# 一站式跨專科門診 支援遺傳病病人

# *Genetic unit offers one-stop service to patients*



陸浩明醫生表示，很多時根據病人病歷和詳細家族史，已大概知道病人是否患遺傳病。

Dr Luk Ho-ming says through detailed family history and reviewing of medical record, genetic doctor can tell if patients have genetic conditions in most of the cases.

醫管局去年接辦衞生署的醫學遺傳服務，在香港兒童醫院（兒童醫院）提供一站式遺傳病評估、檢驗、診斷、治療、輔導及預防服務。該院更設跨團隊專科門診，由醫學遺傳科聯同其他專科團隊會診，幫助患者及其家屬制定管理計劃和縮短病人輪候時間。「很多時病人看過很多專科，直至遺傳科醫生作評估或基因檢測，才知道自己患甚麼病。病人早一日確診，早一日安心，並且進行個人化治療，認識未來的生育風險等。」該院醫學遺傳科顧問醫生**陸浩明**說。

## 首次會診「查三代」

兒童醫院遺傳科服務需由公私營機構轉介，服務範圍涵蓋任何年紀、懷疑或確診遺傳病的病人及其家人。陸醫生說，遺傳病出現誤診或延遲診斷在全球很常見，「由於遺傳病沒有特定的病徵，根據文獻，在沒有遺傳科醫生協助及基因檢測的時代，平均五至七年才能作出診斷。通過遺傳科提供的評估和檢測，希望及早為病人作出診斷並盡快提供治療。」

遺傳科醫生在專科門診首次會診時會詳細詢問家族病史如「查三代」，繪畫家族譜系圖，然後為病人進行身體檢查，如有需要會進行基因檢測，及為其他家人轉介或進行檢測。此外，兒童醫院醫學遺傳科亦會為該院懷疑患遺傳病的病童，提供臨床住院諮詢服務。

## 減省病人輪候時間

陸醫生強調，跨專科門診非常重要。「不是所有醫生都認識遺傳病，不能單靠一人或一個專科之力了解所有病人個案，由遺傳科加上其他專科或亞專科如心臟科、內分泌科或皮膚科等一起為病人提供專業意見，大大減省病人輪候時間，也可整體檢視病人情況，為複雜的個案制定個人化的管理計劃。」

他曾遇過一名病人透過不同臨床及研究基因檢測，多年後終確診杜興氏肌肉營養不良症，母親是基因攜帶者。她清楚自己的生育風險和選擇，將來再生育的話，需要做產前篩選。「知道答案後，全家都很開心。雖然多數遺傳病目前沒有針對性治療，誰知將來有沒有呢？有準確的遺傳診斷，最少病人日後的路也清晰許多。」




香港兒童醫院醫學遺傳科設跨專科門診，由不同專科的醫生會診，一起為病人提供意見。

The cross-specialty clinic that set up by CGSU and different specialties provides assessment and gives professional advice for patients in a one-stop manner.

### 遺傳病教室
### Understand genetic diseases

**病人在哪種情況下需要轉介？**
**When does a patient require a referral to CGSU?**

在以下情況，醫生可考慮轉介予遺傳科評估及遺傳諮詢：異常的發病年齡，如小朋友罹患年長人士較常見的疾病，如癌症；家族中其他成員都有相同病症或病狀；有較嚴重或非典型病徵，如多重器官有結構問題等。

A doctor may refer a case to the CGSU for assessment and genetic counselling in the following circumstances: unusual age of onset for diseases like a child suffers from a disease that usually affects older people such as cancer, when other family members have the similar disease or symptoms, and/or when a patient has unusual serious or non-typical symptoms such as a condition affecting different organs at the same time in the patient.

**為甚麼有些遺傳病「傳男不傳女」？**
**Why do some genetic diseases mostly affect males?**

男性的染色體是 XY，女性是 XX。X染色體連鎖遺傳病如色盲、血友病、蠶豆症、杜興氏肌肉營養不良症等，由X染色體的基因突變引起。由於女性有兩個 X，其中一個 X出現問題，另一個 X多數可以彌補，所以女性如果是基因攜帶者，大部分不會出現病徵。反之，因為男性只有一個 X，X染色體有問題的話，男性必定會發病。

Males have one X chromosome and one Y chromosome, while females have two X chromosomes. Some X linked diseases like color blindness, haemophilia, favism (G6PD deficiency), and Duchenne muscular dystrophy, etc. are caused by genetic mutation in genes located in X chromosome. As females have two X chromosomes, if the gene in one X chromosome is faulty, the same gene in another X chromosome may help compensate. When this happens, a female genetic carrier may not have symptoms, however, a male with X linked disease must be symptomatic.



The Hospital Authority (HA) took over the Clinical Genetic Service previously run by the Department of Health last year. The Hong Kong Children's Hospital (HKCH) now provides a one-stop genetics and genomics service covering assessment, investigation, diagnosis, treatment, counselling, and prevention. Cross-specialty clinic has also been set up to provide consultations by the Clinical Genetics Service Unit (CGSU) and other specialties, which helps our patients and families to formulate their management plan and reduces the waiting times. CGSU Consultant Dr **Luk Ho-ming** explains, "most patients are not diagnosed until they are assessed by genetic doctors or undergo genetic testing after consulting many specialists. As soon as patients get a diagnosis, they feel relieved, and they can have personalised treatment plan and understand their future reproductive risk in the family."

## Asking family history on first appointment

The CGSU at the HKCH serves patients of all ages with suspected or confirmed genetic conditions, referred from both the public and private sectors. Misdiagnosis and delay in diagnosis are common for genetic diseases all around the world. "Because most genetic diseases do not have specific signs and symptoms, without the inputs of genetic doctor and genetic testing, patients are often only diagnosed after average of five to seven years as mentioned in literatures," says Dr Luk. "Assessments and testing provided by the genetic team can help patients to have early accurate diagnosis and receive personalised treatment as soon as possible."

On the first appointment, the genetic doctor will ask for the patient's detailed family history, draw a family tree for analysis and perform a thorough physical examination. Genetic testing is then provided where appropriate. The doctor will also consult the patients and their families when needed afterwards. CGSU will also provide inpatient consultations for children in HKCH who are suspected to have a genetic disease.

## Improving waiting times

Cross-specialty consultation is an essential part of the process, Dr Luk emphasises. "Not every single doctor knows all genetic diseases well, and it is not possible to lean on any single person or specialty to understand all patient cases," he says. "The CGSU collaborates with different specialties and subspecialties like cardiology, endocrinology, and dermatology to provide professional advice for patients. