.2bn
Approximate no. of base pairs in the human genome

Genes

- The basic physical and functional unit of heredity, made up of DNA with a code for a specific instruction
- Every person usually has two copies of each gene, one inherited from each parent

20,000
Estimated no. of genes in the human genome

Chromosomes

- DNA molecules and genes are packaged into thread-like structures called chromosomes
- In humans, most cells normally contain 23 pairs of chromosomes

23
No. of chromosome pairs contained in most human cells

Genome

- A genome is an organism's complete set of DNA, including all of its genes and the DNA in between

>99%
DNA the same among all human beings

Source of information: US National Library of Medicine

Vision and Scope

What We Aspire and What the Framework Is About

Patients with a genetic disorder, or genetic susceptibility to a disease, need timely access to G/G services that are well organised and appropriately integrated into their clinical pathways in order for them to receive early diagnosis and targeted treatment for their condition. This includes the availability of appropriate and affordable G/G tests that are supported by evidence in terms of their safety and effectiveness. It is also important for the patients to have a clear understanding of how their G/G information will be handled, who might have access to it and any future implications.

At the same time, there is a need to consider the possible implications to their family members and take a family-based perspective in G/G services since many genetic conditions are often inherited. In view of the rapid pace of advances in G/G technology and application, it is also crucial for the healthcare professionals to be equipped with the appropriate expertise, knowledge and skills. With these elements in mind, the following paragraphs describe our aspirations for G/G services in HA and what the Framework is about.

Vision for HA Genetic and Genomic Services

Hospital Authority aims to provide structured and coordinated G/G services that are evidence-based and keeping pace with advances in G/G development, through professional staff with the relevant skills and expertise to meet patients' healthcare needs in a timely and equitable manner.

Scope of the Framework

The Framework sets out the strategies and future service model to guide the development of HA's clinical and laboratory G/G services over the next five to ten years, including the associated supporting services, system infrastructure and performance monitoring. It aims to address the existing issues in HA's G/G services and improve service quality and sustainability.

Although G/G in healthcare services generally encompass human and microbial genetics, the scope of the Framework covers only the organisation and development of **human G/G services**, which are currently lagging behind despite being a significant component of HA's G/G service needs. Nevertheless, many of the strategies and directions outlined in this Framework could also serve as a useful reference for the microbial G/G services in HA. Meanwhile, considering that cancer, prenatal and paediatric services are the key clinical areas in human G/G services worldwide, they have been used as illustrative examples in the Framework.

The scope of the Framework does not cover the overall development of G/G services in Hong Kong, as well as the policy and legal aspects relating to the application of G/G technologies, as these are more appropriate to be addressed at the Government level by the Food and Health Bureau's Steering Committee on Genomic Medicine.

Although the specific operational details for implementation are beyond the scope of this Framework, it provides an overarching blueprint for HA clinicians and executives to align their initiatives for developing HA's G/G services.

Planning Context

Local Situation and Contextual Factors Driving the Development of Genetic and Genomic Services

Outlined in this chapter are some contextual factors that have provided the impetus for the development of the Strategic Service Framework for G/G Services in HA, particularly those that are contributing to growing demand such as rapid advances in technology, expanding research and knowledge base, rising patient expectations, and increasing provider availability. Another important context in the planning of HA's G/G service development is the local service landscape, which includes key service providers, expertise and training, as well as Government policy and initiatives, an overview of which is also provided here.

Factors Driving the Demand for G/G Services

Rapid Advances in Technology

Worldwide a number of factors are driving the rapid pace of development and demand for G/G in healthcare. In particular, technological and scientific advances have enabled G/G sequencing to be carried out faster, more efficiently and at lower costs, allowing greater affordability for healthcare systems to adopt the technology and expand its application in clinical practice.

The advent of Next Generation Sequencing (NGS), in particular, has become a revolutionary and widely adopted technology in academic research as well as clinical practice, and helped to accelerate the development of diagnostics. NGS refers to a type of high throughput sequencing technology that has the capability to enable millions of DNA strands to be sequenced in parallel, enabling multiple genes up to the whole genome to be studied at the same time. This has been transformational in the development of multigene panels to study certain diseases like cancer, or in diagnosing uncommon conditions that may require detailed knowledge about a patient's whole genome. However, despite the advances in sequencing technologies, interpreting the huge amounts of data generated and its clinical significance remains complex and requires specialised expertise.

Expanding Research and Knowledge Base

Along with technological developments, scientific advancement and research has led to a dramatic increase in knowledge and the evidence-base around G/G, especially since the mapping of the human genome over a decade ago. This has included better understanding of the genetic basis for certain disorders and growing recognition that many diseases are heterogeneous (especially in cancer); development of novel screening and diagnostic techniques, such as based on cell-free / circulating DNA; and a growing number of targeted therapies as well as more accurate sequencing and analytical techniques.

In recent years, a number of countries such as the United Kingdom, Australia, Singapore and the United States have embarked on large-scale genome sequencing projects to enhance the clinical and scientific evidence-base and catalyse research. The projects are contributing to increasingly sophisticated data analytics and enhanced capabilities in linking big genetic data with other information, like medical records, to provide new insights into the genetic basis of disease and the identification of more effective treatment. In Hong Kong, the Government is also in the process of setting up a large-scale Hong Kong Genome Project (HKGP).

Rising Patient Expectations

Increasing awareness and interest among the general public on the potential application of G/G testing also drives demand. In particular, nowadays patients and their families are able to find out from Internet resources how certain G/G tests or services could benefit them, potentially leading them to request these from their doctors. For instance, G/G profiling of tumours to inform on targeted therapy is becoming increasingly important and popular in cancer treatment, especially for breast, colorectal and lung cancer patients. In fact, G/G testing associated with Pharmacogenomics, which is the study of how genes affect a person's response to drugs[6], is gaining traction in helping to determine whether a medication will be effective for a particular patient and to prevent adverse drug reactions (i.e. negative side effects).

Furthermore, G/G services are often anticipated for patients with an undiagnosed or uncommon disorder, particularly for paediatric patients, since it is estimated that around 80% of uncommon disorders have a genetic origin and half of all new cases are children[7]. Cascade G/G testing may also be expected among the family members when a patient is diagnosed with a hereditary genetic condition, in order to find out whether they also have the condition and the risk of the condition being passed on to the next generation.

6 US National Library of Medicine. *What is Pharmacogenomics?* www.ghr.nlm.nih.gov/primer/genomicresearch
7 Department of Health (2013). *The UK Strategy for Rare Diseases*. London: The Stationery Office.

Increasing Provider Availability

DNA sequencing was historically very expensive, but with rapid advances in the technology, particularly in NGS, the costs have been falling. As a result, there has been a proliferation in the availability of G/G testing through private providers, including direct-to-consumer tests. Hence, there are increasing opportunities for members of the public to have their genomes tested for genetic disorders or predisposition to certain conditions. Locally, there are instances of patients seeking explanation and counselling or even active intervention from HA doctors regarding the DNA sequencing results they have received through private providers, which could place additional demand on HA's G/G services.

Overall, the global trend is towards greater integration of G/G testing and analysis into patient pathways and routine clinical care. Although the pace and level of adoption is likely to vary among different clinical specialties, depending on their service needs and readiness, G/G is set to play an increasingly vital role in healthcare delivery.

Local Situation of G/G Services

Key Service Providers

In Hong Kong, the public Clinical Genetic Service (CGS) was first set up by the former Medical and Health Department (MHD) of the Government in the early 1980s. When MHD eventually split into DH and HA, with the latter established as a statutory body in 1990, the CGS, unlike other specialist services that were transferred to HA, remained with DH. As a result, the DH CGS is currently Hong Kong's main provider of public clinical and laboratory G/G services for constitutional genetic disorders, but it does not offer treatment.

In parallel, HKU and CUHK are major players in the field of G/G both locally and internationally, with their services being mostly research-based which cater for a limited number of patients. At the same time, G/G services have also become increasingly accessible over the years to members of the public through private hospitals and clinics, as well as via non-government organisations (NGOs), etc.

HA's G/G services, on the other hand, are less established and mature compared to those of DH, HKU and CUHK due to the absence of a dedicated clinical department for service provision. As a result, there has been heavy reliance on these service providers by HA. A brief description of HA's G/G services is given in the chapter *Overview of HA's Genetic and Genomic Services*, while a summary of other key players, particularly in relation to their interface with HA, is provided below.

Department of Health Clinical Genetic Service

The service of DH CGS is led by a Consultant Clinical Geneticist and is provided by a team of doctors, nurses, as well as laboratory and other supporting staff. It receives a significant number of referrals each year from HA, mostly from Paediatrics, followed by Obstetrics & Gynaecology (O&G) and Medicine specialties. For example, in 2015 around 88% of the referrals to DH CGS were from HA. Public patients are mainly referred to the DH for genetic counselling[8] and testing, while follow-up treatment and management is provided by HA. Other sources of referral to DH CGS include private doctors and the Family Planning Association.

The DH Genetic Laboratory Service provides chromosome analysis and molecular testing to help identify the underlying genetic causes of various genetic diseases in patients and their at-risk family members. The laboratory mainly supports the Genetic Counselling Service and generally does not accept specimens for testing from outside. Hence, patients are referred to DH CGS for assessment and/or genetic counselling before the tests are ordered by the CGS doctors if deemed necessary.

DH CGS is scheduled to move to the premise of Hong Kong Children's Hospital (HKCH) in late 2019, and more collaboration is expected with HA's G/G services when that happens.

University Genetic and Genomic Services

HKU has well established services for hereditary cancer syndrome, in particular for breast and ovarian cancer syndrome as well as Lynch syndrome[9], covering patient and family assessment, testing and genetic counselling. Since G/G service for hereditary cancer syndrome is not available in HA, patients and their families at risk of having these conditions are referred to HKU by the clinicians, albeit on an *ad hoc* basis.

Both HKU and CUHK offer diagnosis and counselling for uncommon genetic disorders, which are provided by the clinical genetic teams led by G/G sub-specialists in Paediatrics from the respective universities. They are also active in supporting the investigation of patients with undiagnosed conditions to help identify the molecular basis for their condition, through the use of advanced technologies like NGS for whole genome sequencing (WGS). These university services are often offered as private, but subsidised services are also available for HA patients. Other examples in this respect include pre-implantation genetic testing[10] provided by the respective universities' clinic for assisted reproduction.

8 According to WHO, genetic counselling is a communication process through which knowledge about the genetic aspects of illnesses is shared by trained professionals with those who are at an increased risk of having a heritable disorder or passing it on to their offspring.

9 Lynch syndrome, also known as hereditary non-polyposis colorectal cancer (HNPCC), is an inherited disorder that increases the risk of many types of cancer, particularly cancers of the colon and rectum. Based on US National Library of Medicine. www.ghr.nlm.nih.gov/condition/lynch-syndrome

10 Pre-implantation genetic testing (PGT) is performed to analyse the DNA from oocytes or embryos for HLA-typing or for determining genetic abnormalities. These include for aneuploidies, monogenic/single gene defects, and chromosomal structural rearrangements.

Overall, the G/G services of HKU and CUHK are mostly provided at the two teaching hospitals, i.e. Queen Mary Hospital (QMH) and Prince of Wales Hospital (PWH) respectively. For instance, CUHK has set up at PWH a Joint Centre for Medical Genetics in collaboration with the Department of Molecular and Human Genetics from the United States' Baylor College of Medicine (BCM), providing a one-stop genetic clinic for genetic counselling, risk assessment and G/G testing for a wide range of conditions[11].

In addition, two research institutes, namely the Centre for Genomic Sciences and the Li Ka Shing Institute of Health Sciences have been set up at HKU and CUHK respectively to spearhead cutting-edge research and innovation in G/G. The universities have also participated in a number of international collaborations over the years, such as the International HapMap Project[12].

Private Sector and Non-government Organisations

There are a number of private hospitals and clinics in Hong Kong that provide cancer genetic screening, targeted cancer therapy, prenatal and newborn screening, pre-implantation genetic testing, and genetic counselling. Moreover, a variety of G/G tests including those for assessing predisposition for cancer and other specific conditions, or relating to family planning, general health, etc. are also available in some private clinics and laboratories. However, many of the patients seek further treatment from HA when a genetic condition is found.

In parallel, some NGOs are providing charitable support for certain genetic conditions. For example, the Hong Kong Cancer Fund is supporting the Hereditary Gastrointestinal Cancer Genetic Diagnosis Laboratory at HKU in providing a charitable genetic diagnosis service for patients with suspected hereditary gastrointestinal and other cancers; and the Hong Kong Hereditary Breast Cancer Family Registry provides funding for genetic testing and counselling and conducts public awareness activities. In addition, the Sudden Arrhythmias Death Syndrome (SADS) Foundation in 2014 supported a two-year study on hereditary cardiac disorders conducted by Princess Margaret Hospital (PMH) and DH, which involved genetic screening for family members of young sudden cardiac death victims and NGS investigations into the causes of death.

11 Department of Obstetrics and Gynaecology, CUHK. *The CUHK and Baylor College of Medicine One-stop Genetic Clinic*. www.obg.cuhk.edu.hk/fetal-medicine/fetal-medicine_services

12 The goal of the International HapMap Project was to develop a haplotype map of the human genome for describing the common patterns of human genetic variation. Based on National Human Genome Research Institute. www.genome.gov/11511175

G/G Expertise and Training in Hong Kong

Currently, there are five Clinical Geneticists in Hong Kong, all of whom are paediatricians by training. Two of them are at DH CGS, two employed by HKU and CUHK, and one from the private sector. As for genetic counselling, while the training requirement of genetic counselling is not well defined in Hong Kong, some clinicians, nurses and midwives provide genetic counselling if needed, albeit to varying degrees based on their knowledge and experience. With regard to Bioinformaticians, a significant amount of expertise is vested in the universities, and there is one Bioinformatician employed by HA at the HKCH.

Overall, recognising the need to build up a critical mass of expertise for the development of G/G services in Hong Kong, the Hong Kong Academy of Medicine has previously set up a Working Group on training in G/G, involving the Colleges of Community Medicine, Obstetricians and Gynaecologists, Paediatricians, Pathologists and Physicians[13]. Subsequently, the Hong Kong College of Paediatricians has established a subspecialty in Genetics and Genomics, while the Hong Kong College of Pathologists is rolling out a post-fellowship training programme and is inviting applications for First Fellows in Genetic and Genomic Pathology, with the first batch of Fellows expected to be conferred in one to two years.

There are also a number of degree programmes and courses provided by HKU and CUHK in G/G. For example, HKU runs a Postgraduate Diploma in Molecular and Diagnostic Pathology; while CUHK runs a Master of Science in Genomics and Bioinformatics, and a Master of Science in Medical Genetics, and has also set up the CUHK-BGI Innovation Institute of Trans-omics as a training ground for genomics, proteomics and bioinformatics.

Government Policy and Initiatives

The Government is proactively driving the use of new technology and innovation for better public health and clinical outcomes. Specifically, in 2017 the Government established a Steering Committee on Genomic Medicine to map out the strategies for developing genomic medicine in Hong Kong, as well as propose the broad directions for addressing regulatory and ethical issues surrounding the application of genomics. Subsequently, in the 2018 Policy Address the Government announced it had accepted a preliminary recommendation from the Steering Committee to conduct a large-scale Hong Kong Genome Project (HKGP) and would provide funding for this purpose.

In November 2018, the Food and Health Bureau (FHB) set up a Working Group on HKGP comprising experts from the academic, clinical and research sectors to discuss the details of the project. The HKGP aims to enhance the clinical application of genomic medicine and promote research in genomic medicine through the establishment of a genome database of the local population, testing infrastructure and talent pool. Key stakeholders in the HKGP are anticipated to include FHB, DH, HA, universities, private hospitals, and the research and development sector[14].

13 Hong Kong Academy of Medicine. www.hkam.org.hk/hkamweb/pages_1_220.html

14 Legislative Council Panel on Health Services (2019). *Hong Kong Genome Project*. LC Paper No. CB(2)607/18-19(03).

Planning Process

How We Developed the Strategic Service Framework for Genetic and Genomic Services

Project Governance

Under the policy direction and guidance of the Medical Services Development Committee (MSDC) and Directors' Meeting, a designated Taskforce was set up to oversee the development of the Framework. The Taskforce was co-chaired by the Director of Strategy and Planning Division and the Director of Quality and Safety Division of the HA Head Office. The terms of reference and membership of the Taskforce are set out in Appendix 1.

Under the Taskforce, three Working Groups (Appendices 2 to 4) were formed to advise on the specific areas of (i) Governance and Service Development, (ii) Cancer Genetic Services, and (iii) Prenatal and Paediatric Genetic Services. To facilitate the discussions on governance and service development, three Subgroups were set up for in-depth deliberation on (i) Service Organisation and Funding, (ii) Staff Expertise and Role Delineation, and (iii) Introduction of New Technology and Performance Measures (Appendices 5 to 7). A project team from the Strategy and Planning Division as well as Quality and Safety Division of HA Head Office provided the overall executive support for developing the Framework. The governance structure of the project is illustrated in Figure 1.

Figure 1. Project Governance Structure

Formulation Process

The formulation process for the Framework commenced in late 2017, with background research that included a literature review of overseas experience to understand international G/G development and service models, as well as situation analysis of the local G/G service landscape, particularly with regard to HA's existing services.

Following this, a series of consultation activities were carried out to engage key stakeholders in identifying the key issues of HA's G/G services and make suggestions on the strategies for addressing them. A highly participative and broad engagement approach was adopted with the involvement of frontline healthcare staff and representatives from Coordinating Committees (COCs), Central Committees (CCs), HKU, CUHK, DH CGS, as well as HA executives.

One of the consultation activities that kick-started the deliberation process was a workshop conducted in April 2018 with Dr Ron Zimmern, a renowned expert from the United Kingdom who had previously carried out a consultancy study regarding G/G services in Hong Kong[15]. Besides discussion and exchange of views on the current issues in HA's G/G services and possible ways to address them, there was also sharing on overseas experience. The workshop was attended by around 50 key stakeholders involved in the planning and delivery of G/G services, including representatives from various COCs/CCs, HKU, CUHK, DH CGS, as well as executives from the Clusters and Head Office.

15 *A Review of Genetic and Genomic Services in Hong Kong* (2011) – An internal consultancy report.

Following the workshop, a specialty-based questionnaire survey was carried out to gather information about HA's clinical and laboratory G/G services as well as the different specialties' aspirations for future G/G service development. The specialties covered were Pathology, O&G, Paediatrics, Clinical Oncology, Internal Medicine, Surgery and Psychiatry. The questionnaires were also sent to HKU, CUHK and DH CGS to better understand their service provision. Subsequent to this, meetings were organised with relevant COC/CC representatives and survey respondents to discuss the survey findings and further explore the service gaps and future developments.

In parallel, hospital visits were conducted to better understand the G/G service operation and facilities at the pathology departments of Queen Elizabeth Hospital (QEH), PMH and Pamela Youde Nethersole Eastern Hospital (PYNEH). Site visits were also made to HKU, CUHK and DH CGS to learn more about their services and facilities, as well as the research being undertaken by the universities.

The Working Groups and Sub-groups set up under the Taskforce on GGS SSF held meetings for detailed discussions in their respective areas, which included strategies for developing HA's G/G services, care pathways and hospital role delineations, as well as the development of illustrative examples for cancer, prenatal and paediatric G/G services.

The findings and recommendations gathered from the above-mentioned consultation activities were put forward to the Taskforce for formulating the Framework. Reports were also made to the Directors' Meeting and MSDC on a regular basis, with direction and advice sought from members.

Consultation on the draft Framework was conducted in June 2019 to solicit feedback and suggestions from key stakeholders. These included senior management, frontline healthcare staff, relevant COCs/CCs, and representatives from HKU, CUHK and DH CGS who had participated in the formulation process. The comments and feedbacks received were carefully considered and deliberated by the Taskforce. Subsequently, the refined framework was submitted to the Directors' Meeting for endorsement, followed by the MSDC for final approval before publication.

Overview of Genetic and Genomic Services in HA

What We Are Doing Now

As mentioned before, due to the historical development of public clinical genetic service in Hong Kong, there is no dedicated clinical department providing G/G services in HA. Rather, HA's G/G services have mostly developed at the local level and on an independent basis by individual clinicians or hospitals according to the interest and expertise of the respective staff and local needs, and have largely been driven through laboratory initiatives. As a result, there is wide variation in the types and complexity of G/G services provided across all seven Clusters, and overall the provision in HA is very limited.

This chapter provides a brief overview of HA's G/G services, covering governance, service scope as well as workforce and training.

Governance

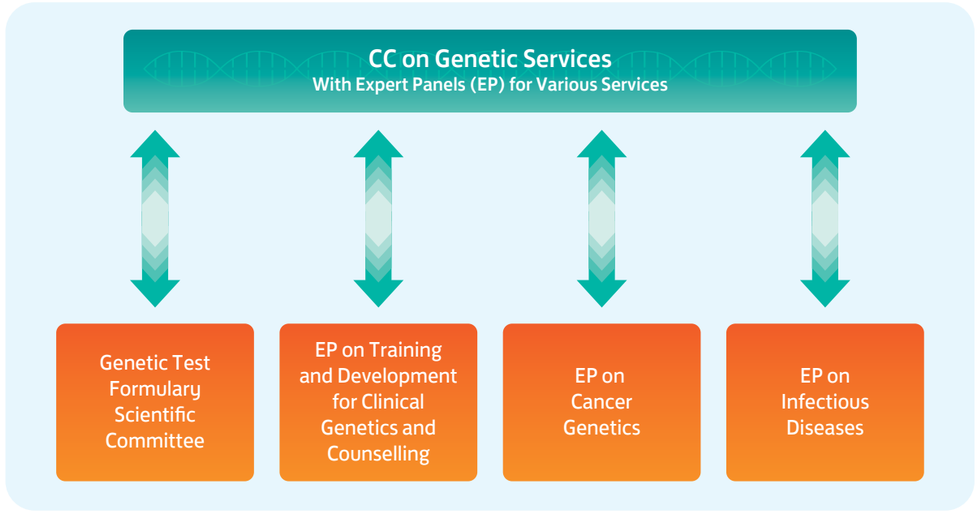
At the corporate level, CC(Genetic Services) is responsible for providing advice on the development of clinical and laboratory G/G services in HA, as well as workforce and training needs. Its membership is cross-specialty and comprises representatives from the COCs in Pathology, O&G, Paediatrics, Clinical Oncology and Medicine, as well as from the Allied Health Committee of Pathology and CC(Diabetic Service). There are also representatives from HKU, CUHK and HKCH, as well as a co-opted member representing DH CGS. Its membership and terms of reference are set out in Appendix 8.

Under CC(Genetic Services), four Expert Panels (EP) have been established, which are (i) Genetic Test Formulary Scientific Committee[16], (ii) EP on Training and Development, (iii) EP on Cancer Genetics, and (iv) EP on Infectious Diseases (Figure 2).

16 The HA Genetic Test Formulary is a list of the G/G tests available in HA hospitals and DH CGS, posted on HA's intranet (http://gtf.home/ha_genetic_test_formulary.aspx).

At the Cluster or hospital level, there are currently no committees set up specifically for the governance of G/G services. Hence, deliberations on the introduction of new G/G services or tests are mostly carried out at the local department level.

Figure 2. Structure of CC(Genetic Services)



Service Scope

Clinical Service

Across HA, the pace and level of adoption of G/G varies markedly among the different clinical specialties, reflecting their individual service needs as well as clinical interests. Specialties that are key providers of G/G services in HA include O&G, Paediatrics, Clinical Oncology, and Medicine. While the specific repertoire of G/G services depends on the specialty, the major scope currently available in HA covers prenatal genetics, newborn screening, hereditary diseases, cancer care and pharmacogenomics, which is summarised in Table 1.

As far as genetic counselling is concerned, with the exception of O&G for prenatal genetic services, it is generally not provided by the various specialties, although some clinicians and nurses may provide G/G information to patients if needed.

Table 1. Major Scope of Clinical G/G Services in HA

Scope	Service
Prenatal Genetics	Prenatal Down syndrome screening provided as part of antenatal care for pregnant women at HA hospitals.
	Screening for thalassemia. If both parents-to-be have thalassemia, diagnostic tests of the foetus will be conducted to determine whether the disease has been passed on.
	Basic genetic counselling, if needed, is provided by O&G clinicians, nurses or midwives depending on the case complexity and professional experience.
Newborn Screening	Expanded Newborn Screening Programme for Inborn Errors of Metabolism (IEM), which is a Government funded programme covering 24 IEM conditions. Blood specimens are sent to HKCH for testing, and babies with uncertain or abnormal screening results are followed up by HKCH's metabolic medicine team.
Hereditary Diseases	Diagnosis using molecular testing to confirm the genetic basis of IEM diseases and for single gene diseases such as Huntington disease, cystic fibrosis and muscular dystrophy. The service is highly site-specific, depending on the awareness and expertise of the clinical team.
	For adult IEM, PMH is currently the designated centre for Enzyme Replacement Therapy for lysosomal storage disorders in new adult cases.
Cancer Care	Diagnosis, prognosis and targeted drug therapy guided by genetic profiling of a patient's cancer, mainly for breast, colorectal, lung, melanoma and gynaecological cancers, as well as certain blood cancers and brain tumours.
Pharmacogenomics	Guided prescribing using biomarker tests to determine whether a drug will be effective for a particular patient and the right dosage, or whether there is a risk of adverse reaction based on information about specific genes in the patient.
	The service is quite limited in HA, and mainly applies to the prescription of allopurinol (for decreasing uric acid levels), carbamazepine (anticonvulsant) and azathioprine (immune-suppressant).

Laboratory Service

The development of laboratory G/G service in HA has been hospital-specific. The tests are often developed in response to *ad hoc* requests from clinicians, and many a time using baseline budgets to meet the requests. According to the HA Genetic Test Formulary, there are 10 hospitals with 18 laboratories across HA that are providing G/G tests. These involve different pathology disciplines including anatomical pathology, chemical pathology, haematology, immunology and microbiology, except for the laboratory in Tsan Yuk Hospital (TYH) which is run by the O&G Department. In addition, the pathology department at the recently commissioned HKCH is also providing G/G tests. These 11 hospitals are indicated in Figure 3.

Figure 3. HA Hospitals with Laboratories Providing G/G Tests*

* Based on the HA Genetic Test Formulary, and also including HKCH

† The Laboratory at TYH is run by the O&G Department

The laboratory G/G services available in HA can be broadly categorised as cytogenetic[17], molecular, haematological and biochemical, which mainly support clinical investigations into cancers, chromosomal abnormalities and inherited diseases, as well as miscellaneous applications like specimen identification. Within these categories a wide spectrum of different tests are provided. These include routine tests conducted at most of the major hospitals, e.g. genetic profiling of certain cancers and pharmacogenomics tests to inform on the use of drugs; and more specialised tests that are only available at particular hospitals / laboratories. For instance, G/G tests related to transplantation[18] and immuno-genetics[19] are concentrated at QMH, while specific gene panel tests for genetic conditions are provided at PWH, PMH and QMH. Meanwhile, prenatal screening and diagnostic tests are mainly provided by the laboratories at TYH and PWH run by the O&G departments.

The different pathology departments may send specimens to other HA pathology laboratories for testing if the test is not available in-house. The referral patterns for sending specimens out for testing vary by department, although the hotspots for receiving test requests are the laboratories in QMH, QEH and PWH, which generally reflect the more comprehensive range of G/G tests available at these hospitals. They are also the network laboratory centres for cytogenetic tests for blood cancers, covering Hong Kong Island, Kowloon and the New Territories respectively. Moreover, following the service commencement of HKCH in late 2018, it has been designated to provide laboratory testing for the Newborn Screening Programme for IEM.

In terms of facility set-up, most of the pathology departments have organised their facilities in different ways for the provision of G/G tests. For example, some pathology departments have established dedicated laboratories to concentrate and share equipment / technology, like NGS machines, among the different pathology disciplines, while others have separate discipline-based laboratories for G/G tests. Only Hong Kong East Cluster has streamlined its G/G laboratory service through a Cluster-based centralised pathology department and molecular laboratory at PYNEH. Overall, the laboratories are recognised by the relevant accreditation bodies for performing specific G/G tests and participate in external quality assurance programmes, as well as undertake their own in-house service monitoring.

17 Cytogenetics is the study of chromosomes and their role in heredity. Based on Nature Journal. www.nature.com/subjects/cytogenetics
18 Pre-transplantation evaluation of patients is carried out using immuno-genetics to optimise donor matching in blood, tissue and organ transplants.
19 Immuno-genetics is the study of the genetic basis of the immune response. Based on Nature Journal. www.nature.com/subjects/immunogenetics

Referral to Other Service Providers

HA patients requiring G/G services may be referred to other service providers, such as DH CGS, HKU and CUHK, when the required clinical expertise or laboratory tests are not available in-house. Specimens may also be sent to overseas laboratories for specialised testing if the G/G tests are not available locally. However, the referrals are generally made at individual staff's own accord and through informal links rather than based on standard protocols or formal networks. Examples include referrals to HKU's research-based services for breast and ovarian cancer syndrome, as well as Lynch syndrome; and referrals to HKU and CUHK for investigations into uncommon or undiagnosed diseases.

Workforce and Training

Currently, there are no Clinical Geneticists in HA, with services mainly provided by clinicians in the individual specialties. As for genetic counselling, while the training requirement of genetic counselling is not well defined in Hong Kong, there are a number of clinicians and nurses in HA providing basic genetic counselling, mainly in prenatal genetic services.

With regard to laboratory services, genetic tests are generally provided by the different pathology disciplines, involving pathologists with special interest or expertise in G/G, as well as relevant laboratory technical staff. There is currently one Bioinformatician recruited for HKCH in the grade of Scientific Officer (Medical), for developing algorithms to analyse and interpret genomic data generated from advanced technology like NGS.

In terms of training, over the past few years HA has rolled out specific training around G/G for its staff. For instance, Corporate Scholarship Programme for Doctors as well as Commissioned Training to beef up the skills and knowledge among clinicians and nurses in areas such as clinical genetics and genetic counselling respectively, as well as among pathology staff in G/G laboratory testing.

Key Issues in HA's Genetic and Genomic Services

What We Need to Improve On

Based on the analysis of the current situation and inputs gathered from the consultation exercise with key stakeholders, key issues have been identified in HA's G/G services, so that strategies could be formulated in this Framework to address them. The issues, which are delineated in this chapter, pertain to the following five major areas for improvement: (i) service organisation; (ii) financial support; (iii) governance; (iv) talent and expertise; and (v) performance monitoring.

Service Organisation

No Structured Clinical Genetic Service in HA

The absence of a structured clinical genetic service in HA has resulted in the lack of coordinated development of clinical and laboratory G/G services by individual hospitals at the local level. As a result, there are significant service gaps in HA, such as for hereditary cancer syndrome and uncommon or undiagnosed disorders. Patients with these G/G service needs often need to be referred to service providers outside HA for diagnosis and genetic testing and counselling.

Moreover, as most of the clinical genetic expertise lie outside of HA in the universities and DH CGS, many clinicians encounter significant difficulties in accessing timely advice and support for G/G cases under their care, particularly in the inpatient setting because DH CGS services are mainly provided on an outpatient basis.

Inequitable Access to G/G Testing

Because pathology departments have developed G/G tests independently based on local needs, hospitals and Clusters in HA have a different repertoire of G/G tests. Moreover, as G/G tests are often developed using baseline budgets to meet *ad hoc* clinical needs and are not HA standard service, they may not be widely publicised or are only available as self-financed items (SFI). As a result, clinicians have to call around different hospitals to find out what tests are available, and patients in different hospitals or Clusters do not have equitable access to the testing. Although the HA Genetic Test Formulary was set up a few years ago to help promulgate information about available tests, it is not updated on a regular basis and the information is not orientated for clinicians' needs.

At the same time, some advanced or specialised G/G tests may only be available outside of HA or even Hong Kong, such as in overseas laboratories. However, there is currently no formal channel or mechanism to coordinate such testing or referrals, which further exacerbates the variability in access.

Mismatch between Clinical Needs and Laboratory Support

In addition to inequitable access to G/G testing, there is a mismatch between the provision of laboratory tests and overall clinical needs, particularly in the inadequate development of pharmacogenomics. For example, the introduction of biomarker tests has not kept pace with the increasing use of targeted cancer therapy. As a result, some of the cancer drugs are not usable in the absence of biomarker tests which are often not introduced along with new drugs added to the HA Drug Formulary (HADF). Patients often have to rely on sponsorship from pharmaceutical companies for receiving the tests provided by private laboratories.

The management of some cancers is also lagging behind international guidelines due to the unavailability of G/G tests, such as for minimal residual disease[20] (MRD) monitoring in leukaemia. Moreover, many advanced diagnostic tests, such as whole exome sequencing (WES) to facilitate time-sensitive patient care decisions, are only available outside of HA or as SFI.

Inadequate Formal Network Arrangements

There are very few standard protocols or test criteria for G/G services in HA, except for Government-funded programmes like the prenatal Down Syndrome Screening and Newborn Screening for IEM. Therefore, referral practices are considerably variable and haphazard across hospitals, even within the same specialty, and often rely on informal links between individuals or departments rather than on formal networks and referral criteria. It means the referrals are highly dependent on individual staff and carried out on an *ad hoc* basis.

The referrals from HA to other G/G service providers are also not based on a formal network arrangement. Furthermore, without standard protocols it is not always clear how follow-up care could be provided, such as long-term surveillance for monitoring the condition of family members who are screened positive for hereditary cancer syndrome by HKU. Along with the absence of a formal channel or mechanism to coordinate referrals to overseas laboratories, the *ad hoc* arrangements contribute to inequitable access to G/G services as well as result in varying levels of care across HA for patients with the same condition.

20 Minimal residual disease (MRD) refers to the small number of leukaemic cells (cancer cells from the bone marrow) left in the body after treatment, and is the major cause of relapse.

Financial Support

Variable Funding Mechanism for G/G Tests Received by Patients

Largely as a result of the independent development G/G tests across HA as well as unavailability of certain tests as HA standard service, there are variable funding arrangements for G/G tests received by patients. The funding mechanisms can include HA funding, SFI for advanced diagnostics tests, funding by universities, and sponsorship by pharmaceutical companies for biomarker tests provided by private laboratories to inform on the use of drugs.

In addition, some HA laboratories may cross-charge other HA hospitals and departments for tests that have been referred to them. This is mainly because they are developing and providing the tests using their baseline budgets without additional resources. An underlying reason is that funding for new G/G tests and technology has to be vetted and bid through the annual planning mechanism, which involves a long lead time and cannot keep up with the rapid G/G development.

Both clinicians and pathologists alike find the cross-charging arrangements an administrative burden and would prefer a simpler system. For example, there are instances where departments would rather refer cases out to DH CGS for free testing than dealing with cross-charging, despite the long waiting time for assessment and the need for DH CGS to refer the patients back to HA for follow-up.

Governance

Sub-optimal Coordination

Over the years, CC(Genetic Services) has been the only platform for coordinating HA's G/G services. Despite the cross-specialty membership of CC(Genetic Services), coordination of the specialties and hospitals providing G/G services as well as between the clinical and laboratory services is suboptimal. For instance, the Genetic Test Formulary Scientific Committee under CC(Genetic Services) appraises new G/G tests for adoption and inclusion in the HA Genetic Test Formulary, but it does not prioritise them for clinical use. Besides CC(Genetic Services), Annual Plan proposals relating to new G/G services may also be submitted separately by different specialties, COC(Pathology) and Cluster hospitals. As a result, the G/G tests and technologies are not introduced into HA in a systematic or coherent manner, and they are often not in sync with the overall clinical need.

Given the rapid advances and increasing application of G/G in clinical practice, as well as the issues in aligning clinical and laboratory services, there is a need to review the existing coordination and governance structure to facilitate the timely and systematic provision of G/G services by HA.

Mechanism for Introduction of New G/G Services Not Keeping Pace with Clinical Needs and Development

There is currently no mechanism for evaluating when to transpose research-based services into HA, such as the clinics for hereditary cancer syndrome currently run by the HKU teams, and some specialised G/G tests developed by university laboratories. This means well-evidenced services or tests from the academic sector are not transferred into mainstream clinical practice in a timely manner, which could otherwise have benefitted a wider number of patients. All these have contributed to HA's G/G services falling behind international developments. A case in point is the yet-to-be introduced non-invasive prenatal testing (NIPT) using NGS technology as a second-tier screening test for Down syndrome, which was developed by CUHK years ago and now widely available internationally. In view of the fast development in G/G technology, collaboration with academics will be essential for HA to leverage advanced technology in providing evidence-based medicine.

Talent and Expertise

Limited G/G Literacy and Expertise in HA

As far as specialised G/G expertise is concerned there is no "Clinical Geneticist", "Genetic Counsellor", or "Bioinformatician" establishment in HA. Internationally, these are important roles in clinical genetic services and have defined skills and competency criteria. What is more, the role and duties of staff involved in HA's G/G services are not well defined and there are different interpretations among departments on what they should entail. It is also unclear who should perform certain tasks, such as genetic counselling or the ordering of tests. For instance, among clinicians and specialties there are different interpretations about genetic counselling, including what it entails, who should provide it and the skills involved. As a result, there is variability across HA on what staff are involved in G/G services and to what extent.

In addition, there is inadequate genetic literacy among healthcare staff. Many of them may not be able to manage common G/G cases themselves, or recognise conditions with a genetic basis and know when to offer G/G tests or refer patients out for specialised assessment and tests, which may contribute to delays in diagnosis and optimal treatment.

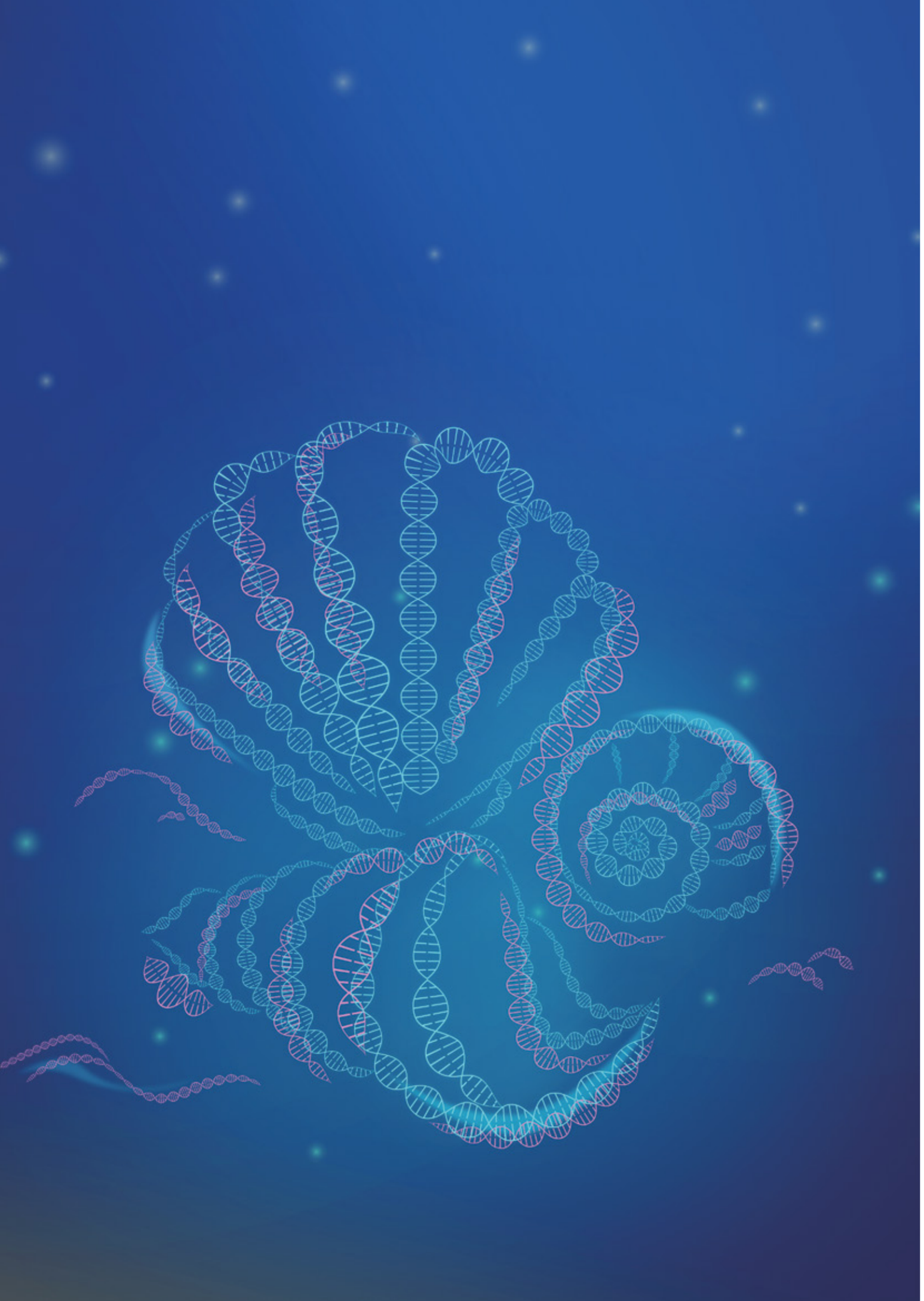
Compounding the issue, the currently siloed service development and delivery means there are dispersed pockets of G/G knowledge and experience across HA. Clinicians in one hospital may be unaware of similar cases being managed elsewhere, such as for uncommon disorders. Together with the absence of a central information or data source to facilitate knowledge sharing on G/G conditions, it means opportunities are lost for collective learning and in some instances may contribute to delays in diagnosis where doctors may not recognise a disorder they have not seen before.

At the same time, different interpretations on the roles and duties have created problems in the planning and development of training to match with service requirements. For example, some frontline staff members have undertaken G/G training, such as via commissioned training programmes, but they have no opportunity to put the skills learnt into practice at their workplace.

Performance Monitoring

Lack of Systematic Monitoring to Inform on G/G Service Improvements

At present there is no systematic monitoring of HA's G/G services in terms of quality and performance. The absence of routine data is a barrier to identifying service gaps and bottlenecks in the service to facilitate targeting of resources and improvement measures. With the exception of the O&G specialty (which regularly reports on the Down syndrome screening programme), most service monitoring is performed at the department level and is not standardised across HA. Standardised indicators have yet to be developed for systematic performance monitoring, to inform on continuous quality improvement and service planning.



Strategic Service Framework for Genetic & Genomic Services

What We Are Going to Do

To address the key issues identified in HA's G/G services, this Framework sets out five strategic directions for HA to develop and improve its clinical and laboratory G/G services. Under each direction a number of interrelated strategies have been crystallised, which together will help HA move towards its vision of providing structured and coordinated G/G services that are evidence-based and keeping pace with advances in G/G development, through professional staff with the relevant skills and expertise to meet patients' healthcare needs in a timely and equitable manner.

The strategic directions of the Framework outline the broad directions HA will pursue in developing its G/G services, while the strategies map out what needs to be done. In parallel, key enablers, specifically IT support, to facilitate effective implementation of the strategies are also highlighted. Presented below is the overall Framework, which illustrates how the strategies relate to the key areas for improvement in HA's G/G services, while details of the strategic directions and the associated strategies are outlined subsequently in this chapter.

Areas for Improvement *(What we can do better)*	Strategic Directions *(Where we are going)*	Strategies *(How we will get there)*
Service Organisation	Strengthen the coordination and collaboration of different G/G services to enhance service quality and accessibility	Develop structured and coordinated G/G services by organising different services based on a tiered approach
		Establish collaborative G/G service networks according to a programme-based hub-and-spoke service model
		Develop a G/G Service Directory to facilitate standardised service provision and information sharing
Financial Support	Provide timely financial support for G/G service provision and development to help keep pace with G/G advancements	Establish a designated fund to expedite the development and introduction of G/G tests
		Develop programme-based funding to support the collaborative G/G service delivery model
Governance	Enhance the governance of G/G services for better coordination	Strengthen the governance structure and process for overseeing G/G service provision and development
		Establish a central mechanism for assessing and prioritising the introduction of new G/G services in a timely and systematic manner
Talent and Expertise	Nurture a skilled and competent G/G workforce in HA	Set out the competency requirements and build up relevant expertise for the delivery of advanced G/G services
		Take steps to raise the G/G literacy of healthcare staff for enhanced awareness and capabilities
Performance Monitoring	Promote performance monitoring for continuous quality improvement	Identify key domains and develop indicators for evaluating and monitoring patient outcomes and service quality
		Enhance data collection, including standardisation of data capture and alignment of measurement tools

STRATEGIC DIRECTION 1

Strengthen the Coordination and Collaboration of Different G/G Services to Enhance Service Quality and Accessibility

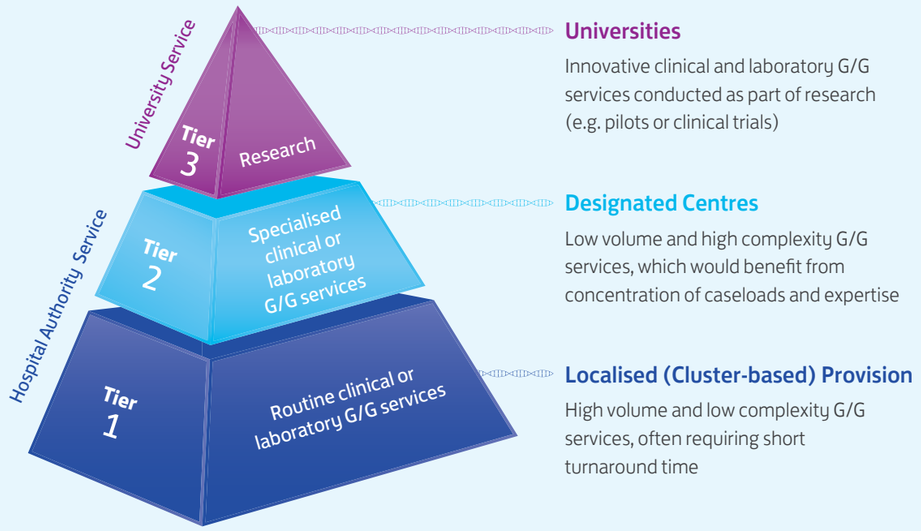
Most of the current issues in HA's G/G services, especially significant service gaps and inequitable access, stem from fragmented service development and provision. Hence, the first and foremost direction is to improve the service organisation by strengthening the coordination and collaboration of different G/G services so that they are more structured and systematic. The relevant strategies for enhancing service organisation are as follows:

- Develop structured and coordinated G/G services by organising different services based on a tiered approach.
- Establish collaborative G/G service networks according to a programme-based hub-and-spoke service model.
- Develop a G/G Service Directory to facilitate standardised service provision and information sharing.

Develop Structured and Coordinated G/G Services by Organising Different Services Based on a Tiered Approach

Based on the principle of "localising where possible and centralising where necessary" in order to enhance service coverage, a tiered approach will be adopted for the organisation of HA's G/G services. In this regard, the services will be organised into three tiers according to service complexity and expertise requirement, as illustrated in Figure 4. Tiers 1 and 2 relate to HA standard service, while Tier 3 pertains to research-based services under the ambit of the universities.

Figure 4. Structured Genetic and Genomic Services Based on a Tiered Approach



Tier 1 comprises routine clinical or laboratory G/G services, which are mostly of high volume and low complexity and often requiring short turnaround time. These will be provided in local hospitals by the relevant specialties through a Cluster-based approach. Examples of Tier 1 G/G services include pharmacogenomics, targeted cancer therapy, and routine prenatal screening.

Tier 2 consists of specialised clinical or laboratory G/G services, which are typically of low volume and high complexity. In this regard, the services will be provided at designated centres to facilitate the concentration of caseloads and expertise. Currently in HA, the clinical services that would fall within this Tier are not well developed, but future examples could include clinics for hereditary cancer syndrome, minimal residual disease (MRD) testing for certain cancers, diagnosis and genetic counselling for uncommon or undiagnosed disorders, etc.

Tier 3 involves innovative clinical and laboratory G/G services, which are mainly provided in the teaching hospitals (i.e. QMH and PWH) by the universities as part of research. In general, these include pilot programmes or clinical trials. Examples of innovative services yet to be introduced, but which may be of possible relevance, include gene- or cell-based therapies like CAR T-cell therapy[21] for certain cancers. Another example is collaboration with the HKGP, subject to the project details to be determined by FHB's Steering Committee on Genomic Medicine.

21 CAR T-cell therapy (Chimeric antigen receptor T-cell therapy) is a novel technique to genetically engineer T-cells with the new ability to target specific proteins, which in some overseas countries has been applied to help treat certain patients with leukaemia or lymphoma.

The tiered approach facilitates the stratification of patients during the course of their care according to their level of G/G service needs, and will help to match them with services managed by healthcare professionals with the right skills and expertise. Underpinning this will be a structured referral mechanism and standard referral criteria to streamline care pathways and help reduce variations in the level of care for patients with the same condition.

The tiered approach also provides a structure through which services can move across tiers over time, including the translation of research-based services into HA and the movement of services from designated centres to local provision as they become more commonly used in mainstream practice. In this regard, the services would generally move from Tier 3 to Tier 2, and from Tier 2 to Tier 1.

Establish Collaborative G/G Service Networks According to a Programme-based Hub-and-Spoke Service Model

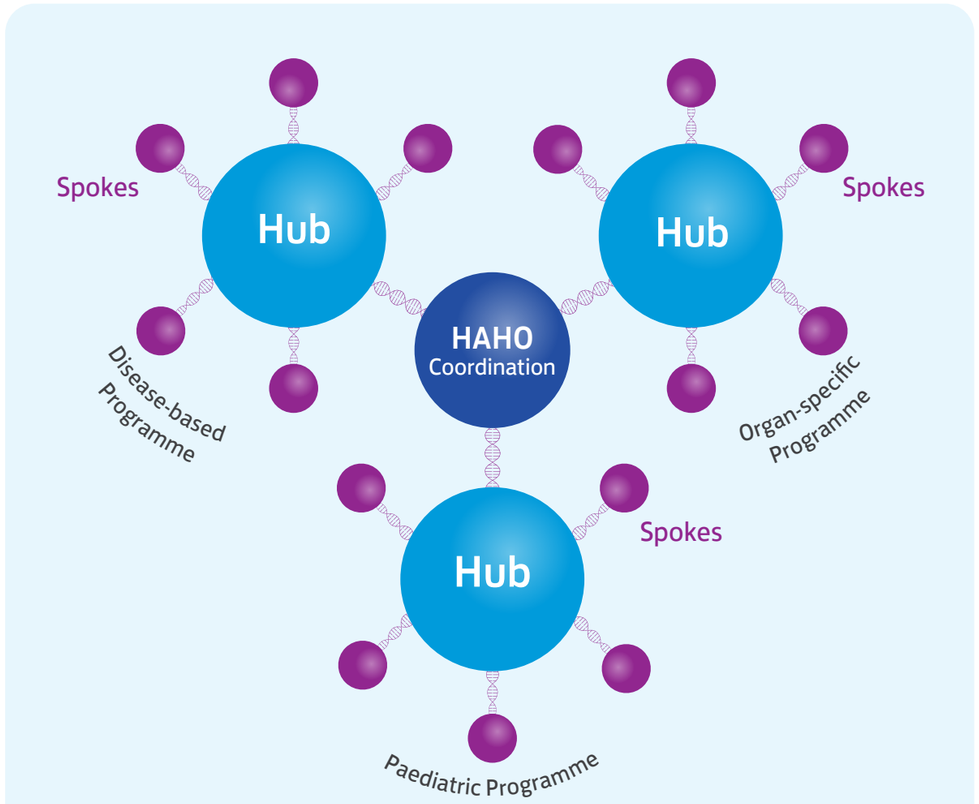
A programme-based hub-and-spoke service model will be adopted to organise the different tiers of G/G services, particularly Tiers 1 and 2 services, into coordinated networks. In the model, the "hubs" are Tier 2 designated centres providing specialised services, while the "spokes" are Tier 1 routine services provided locally.

The different networks could be grouped under the following three themes:

- Paediatric Programmes – G/G services covering pregnancy, infancy, childhood and adolescence, e.g. prenatal screening and diagnosis, newborn IEM screening, diagnosis and genetic counselling for children with uncommon or undiagnosed disorders.
- Disease / Condition-based Programmes – G/G services for diseases affecting a range of organs or are more systemic in nature, such as cancer, adult IEM, haematological and immune disorders.
- Organ-specific Programmes – G/G services for diseases affecting discrete organs or systems of the body, e.g. heart, brain and nervous system, kidney, digestive system and musculoskeletal system.

HA Head Office will act as the central coordinator and sponsor for the programmes, with each programme working out its own hub-and-spoke service arrangement. Illustrated in Figure 5 is the overall service model.

Figure 5. Illustration of the Programme-based Hub-and-Spoke Service Model



Develop a G/G Service Directory to Facilitate Standardised Service Provision and Information Sharing

In order to support structured and standardised service provision as well as facilitate the dissemination of service information, a G/G Service Directory will be established. To be developed by CC(Genetic Services) and orientated for clinicians' use, the Directory will supersede the HA Genetic Test Formulary and serve as the definite up-to-date reference on where HA funded G/G clinical services and laboratory tests are available under the tiered approach and programme-based model. It will be accessible online in electronic version.

By detailing the criteria for different G/G tests, including the clinical indications for testing as well as who can order the tests, the Directory will help support clinicians in ordering appropriate tests. In addition, the testing and referral criteria will also help to strengthen gatekeeping at the point-of-use, so that G/G services are used safely and judiciously.

STRATEGIC DIRECTION 2

Provide Timely Financial Support for G/G Service Provision and Development to Help Keep Pace with G/G Advancements

The development of structured and accessible G/G services in HA requires appropriate planning and allocation of financial resources. To tie in with the fast pace of G/G service development and the reengineered service model, HA will pursue the following dual strategies:

- Establish a designated fund to help expedite the introduction of new G/G tests into HA.
- Develop programme-based funding to support the collaborative service model.

Establish a Designated Fund to Expedite the Development and Introduction of G/G Tests

To address the issue of laboratories having to develop new G/G tests using their existing resources due to the long lead time involved in the annual planning cycle, HA will introduce a designated fund for allocation to the relevant departments on a time-limited basis to pilot and evaluate new tests. Not necessarily limited to one year, the time-limited funding would help to bridge the gap where there is a need to rapidly develop a new G/G test to meet clinical need. Recurrent funding, if needed, could subsequently be sought through the annual planning process.

Overall, the designated funding provides a more consistent and transparent basis through which laboratories could be supported in expediting the introduction of new G/G tests into HA and keeping in pace with the G/G development internationally.

Develop Programme-based Funding to Support the Collaborative G/G Service Delivery Model

To tie in with the new service model and facilitate equitable access, HA will move towards a programme-based funding arrangement for its G/G services. Under the approach, funding will be distributed among the clinical departments and laboratory services involved in a specific G/G programme for meeting their operational needs. This will include the allocation of funding to designated centres for providing specialised clinical services or laboratory tests across HA so as to avoid the need for cross-charging.

Furthermore, in order to enhance the synchronisation of laboratory G/G tests provision with clinical needs, HA will take steps to provide and fund the relevant biomarker tests for drugs in the HA Drug Formulary.

STRATEGIC DIRECTION 3

Enhance the Governance of G/G Services for Better Coordination

Robust governance is essential to ensure the provision and development of HA's G/G services is carried out in an effective manner with transparency and accountability. Specifically, given the complexity of HA's G/G services with the involvement of a wide range of clinical specialties and pathology disciplines, it is crucial to put in place an effective governance structure and process for their coordination and alignment. At the same time, there is a need to strengthen the governance in determining which innovations or new technologies should be introduced and how to introduce them. The key strategies in this regard are as follows:

- Strengthen the governance structure and process for overseeing G/G service provision and development.
- Establish a central mechanism for assessing and prioritising the introduction of new G/G services in a timely and systematic manner.

Strengthen the Governance Structure and Process for Overseeing G/G Service Provision and Development

To facilitate systematic G/G service provision and development, the existing governance structure will be revamped and beefed up, as illustrated in Figure 6. A newly established Steering Group on Genetic and Genomic Services in HA, chaired by the Chief Executive, will steer the overall development with regard to the strategies laid out in this Framework. The membership and terms of reference of the Steering Group are provided in Appendix 9.

Under the guidance of the Steering Group, CC(Genetic Services) will oversee the development and coordination of G/G clinical services and laboratory tests. In addition, various Expert Panels for clinical and laboratory work streams as well as for the different programmes in the new service model will be set up under the CC(Genetic Services) to provide advice on the development of different G/G services and tests, including consideration of the ethical, legal and social implications[22]. The Expert Panels will include representatives from the universities and DH CGS as appropriate. The HA Head Office will provide executive support and coordination for the governance structure.

22 Examples of the ethical, legal and social implications include how to deal with incidental findings and genetic variants of unknown significance; whether there is a need to revisit a patient's G/G sequence information in the light of a growing knowledge base; and how to manage the implications of G/G findings on a patient's relatives.

Figure 6. The Revamped Governance Structure for HA's G/G Services

Establish a Central Mechanism for Assessing and Prioritising the Introduction of New G/G Services in a Timely and Systematic Manner

Under the governance structure, a central mechanism will be established for assessing and prioritising the introduction of new G/G services, so that HA can keep pace with clinical needs and developments in a timely and systematic manner. On the whole, HA takes an evidence-based approach to the introduction of new technology and is a "consensus adopter," which means implementing health technology that is generally accepted and broadly available in the market. Given the rapid pace of G/G development, the central mechanism will be crucial in enabling HA to be more adaptive and responsive to clinical G/G service needs and help to synchronise the provision of laboratory support.

The mechanism will cover all G/G services that are proposed to be included as HA standard service and which require additional resources, particularly for laboratory tests. At the same time, G/G clinical and laboratory services that are proposed to be transferred from one service tier to another, including the translation of research-based services into HA standard service, will also be vetted through the mechanism. Only those proposals that are endorsed through the governance structure will be incorporated into the G/G Service Directory.

STRATEGIC DIRECTION 4

Nurture a Skilled and Competent G/G Workforce in HA

Fundamental to the provision of HA's G/G services in alignment with the reengineered service delivery model is a workforce with the right mix of skills and competencies. The priority is to strengthen the workforce for delivering the different tiers of G/G services, along with developing clear roles and responsibilities. At the same time, it is important to upskill and improve the general G/G knowledge of the broader healthcare workforce, so that they are more aware of the services available and can make appropriate referrals. Hence, a multifaceted approach will be adopted along the following strategies:

- Set out the competency requirements and build up relevant expertise for the delivery of advanced G/G services.
- Take steps to raise the genetic literacy of healthcare staff for enhanced awareness and capabilities.

Set out the Competency Requirements and Build up Relevant Expertise for the Delivery of Advanced G/G Services

As a first step for developing the G/G workforce, HA Head Office will coordinate the development of the standard skill and competency requirements for delivering the different tiers of the G/G services. This will include the qualifications, professional competencies and associated training for specialised roles.

Under HA Head Office coordination, each G/G programme will participate in determining their own staffing requirements based on service complexity and workload. In general, it is anticipated that the level of staff expertise would align with the G/G service tiers as broadly outlined in Table 2, and in time, a system of credentialing could be introduced to help ensure staff expertise and competency at the relevant tiers.

Table 2. Future Alignment of Staff Expertise with HA's G/G Service Tiers

Staff	Tier 2 (Hubs) Specialised clinical or laboratory G/G services	Tier 1 (Spokes) Routine clinical or laboratory G/G services
Clinicians	Clinicians with specialist qualifications (e.g. Clinical Geneticist)	Clinicians with special interest or experience in G/G
Genetic Counsellors	Genetic counselling by qualified Genetic Counsellors	Basic genetic counselling provided by clinicians and nurses
Pathologists & other Laboratory Staff	Pathologists with special interest or expertise in G/G services	
	Scientific Officers and technical laboratory staff with relevant training and experience according to service complexity	
Bioinformaticians	Bioinformaticians to support advanced genome sequencing (e.g. NGS)	Not Applicable

Since HA currently lacks G/G expertise, the talent pool will need to be built up through a multi-faceted approach. This could include recruiting doctors with specialist G/G qualifications from overseas or locally as required; training nurses and certain allied health professionals to take up genetic counselling roles; leveraging on existing G/G expertise in the universities and DH CGS, such as through honorary appointments, secondments, service partnerships and collaborations; and providing in-house and external training for relevant staff.

Take Steps to Raise the G/G Literacy of Healthcare Staff for Enhanced Awareness and Capabilities

Given the growing application of G/G services in healthcare, it is also incumbent on HA to support the different specialties in raising the G/G literacy of their staff, so that they could manage common G/G cases and know when to appropriately refer to specialised G/G services. This could include targeted engagement and communication activities, such as publicising G/G services at the hospital / department level and using existing platforms like HA Convention, talks and seminars to share relevant information. Each specialty could determine their own training requirements for raising G/G literacy, and how best to disseminate knowledge and learning through their existing networks.

STRATEGIC DIRECTION 5

Promote Performance Monitoring for Continuous Quality Improvement

Systematic monitoring of clinical and laboratory G/G services is important to help drive improvements and ensure the safety, quality and sustainability of the services for maximising patient outcomes. Importantly, it would provide a consistent basis for examining the different programme-based services in order to identify areas of best practice for disseminating across HA, as well as areas that can be improved for organisation-wide learning. In order to promote performance monitoring for continuous quality improvement, the following strategies will be pursued:

- Identify key domains and develop indicators for evaluating and monitoring patient outcomes and service quality.
- Enhance data collection, including standardisation of data capture and alignment of measurement tools.

Identify Key Domains and Develop Indicators for Evaluating and Monitoring Patient Outcomes and Service Quality

As an initial step to developing a more robust monitoring system, HA Head Office will coordinate the identification of key domains for the evaluation and monitoring of patient outcomes and service quality in HA's clinical and laboratory G/G services, taking reference to local and international experiences. For example, this could include domains around patient experience, clinical and laboratory workload / throughputs, waiting times for clinical genetic services and genetic counselling, laboratory reporting times, and adherence to testing criteria. The subsequent development of standardised indicators will facilitate the benchmarking of G/G services across HA to help reduce unwarranted variations in care, identify bottlenecks in service provision, and target areas with room for improvement.

Enhance Data Collection, Including Standardisation of Data Capture and Alignment of Measurement Tools

In order to support the efforts in continuous quality improvement, the various parameters for performance monitoring will need to be deliberated and agreed upon to support systematic data capture. In particular, standard data definitions and automated data collection mechanisms that integrate seamlessly into clinical and laboratory workflows, without the need for additional work by frontline staff, will be important. Collectively, this will help to ensure high quality data for more robust and meaningful analysis.

Illustrative Examples

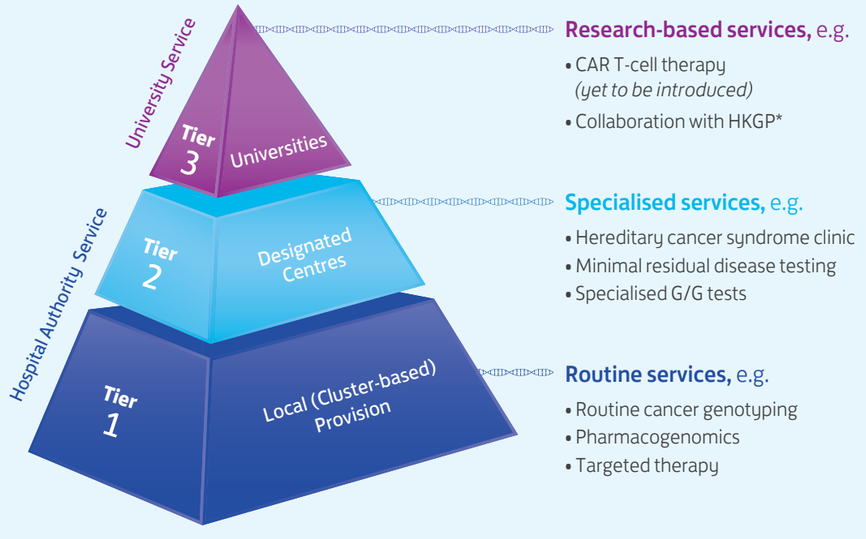
To illustrate the service model as set out in this Framework, a number of specific examples covering cancer as well as prenatal and paediatric G/G services are presented below.

Cancer G/G Services

Cancer is caused by mutations of genes that control the way cells function. Those caused by mutations inherited from the parent(s) are called germline cancers, examples of which include hereditary cancer syndromes like breast & ovarian cancer syndrome and Lynch syndrome. On the other hand, cancers that are not inherited but occur during a person's lifetime are known as somatic cancers, which include lung cancer and leukaemia.

Illustrated in Figure 7 are the different tiers of cancer G/G services in HA according to the future service model. In Tier 1 are routine services like cancer genotyping, targeted therapy and pharmacogenomics. Tier 2 services to be provided at designated centres (i.e. the hubs) include clinics for hereditary cancer syndrome and minimal residual disease testing, which are currently not available in HA. Subject to deliberation, CAR T-cell therapy is an example of research-based services that could be introduced by the universities as a Tier 3 service. Collaboration with the HKGP could also be at Tier 3, depending on the project details to be determined by FHB's Steering Committee on Genomic Medicine.

Figure 7. Future Stratification of Cancer G/G Services in HA



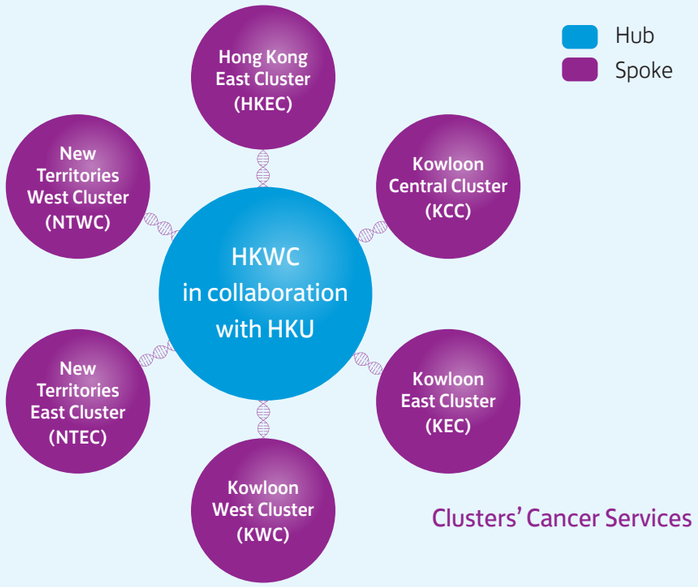
* Subject to details of the HKGP to be determined by FHB's Steering Committee on Genomic Medicine

Examples on Adult Germline Cancer: Breast & Ovarian Cancer Syndrome and Lynch Syndrome

Clinics for breast & ovarian cancer syndrome as well as Lynch syndrome are currently provided by HKU at Tung Wah Hospital and QMH respectively. In the future, the research-based services for these two hereditary cancer syndromes will be translated into HA as a standard service so as to benefit a larger number of patients and their families in a systematic way.

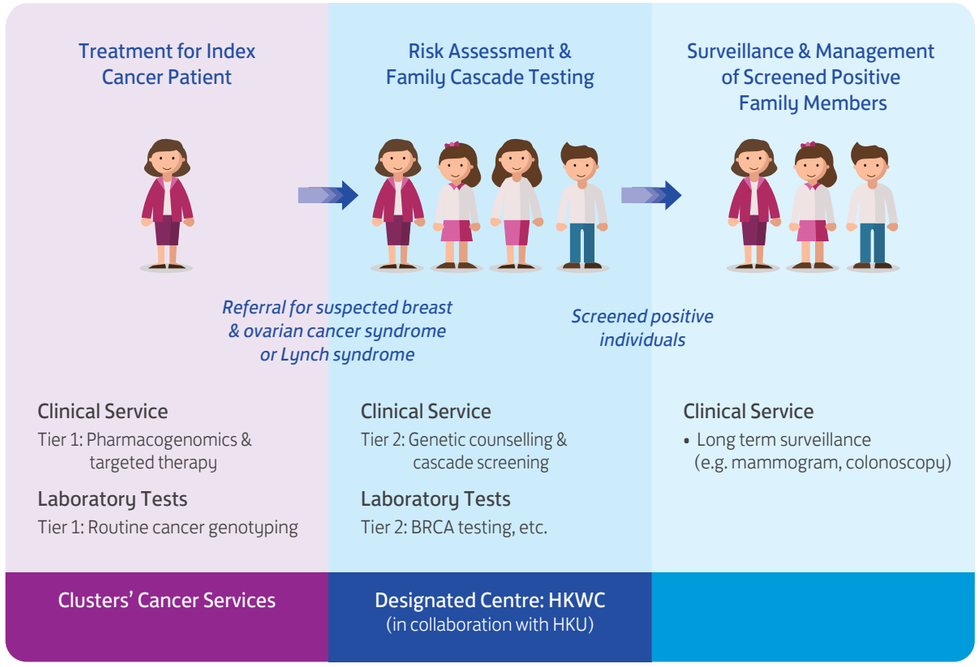
In view of the existing expertise, Hong Kong West Cluster (HKWC) (in collaboration with HKU) will be the designated hub for these two services in the first instance, with the Clusters' cancer services across HA forming the spokes (Figure 8). Depending on future caseloads, the hub-and-spoke model could be further developed taking into account the service demand and local expertise.

Figure 8. Future Hub-and-Spoke Service Model for Breast & Ovarian Cancer Syndrome and Lynch Syndrome



In the future care pathway, as illustrated in Figure 9, the Clusters' cancer services will continue to provide routine diagnosis and treatment for the index cancer patient. If breast & ovarian cancer syndrome or Lynch syndrome are suspected, the patient and his/her family members will be referred to the respective hereditary cancer syndrome clinic at HKWC for risk assessment and family cascade screening according to standard protocols. Long-term surveillance will be carried out for family members who are screened positive for hereditary cancer syndrome, but further deliberation is needed on the service provider(s).

Figure 9. Future Care Pathway for Breast & Ovarian Cancer Syndrome and Lynch Syndrome



Examples on Adult Somatic Cancer: Lung Cancer and Leukaemia

Taking somatic cancers like lung cancer and leukaemia as examples, *clinical* G/G services such as protocol-driven targeted therapy and disease monitoring will continue to be provided locally by the relevant clinical units at Tier 1. However, there will be differentiation of Tier 1 and Tier 2 in the provision of *laboratory* support.

Specifically, in view of the high volume of cases and the routine nature of the existing G/G tests for lung cancer, such as EGFR, ALK and ROS1 testing[23], they are recommended to be provided as a Tier 1 service via a Cluster-based approach. In other words, the cancer centres of the respective Clusters will provide the lung cancer G/G tests. On the other hand, MRD testing for leukaemia is a specialised G/G test which will be provided by designated centres as a Tier 2 service. Taking reference to the current service network for cytogenetic tests for blood cancers as shown in Table 3, it is recommended for QMH, QEH and PWH to be the designated centres for MRD testing.

Nevertheless, new G/G tests for lung cancers will initially be introduced at Tier 2 in designated centres, and may move to Tier 1 later on if appropriate, subject to deliberation through the governance structure for G/G services in HA.

Table 3. Current Networking Arrangement for Cytogenetic Tests for Blood Cancers

Designated Centre	Networked Clusters
QMH	HKEC and HKWC
QEH	KCC, KEC and KWC
PWH	NTEC and NTWC

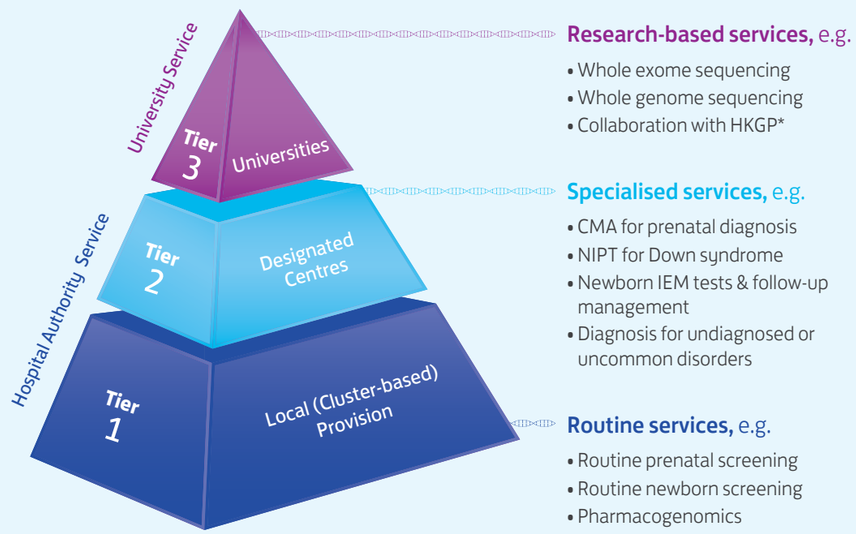
23 These biomarker tests can be used to provide information on the molecular characteristics of a patient's lung cancer, to help guide decisions about targeted therapies and to inform on prognosis. For example, to identify mutations in the Epidermal Growth Factor Receptor (EGFR) gene, or rearrangements in the Anaplastic Lymphoma Kinase (ALK) gene or ROS proto-oncogene 1 receptor tyrosine kinase (ROS1) gene. Based on American Lung Association. www.lung.org/assets/documents/lung-health/what-you-need-to-know-about-tumor-testing.pdf

Prenatal and Paediatric G/G Services

The different tiers of prenatal and paediatric G/G services in HA are illustrated in Figure 10 according to the future service model. Tier 1 services to be provided locally include routine prenatal and newborn screening, pharmacogenomics, etc. In Tier 2 are specialised services to be provided in designated centres, such as chromosomal microarray (CMA)[24] for prenatal diagnosis, Non-Invasive Prenatal Testing (NIPT) for Down syndrome, as well as genetic counselling and testing for uncommon or undiagnosed disorders. Subject to deliberation, whole exome sequencing (WES) and whole genome sequencing (WGS) for genetic disorders are examples of research-based services that could be introduced by the universities as a Tier 3 service. Another possible example is collaboration with the HKGP.

With the service commencement of HKCH and its designated role as a tertiary referral centre for complex, serious and uncommon paediatric cases requiring multidisciplinary management, laboratory testing, diagnosis and family counselling[25], it will be a referral centre for advanced prenatal and paediatric G/G services.

Figure 10. Future Stratification of Prenatal and Paediatric G/G Services in HA



* Subject to details of the HKGP to be determined by FHB's Steering Committee on Genomic Medicine

24 Chromosomal microarray (CMA) refers to methods used to detect copy number variants (losses or gains of chromosome material), which may be benign, pathogenic, or of uncertain clinical significance. A far more sensitive method than traditional karyotyping, CMA detects both large and small copy number variants. Depending on the method used, CMA may involve scanning of the whole genome, targeted regions of the genome, or a specific chromosome or chromosome segment. Based on National Center for Biotechnology Information (NCBI). www.ncbi.nlm.nih.gov/books/NBK5191/#IX-C

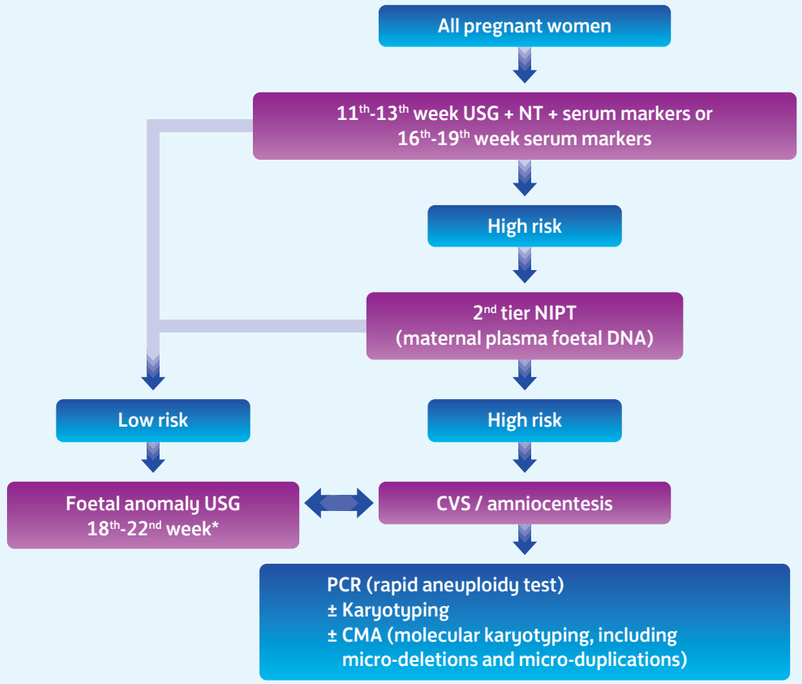
25 Legislative Council Panel on Health Services (2018). *Hong Kong Children's Hospital.* LC Paper No. CB(2)231/18-19(06).

Example: Prenatal Testing and Diagnosis for Down Syndrome

In general for prenatal G/G services, the *clinical* aspects such as routine screening and basic genetic counselling will continue to be carried out locally by the Clusters' Obstetric units at Tier 1. The *laboratory* support, on the other hand, will be provided in designated centres as Tier 2 services.

At present, routine prenatal screening for Down syndrome is a well-established Government-funded programme. In addition, HA is making preparations to introduce NIPT as a second-tier screening test for Down syndrome for high-risk cases. In line with international trend, the advanced technology of CMA has also been introduced into HA for prenatal diagnostic testing. Hence, in future the algorithm for prenatal diagnosis in HA will be as illustrated in Figure 11.

Figure 11. Future Algorithm for Prenatal Diagnosis in HA



* All pregnant women should ideally be offered screening of foetal structural abnormalities
(Source: Hong Kong College of Obstetricians and Gynaecologists – Guidelines on Antenatal Care (Part II), 2008)

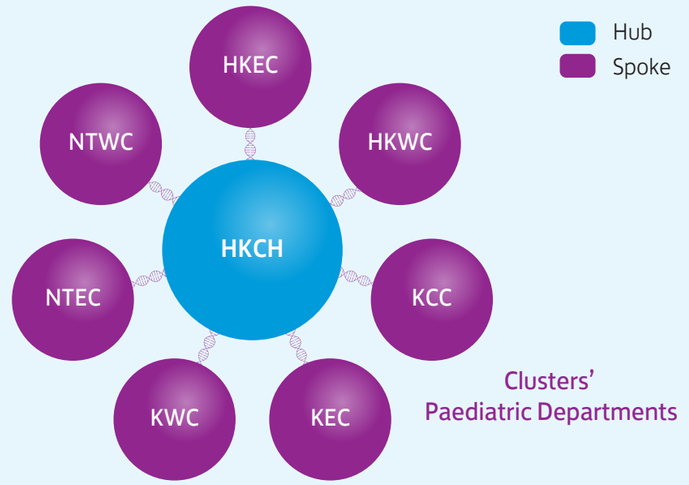
Abbreviations:
CMA = Chromosomal microarray
CVS = Chorionic villus sampling
NIPT = Non-invasive prenatal testing
NT = Nuchal translucency
PCR = Polymerase chain reaction
USG = Ultrasonogram

In the service model, Clusters' Obstetric units will continue to provide routine Down syndrome screening as part of antenatal care, including nuchal translucency ultrasound measurement and maternal serum markers. They also carry out invasive prenatal tests (chorionic villus sampling and amniocentesis) for high risk Down syndrome screening results, and for structural foetal abnormalities detected by foetal ultrasound examination. Meanwhile, the samples are sent to the O&G laboratories at TYH and PWH for screening and diagnostic tests as with the current arrangement. CMA will continue to be carried out by the two laboratories in collaboration with HKU and CUHK respectively. HKCH, on the other hand, will be the designated centre for the NIPT programme. Blood specimens will be sent to the laboratory at HKCH for NIPT as the second-tier screening tests. Genetic counselling will remain to be provided by the Obstetric units, supported by the two universities.

Example: Paediatric Undiagnosed or Uncommon Disorders

In general, HKCH is the designated centre for the assessment and management of uncommon or undiagnosed disorders in paediatric patients (Figure 12). These patients will be referred to HKCH via the Clusters' Paediatric units when more specialised clinical expertise or laboratory tests are required.

Figure 12. Hub-and-Spoke Service Model for Paediatric Undiagnosed or Uncommon Disorders

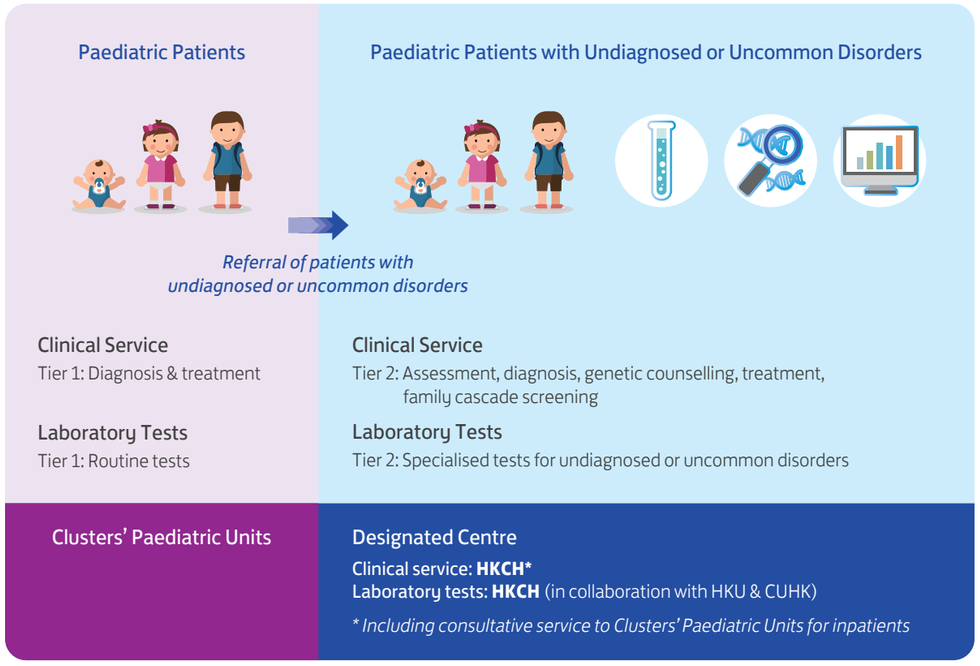


In the future care pathway, the Paediatric units will provide routine services, which could include the diagnostic work up and management of paediatric patients with undiagnosed or uncommon disorders, depending on the local skills and expertise. Paediatric patients that cannot be diagnosed or managed in the Paediatric units will be referred to HKCH for specialised tests, diagnosis, genetic counselling, treatment, and family cascade screening as appropriate (Figure 13). Follow-up care, on the other hand, could either be provided by HKCH or the referring Paediatric unit, depending on case complexity.

While specialised G/G laboratory services will be provided at HKCH, the hospital may also leverage on the existing laboratory expertise at HKU and CUHK if needed. Moreover, for some patients who are unable to be transferred to HKCH for assessment, such as patients in the Clusters' paediatric intensive care units (PICU), or neonatal intensive care units (NICU), HKCH may provide an outreach consultative service for inpatient assessment.

The operational details regarding the care pathways and referral arrangements will be discussed and mapped out during the implementation phase.

Figure 13. Care Pathway for Paediatric Undiagnosed or Uncommon Disorders



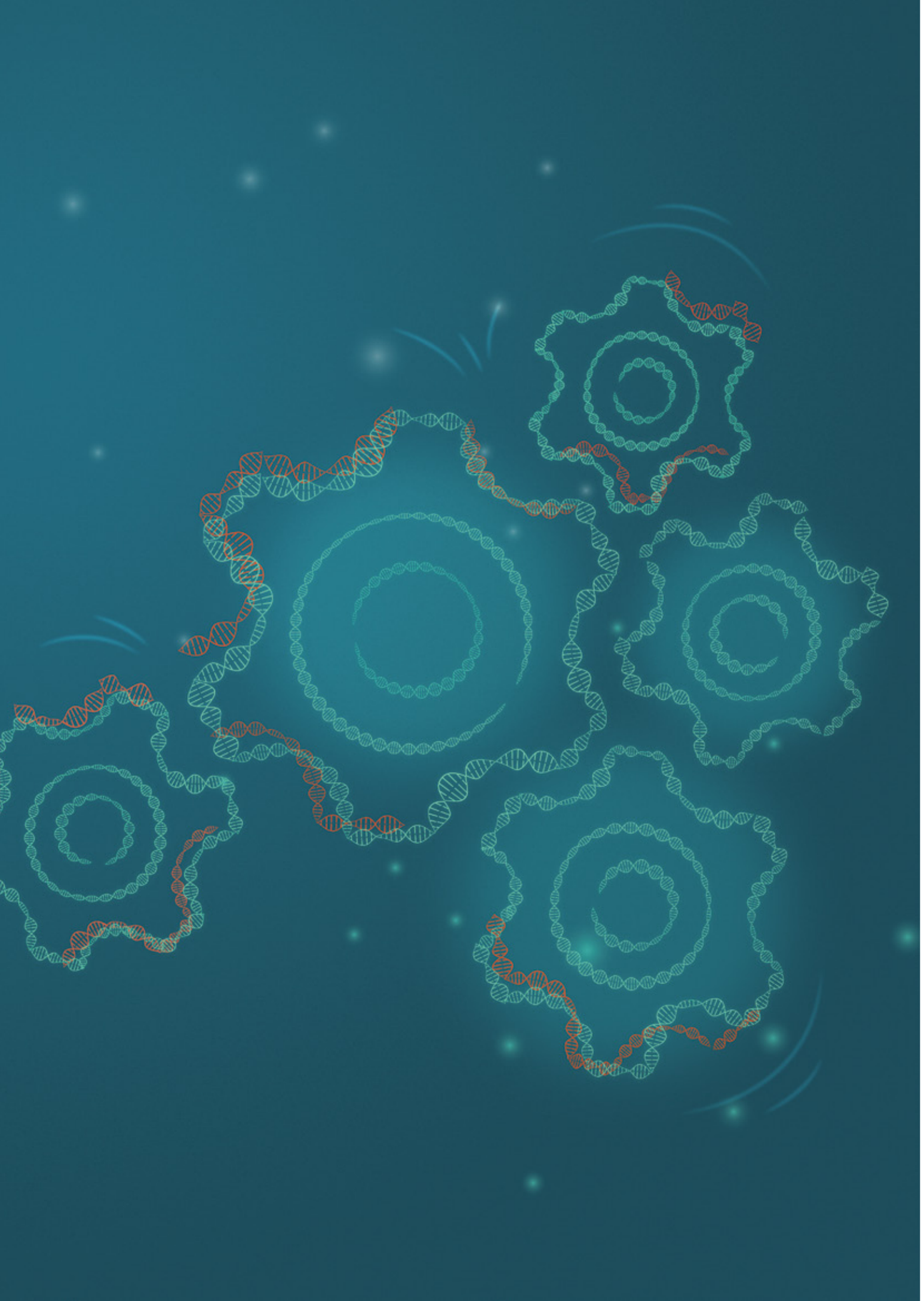
Key Enablers

Successful implementation of the strategies outlined in this Framework will require key enablers, in particular Information Technology (IT), to support the development of the service delivery model. Specifically, IT system infrastructure will be crucial in enabling the workflow, communication and coordination of different clinical specialties and pathology disciplines during the course of a patient's care, especially if the patients are referred between the different tiers of G/G services.

Moreover, as increasing amounts of genomic information are generated by HA there will be growing demands on the IT infrastructure for a unified platform which can securely store huge amounts of genomic data, while at the same time providing appropriate access and the necessary computing power for its analysis and interpretation, as well as tools to support clinical decision-making.

In addition, centralised information portals are required for supporting the dissemination of information and knowledge among healthcare professionals. These include the development of the electronic G/G Service Directory, and database of G/G cases to make it easier for clinicians to know whether similar cases are being managed elsewhere in HA, especially for uncommon or undiagnosed disorders.

Moreover, along with anticipated greater collaborations with community partners, such as with those involved in the HKGP, the ability to share appropriate and relevant patient information electronically with due diligence to data privacy and security, and in line with patient consent, will become of increasing importance.



Implementation and Monitoring

Realising the Plan : Our Short and Medium-term Goals

The implementation of this Framework requires the concerted efforts of various stakeholders and input of different resources. It will be led by the Steering Group on Genetic and Genomic Services in HA, which will report to the Directors' Meeting and MSDC, as appropriate, on the development of G/G services in HA. While the overall directions and strategies are laid out in this Framework, the operational details for the implementation will be worked out by the key stakeholders under the guidance of the Steering Group.

In this chapter, the short and medium-term implementation priorities for the coming 1-3 years and 4-5 years respectively are briefly outlined. Some of the strategies do not require additional resources and could move forward through the efforts and collaborations of the various stakeholders, while other strategies will incur resources to realise. The HA annual planning process is the mechanism through which resources could be sought to support the implementation of the relevant strategies.

Implementation Priorities

Short Term

Strengthen the Governance Structure

Robust governance is essential to the systematic development of G/G services in HA. As the key initial step, the governance structure and processes will be strengthened at the corporate level. Following the recent establishment of the Steering Group on Genetic and Genomic Services in HA, the organisational structure of CC(Genetic Services) will be revamped with the setting up of different Expert Panels, including one for laboratory services and those for different clinical programmes. Executive support from HA Head Office will also be strengthened in order to act as the lynchpin for linking up and aligning the Expert Panels. In parallel, the mechanism for vetting and appraising the introduction of new G/G services and tests will be established under the governance structure.

Develop Programme-based Funding and a Designated Fund

Financial support will be enhanced for the systematic development of G/G services in HA. In this connection, steps will be taken to set up a designated fund to facilitate the timely development and introduction of G/G tests. Programme-based funding will also be developed in support of different G/G programmes under the hub-and-spoke service model.

Define the Skill and Expertise Requirements

In order for the G/G services to develop along the lines of the strategies in this Framework, there is a need to work out the standard skill and competency requirements of staff providing different levels of G/G services in HA, for example genetic counselling. The criteria will help to guide the development of initiatives to upskill staff and beef up the G/G expertise in HA. At the same time, HA will explore with the Hong Kong Academy of Medicine and the relevant Colleges ways to develop G/G training for nurturing the local talent pool.

Medium Term

Enhance Data Collection and Develop Indicators for Performance Monitoring

As the service model shifts toward more structured and standardised care, process and outcome measures should be developed to help monitor the quality of HA's clinical and laboratory G/G services. Initially, this will include identifying key domains for monitoring, and developing standard data definitions, data collection and reporting mechanisms. Subsequently, suitable quality indicators will be identified and further developed for benchmarking, accountability and continuous quality improvement.

Enhance IT Support

As a key enabler, IT support will be enhanced for implementing the strategies outlined in the Framework. In particular, IT will be deployed in the establishment of the electronic G/G Service Directory, and in providing system support for the collection, storage, analysis and sharing of high volumes of G/G data in HA.

Monitoring

The implementation of this Framework is an iterative process of developing and improving our G/G services for the benefit of patients. Monitoring the process is the key to ensuring proper implementation of the strategies and effective use of resources. The success of the strategies in achieving the goal of service development and improvement will be evaluated. The monitoring will be carried out at several levels, as follows:

- Service deliverables tied in with resource bidding via the HA annual planning process will be monitored through the existing mechanism.
- Progress on the key implementation milestones mapped out in the operational plans of the Framework over the next five to ten years will be monitored by CC(Genetic Services), with guidance from the Steering Group on Genetic and Genomic Services in HA.
- Quality indicators will be developed for benchmarking, accountability reporting and continuous quality improvement of the overall G/G service in HA.

Conclusion

Rapid advances in the field of G/G have transformed our understanding of the role our genes play in health and disease. The ability to use G/G information and techniques to guide healthcare decisions and treatment, along with the advancements in knowledge and technology, is ushering in a new era of precision and personalised medicine. It is therefore important for HA to harness the potential of G/G appropriately and judiciously for improving patients' health outcomes.

In this connection, we envision the provision of structured and coordinated G/G services that are evidence-based and keeping pace with advances in G/G development, through professional staff with the relevant skills and expertise to meet patients' healthcare needs in a timely and equitable manner.

This Framework has been developed with the invaluable inputs of frontline staff from different clinical specialties and pathology disciplines in HA as well as representatives of DH, HKU and CUHK. It sets out the key interrelated strategies to achieve the vision. Specifically, the governance and service organisation of our G/G services will be revamped and improved to facilitate better coordination and alignment between clinical and laboratory G/G services. In particular, through a tiered approach and programme-based hub-and-spoke service model, collaborative networks will be established to better match the levels of service provision with clinical needs and improve the accessibility of care. At the same time, financial support will be enhanced to meet service needs, and the relevant workforce developed to ensure that necessary G/G skills and expertise are available in HA.

As the blueprint for our G/G services, this Framework sets out the directions for HA to work towards. It will require the continued support and participation of various stakeholders to realise the strategies that have been laid out, thereby contributing to the overall development of G/G in Hong Kong. Successful implementation of this Framework will not only help address the service needs of today, but also lay the foundations for HA to leverage on the huge potential of G/G innovations and advancements to benefit patient care in the years to come.

Abbreviations

CC	Central Committee
COC	Coordinating Committee
CUHK	The Chinese University of Hong Kong
DH	Department of Health
DH CGS	Department of Health Clinical Genetic Service
DNA	Deoxyribonucleic Acid
FHB	Food and Health Bureau
GGS SSF	Strategic Service Framework for Genetic and Genomic Services
G/G	Genetics and Genomics
HA	Hospital Authority
HKCH	Hong Kong Children's Hospital
HKGP	Hong Kong Genome Project
HKU	The University of Hong Kong
IEM	Inborn Errors of Metabolism
IT	Information Technology
MSDC	Medical Services Development Committee
NGS	Next Generation Sequencing
O&G	Obstetrics and Gynaecology
PMH	Princess Margaret Hospital
PWH	Prince of Wales Hospital
PYNEH	Pamela Youde Nethersole Eastern Hospital
QEH	Queen Elizabeth Hospital
QMH	Queen Mary Hospital
TYH	Tsan Yuk Hospital

Appendices

Appendix 1: Taskforce on the HA Strategic Service Framework for Genetic and Genomic Services

Terms of Reference

- To review the current and future service needs for genetic and genomic services in HA
- To advise on the future service model(s), system infrastructure and monitoring of HA genetic and genomic services to address the existing issues and guide service development over the next five to ten years
- To identify priority areas and develop strategies to enhance the quality and outcomes of HA genetic and genomic services
- To formulate a strategic service framework for HA genetic and genomic services for consideration by members of the Directors' Meeting and Medical Services Development Committee

Membership (as at August 2019)

Co-chairs

Dr Libby LEE	Director (Strategy & Planning), HA Head Office
Dr K L CHUNG	Director (Quality & Safety), HA Head Office

Members

Dr Mary TANG	Honorary Consultant (Obstetrics and Gynaecology), Queen Mary Hospital / Clinical Associate Professor, Department of Obstetrics & Gynaecology, Li Ka Shing Faculty of Medicine, The University of Hong Kong – *Representative of CC (Genetic Services)*
Dr Alex CHAN	Service Director (Pathology), Kowloon Central Cluster / Chief of Service (Pathology), Queen Elizabeth Hospital – *Representative of COC (Pathology) (Anatomical Pathology)*

Dr Michael CHAN	Cluster Coordinator (Pathology), New Territories East Cluster / Consultant (Chemical Pathology), Prince of Wales Hospital – *Representative of COC (Pathology) (Chemical Pathology)*
Dr Michael WONG	Chief Manager, Kowloon West Cluster / Deputy Hospital Chief Executive (Operations), Princess Margaret Hospital / Deputy Hospital Chief Executive, North Lantau Hospital – *Representative of COC (Pathology) (Haematopathology)* (up to 12 September 2018)
Dr Eudora CHOW	Service Director (Pathology), Kowloon East Cluster / Chief of Service (Pathology), United Christian Hospital – *Representative of COC (Pathology) (Haematopathology)* (from 12 September 2018)
Dr Mandy CHU	Consultant (Obstetrics and Gynaecology), Queen Mary Hospital – *Representative of CC (Cancer Service)*
Dr W T NG	Clinical Coordinator (Patient Relations) / Deputy Chief of Service (Clinical Oncology), Pamela Youde Nethersole Eastern Hospital – *Representative of COC (Clinical Oncology)*
Dr Pun HUI	Deputy Chief of Service (Clinical Oncology) / Consultant (Clinical Oncology), Prince of Wales Hospital – *Representative of COC (Medicine) (Medical Oncology)*
Dr Albert LIE	Deputy Hospital Chief Executive (Planning) / Consultant (Medicine), Queen Mary Hospital – *Representative of COC (Medicine) (Haematological Oncology)* (up to 12 July 2019)
Dr June LAU	Consultant (Medicine), Queen Elizabeth Hospital – *Representative of COC (Medicine) (Haematological Oncology)* (from 12 July 2019)
Dr H T LEONG	Consultant (Surgery), North District Hospital – *Representative of COC (Surgery)*
Dr W C LEUNG	Chief of Service (Obstetrics & Gynaecology), Kwong Wah Hospital – *Representative of COC (Obstetrics & Gynaecology)*
Dr Brian CHUNG	Clinical Associate Professor, Department of Paediatrics & Adolescent Medicine, Li Ka Shing Faculty of Medicine, The University of Hong Kong / Honorary Consultant (Paediatrics), Queen Mary Hospital – *Representative of COC (Paediatrics)*
Dr Jason SO	Chief of Service (Pathology), Hong Kong Children's Hospital – *Representative of Hong Kong Children's Hospital*
Dr Ivan LO	Consultant Clinical Geneticist, Clinical Genetic Service, Department of Health – *Representative of Department of Health Clinical Genetic Service*
Prof S Y LEUNG	Chair Professor of Gastrointestinal Cancer Genetics & Genomics, Li Ka Shing Faculty of Medicine, The University of Hong Kong – *Representative of the University of Hong Kong*
Prof K F TO	Professor and Chairman, Department of Anatomical and Cellular Pathology, Faculty of Medicine, The Chinese University of Hong Kong – *Representative of the Chinese University of Hong Kong*
Ms Sinley LAI	Advanced Practice Nurse (Obstetrics & Gynaecology), Tuen Mun Hospital – *Representative of Nursing Grade*
Dr Gene LAU	Scientific Officer (Medical), Department of Pathology, Queen Elizabeth Hospital – *Representative of Allied Health Grade (Scientific Officer)*
Mr Lawrence POON	Chief Manager (Nursing) / Chief Nurse Executive, HA Head Office
Dr Rebecca LAM	Chief Manager (Clinical Effectiveness & Technology Management), HA Head Office (up to 12 November 2018)
Ms Rita HO	Deputising Chief Manager (Clinical Effectiveness & Technology Management) / Senior Manager (Technology Management), HA Head Office (from 12 November 2018 to 14 January 2019)
Dr Linda YU	Chief Manager (Clinical Effectiveness & Technology Management), HA Head Office (from 14 January 2019)
Dr Leo WAT	Chief Manager (Strategy, Service Planning & Knowledge Management), HA Head Office (up to 20 August 2018)
Dr Flora TSANG	Chief Manager (Strategy, Service Planning & Knowledge Management), HA Head Office (from 20 August 2018)

Secretary

Ms Looi-looi LOW	Senior Manager (Strategy & Service Planning), HA Head Office

Appendix 2: Working Group on Governance & Service Development

Terms of Reference

- To advise on the development priorities, service model principles, governance structure and service organisation for HA genetic and genomic services, taking into consideration the ethical, legal and social implications
- To advise on the quality assurance and performance monitoring of HA genetic and genomic services
- To report the recommendations to the GGS SSF Taskforce for consideration and formulation of strategies

Membership (as at November 2018)

Co-chairs

Dr Flora TSANG	Chief Manager (Strategy, Service Planning & Knowledge Management), HA Head Office
Dr Rebecca LAM	Chief Manager (Clinical Effectiveness & Technology Management), HA Head Office

Members

Dr Linda YU	Chief Manager (Integrated Care Programs), HA Head Office
Dr N C SIN	Chief Manager (Patient Safety & Risk Management), HA Head Office
Mr Lawrence POON	Chief Manager (Nursing) / Chief Nurse Executive, HA Head Office
Ms Ivis CHUNG	Chief Manager (Allied Health), HA Head Office
Ms Anna LEE	Chief Pharmacist, HA Head Office
Dr T L LEE	Hospital Chief Executive, Hong Kong Children's Hospital
Dr K F WONG	Service Director (Pathology) & Coordinator of Cluster (Public Private Interface), Kowloon Central Cluster / Deputy Hospital Chief Executive (Corporate Affairs) & Consultant Pathologist (Haematology), Queen Elizabeth Hospital / Commissioning Service Coordinator (Pathology), Hong Kong Children's Hospital – *Representative of CC (Genetic Services)*
Dr W CHEUK	Consultant Pathologist (Anatomical Pathology), Queen Elizabeth Hospital – *Representative of COC (Pathology) (Anatomical Pathology)*
Dr Michael CHAN	Cluster Coordinator (Pathology), New Territories East Cluster / Consultant (Chemical Pathology), Prince of Wales Hospital – *Representative of COC (Pathology) (Chemical Pathology)*
Dr Eudora CHOW	Service Director (Pathology), Kowloon East Cluster / Chief of Service (Pathology), United Christian Hospital – *Representative of COC (Pathology) (Haematopathology)*
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Dr C B LAW	Deputy Hospital Chief Executive (Clinical Services), Princess Margaret Hospital / Chief of Service (Medicine & Geriatrics), Princess Margaret Hospital & North Lantau Hospital – *Representative of COC (Internal Medicine)*
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Prof K F TO	Professor and Chairman, Department of Anatomical and Cellular Pathology, Faculty of Medicine, The Chinese University of Hong Kong / Honorary Chief of Service & Chairman (Anatomical & Cellular Pathology), Prince of Wales Hospital – *Representative of the Chinese University of Hong Kong*

Secretary

Ms Looi-looi LOW	Senior Manager (Strategy & Service Planning), HA Head Office

Appendix 3: Working Group on Cancer Genetic Services

Terms of Reference

- To work out the service organisation, care pathway and hospital role delineation for HA's Cancer G/G Programme, according to the service model set out by the GGS SSF Taskforce
- To advise on the staff expertise and training required for HA's cancer genetic services
- To advise on the future developments relevant to HA's cancer genetic services, including translational research
- To develop at least two examples (germline and somatic mutations) to illustrate the Working Group's recommendations
- To report the recommendations to the GGS SSF Taskforce for consideration and formulation of strategies

Membership (as at November 2018)

Co-chairs

Dr Flora TSANG	Chief Manager (Strategy, Service Planning & Knowledge Management), HA Head Office
Dr Rebecca LAM	Chief Manager (Clinical Effectiveness & Technology Management), HA Head Office

Members

Dr Linda YU	Chief Manager (Integrated Care Programs), HA Head Office
Dr W Y SO	Chief Manager (Quality & Standards), HA Head Office
Mr Lawrence POON	Chief Manager (Nursing) / Chief Nurse Executive, HA Head Office
Ms Ivis CHUNG	Chief Manager (Allied Health), HA Head Office
Ms Anna LEE	Chief Pharmacist, HA Head Office
Dr K F WONG	Service Director (Pathology) & Coordinator of Cluster (Public Private Interface), Kowloon Central Cluster / Deputy Hospital Chief Executive (Corporate Affairs) & Consultant Pathologist (Haematology), Queen Elizabeth Hospital / Commissioning Service Coordinator (Pathology), Hong Kong Children's Hospital – *Representative of CC (Genetic Services)*
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Prof Rossa CHIU	Professor of Chemical Pathology and Assistant Dean (Research), Faculty of Medicine, The Chinese University of Hong Kong – *Representative of the Chinese University of Hong Kong*

Secretary

Ms Looi-looi LOW	Senior Manager (Strategy & Service Planning), HA Head Office

Appendix 4: Working Group on Prenatal & Paediatric Genetic Services

Terms of Reference

- To work out the service organisation, care pathway and hospital role delineation for HA's Paediatric G/G Programme (including prenatal, IEM and uncommon disorder services), according to the service model set out by the GGS SSF Taskforce
- To advise on the staff expertise and training required for HA's prenatal and paediatric genetic services
- To advise on the future developments relevant to HA's prenatal and paediatric genetic services, including translational research
- To develop at least two examples (including uncommon disorder) to illustrate the Working Group's recommendations
- To report the recommendations to the GGS SSF Taskforce for consideration and formulation of strategies

Membership (as at November 2018)

Co-chairs

Dr Flora TSANG	Chief Manager (Strategy, Service Planning & Knowledge Management), HA Head Office
Dr Rebecca LAM	Chief Manager (Clinical Effectiveness & Technology Management), HA Head Office

Members

Dr N C SIN	Chief Manager (Patient Safety & Risk Management), HA Head Office
Dr T L LEE	Hospital Chief Executive, Hong Kong Children's Hospital
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Ms Sinley LAI	Advanced Practice Nurse (Obstetrics & Gynaecology), Tuen Mun Hospital – *Representative of COC (Obstetrics & Gynaecology)*
Dr Joannie HUI	Consultant (Paediatrics/IEM), Hong Kong Children's Hospital / Consultant (Paediatrics), Prince of Wales Hospital – *Representative of COC (Paediatrics)*
Dr Alan SO	Chief of Service (Paediatrics), Prince of Wales Hospital – *Representative of COC (Paediatrics)*
Dr Chloe MAK	Consultant (Pathology), Hong Kong Children's Hospital – *Representative of COC (Pathology)*
Dr Liz YUEN	Consultant (Pathology), Hong Kong Children's Hospital – *Representative of Hong Kong Children's Hospital*
Dr Kelvin CHAN	Scientific Officer (Medical), Prenatal Diagnostic & Counselling, Tsan Yuk Hospital – *Representative of Tsan Yuk Hospital Prenatal Diagnostic & Counselling Division - Laboratory Service*
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Dr Ivan LO	Consultant Clinical Geneticist, Clinical Genetic Service, Department of Health – *Representative of Department of Health Clinical Genetic Service*
Dr Brian CHUNG	Clinical Associate Professor, Department of Paediatrics & Adolescent Medicine, Li Ka Shing Faculty of Medicine, The University of Hong Kong / Honorary Consultant (Paediatrics), Queen Mary Hospital – *Representative of the University of Hong Kong*
Prof T Y LEUNG	Chairman & Professor, Department of Obstetrics & Gynaecology, Faculty of Medicine, The Chinese University of Hong Kong / Consultant (Obstetrics & Gynaecology), Prince of Wales Hospital – *Representative of the Chinese University of Hong Kong*

Secretary

Ms Looi-looi LOW	Senior Manager (Strategy & Service Planning), HA Head Office

Appendix 5: Subgroup on Service Organisation & Funding

Terms of Reference

- To advise the Working Group on Governance and Service Development on the service organisation and funding arrangement for HA's genetic and genomic services

- To suggest the future service model and system infrastructure for developing a dedicated clinical genetic service in HA and for the provision of laboratory genetic services

Membership (as at November 2018)

Co-chairs

Dr Flora TSANG	Chief Manager (Strategy, Service Planning & Knowledge Management), HA Head Office
Dr Rebecca LAM	Chief Manager (Clinical Effectiveness & Technology Management), HA Head Office

Members

Dr Linda YU	Chief Manager (Integrated Care Programs), HA Head Office
Dr N C SIN	Chief Manager (Patient Safety & Risk Management), HA Head Office
Dr T L LEE	Hospital Chief Executive, Hong Kong Children's Hospital
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Dr Ashley CHENG	Cluster Clinical Coordinator (Health Informatics), Kowloon West Cluster & Princess Margaret Hospital / Chief of Service (Oncology), Kowloon West Cluster – *Representative of CC (Cancer Service)*
Dr Alan SO	Chief of Service (Paediatrics), Prince of Wales Hospital – *Representative of COC (Paediatrics)*
Dr Ivan LO	Consultant Clinical Geneticist, Clinical Genetic Service, Department of Health – *Representative of Department of Health Clinical Genetic Service*

Appendix 6: Subgroup on Staff Expertise & Role Delineation

Terms of Reference

- To advise the Working Group on Governance and Service Development on the staff expertise and role delineation in providing HA's genetic and genomic services

- To suggest the roles and responsibilities, as well as skills and training requirement, of staff involved in HA's genetic and genomic services

- To suggest ways for improving the genetic literacy of healthcare staff

Membership (as at November 2018)

Co-chairs

Dr Flora TSANG	Chief Manager (Strategy, Service Planning & Knowledge Management), HA Head Office
Dr Rebecca LAM	Chief Manager (Clinical Effectiveness & Technology Management), HA Head Office

Members

Mr Lawrence POON	Chief Manager (Nursing) / Chief Nurse Executive, HA Head Office
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Dr W C LEUNG	Chief of Service (Obstetrics & Gynaecology), Kwong Wah Hospital – *Representative of COC (Obstetrics & Gynaecology)*
Dr Brian CHUNG	Clinical Associate Professor, Department of Paediatrics & Adolescent Medicine, Li Ka Shing Faculty of Medicine, The University of Hong Kong / Honorary Consultant (Paediatrics), Queen Mary Hospital – *Representative of the University of Hong Kong*

Appendix 7: Subgroup on Introduction of New Technology & Performance Measures

Terms of Reference

- To advise the Working Group on Governance and Service Development on the introduction of new technology and performance monitoring in HA's genetic and genomic services
- To suggest the mechanisms for assessing and prioritising new services / tests, and for the translation of research into clinical practice in genetic and genomic services
- To suggest standard measures and indicators for monitoring the service performance and outcomes

Membership (as at November 2018)

Co-chairs

Dr Flora TSANG	Chief Manager (Strategy, Service Planning & Knowledge Management), HA Head Office
Dr Rebecca LAM	Chief Manager (Clinical Effectiveness & Technology Management), HA Head Office

Members

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Dr N C SIN	Chief Manager (Patient Safety & Risk Management), HA Head Office
Ms Anna LEE	Chief Pharmacist, HA Head Office
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Dr W T NG	Clinical Coordinator (Patient Relations) / Consultant (Clinical Oncology), Pamela Youde Nethersole Eastern Hospital – *Representative of COC (Clinical Oncology)*
Prof S Y LEUNG	Chair Professor of Gastrointestinal Cancer Genetics & Genomics, Li Ka Shing Faculty of Medicine, The University of Hong Kong – *Representative of the University of Hong Kong*
Prof K F TO	Professor and Chairman, Department of Anatomical and Cellular Pathology, Faculty of Medicine, The Chinese University of Hong Kong / Honorary Chief of Service & Chairman (Anatomical & Cellular Pathology), Prince of Wales Hospital – *Representative of the Chinese University of Hong Kong*

Appendix 8: HA Central Committee on Genetic Services

Terms of Reference

- To advise HA on service needs and strategies for developing clinical and laboratory genetic services
- To establish principles and criteria for prioritising genetic services and help to coordinate service development
- To identify workforce and training needs
- To monitor the quality of service implementation

Membership (as at August 2019)

Co-chairs

Dr K L CHUNG	Director (Quality & Safety), HA Head Office
Dr W C LEUNG	Chief of Service (Obstetrics & Gynaecology), Kwong Wah Hospital – *Representative of COC (Obstetrics & Gynaecology)*

Members

Dr Alex CHAN	Service Director (Pathology), Kowloon Central Cluster / Chief of Service (Pathology), Queen Elizabeth Hospital – *Representative of COC (Pathology) – Anatomical Pathology*
Dr Michael CHAN	Cluster Coordinator (Pathology), New Territories East Cluster / Consultant (Chemical Pathology), Prince of Wales Hospital – *Representative of COC (Pathology) – Chemical Pathology*
Dr David LUNG	Consultant (Pathology), Queen Elizabeth Hospital – *Representative of COC (Pathology) – Microbiology*
Dr Eudora CHOW	Service Director (Pathology), Kowloon East Cluster / Chief of Service (Pathology), United Christian Hospital – *Representative of COC(Pathology) - Haematology*
Dr Thomas WAN	Scientific Officer (Medical) (Haematology), Prince of Wales Hospital – *Representative of COC (Pathology) – Allied Health Committee of Pathology*
Dr Ashley CHENG	Cluster Clinical Coordinator (Health Informatics), Kowloon West Cluster & Princess Margaret Hospital / Chief of Service (Oncology), Kowloon West Cluster – *Representative of COC (Clinical Oncology)*
Dr Macy TONG	Consultant (Clinical Oncology), Prince of Wales Hospital – *Representative of COC (Clinical Oncology)*
Dr Windsor MAK	Consultant (Medicine), Queen Mary Hospital – *Representative of COC (Internal Medicine) – Neurology*
Dr N S MOK	Consultant (Medicine & Geriatrics), Princess Margaret Hospital – *Representative of COC (Internal Medicine) – Cardiology*
Dr K Y LEUNG	Cluster Coordinator (Medical Records), Kowloon Central Cluster / Chief of Service (Obstetrics & Gynaecology), Queen Elizabeth Hospital – *Representative of COC (Obstetrics & Gynaecology)*
Dr Betty BUT	Chief of Service (Paediatrics), Queen Elizabeth Hospital – *Representative of COC (Paediatrics)*
Dr Alan SO	Chief of Service (Paediatrics), Prince of Wales Hospital – *Representative of COC (Paediatrics)*
Prof Ronald MA	Professor and Head, Division of Endocrinology & Diabetes, Department of Medicine & Therapeutics, Faculty of Medicine, The Chinese University of Hong Kong / Honorary Consultant (Medicine), Prince of Wales Hospital – *Representative of CC (Diabetic Service)*
Prof Dennis LO	Director of Li Ka Shing Institute of Health Sciences & Chairman and Professor of Department of Chemical Pathology, Faculty of Medicine, The Chinese University of Hong Kong – *Expert in Genomics*
Prof P C SHAM	Director of Centre for Genomic Sciences & Chair Professor in Psychiatric Genomics of Department of Psychiatry, Li Ka Shing Faculty of Medicine, The University of Hong Kong – *Expert in Genomics*
Prof T Y LEUNG	Chairman & Professor, Department of Obstetrics & Gynaecology, Faculty of Medicine, The Chinese University of Hong Kong / Consultant (Obstetrics & Gynaecology), Prince of Wales Hospital – *Expert in Obstetrics & Gynaecology*
Dr Mary TANG	Honorary Consultant (Obstetrics and Gynaecology), Queen Mary Hospital / Clinical Associate Professor, Department of Obstetrics & Gynaecology, Li Ka Shing Faculty of Medicine, The University of Hong Kong – *Expert in Obstetrics & Gynaecology*
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Dr Brian CHUNG	Clinical Associate Professor, Department of Paediatrics & Adolescent Medicine, Li Ka Shing Faculty of Medicine, The University of Hong Kong / Honorary Consultant (Paediatrics), Queen Mary Hospital – *Expert in Paediatrics*
Dr T L LEE	Hospital Chief Executive, Hong Kong Children's Hospital – *Representative of Hong Kong Children's Hospital*

Co-opted Member

Dr Ivan LO	Consultant Clinical Geneticist, Clinical Genetic Service, Department of Health – *Expert in Clinical Genetics*

Appendix 9: Steering Group on Genetic and Genomic Services in HA

Terms of Reference

- To advise on HA's role in dovetailing with the Government policy direction in relation to the development of genomic medicine in Hong Kong, as proposed by the Steering Committee on Genomic Medicine (SCGM) including the Hong Kong Genome Project (HKGP)
- To steer the overall development of genetic and genomic services in HA with regard to strategic directions in HA Strategic Service Framework for Genetic & Genomic Services (HA GGS SSF)
- To endorse the action plans and monitor the implementation
- To report to the Directors' Meeting and the Medical Services Development Committee, as appropriate, on the development of genetic and genomic services in HA

Membership (as at August 2019)

Chairman

Dr Tony KO	Chief Executive, HA

Members

Dr K L CHUNG	Director (Quality & Safety), HA Head Office
Dr Libby LEE	Director (Strategy & Planning), HA Head Office
Dr Deacons YEUNG	Director (Cluster Services), HA Head Office
Dr F C PANG	Head of Human Resources, HA Head Office
Dr N T CHEUNG	Head of Information Technology & Health Informatics / Chief Medical Informatics Officer, HA Head Office
Dr Theresa LI	Cluster Chief Executive, Hong Kong West Cluster / Hospital Chief Executive, Queen Mary Hospital & Tsan Yuk Hospital
Dr S V LO	Cluster Chief Executive, New Territories East Cluster / Hospital Chief Executive, Prince of Wales Hospital
Dr Albert LO	Cluster Chief Executive, Kowloon Central Cluster / Hospital Chief Executive, Queen Elizabeth Hospital
Dr T L LEE	Hospital Chief Executive, Hong Kong Children's Hospital
Dr W C LEUNG	Chairman, Central Committee (Genetic Services) / Chief of Service (Obstetrics & Gynaecology), Kwong Wah Hospital
Dr T L QUE	Chairman, Coordinating Committee (Pathology) / Chief of Service (Clinical Pathology), Tuen Mun Hospital, Pok Oi Hospital & Tin Shui Wai Hospital

Secretary

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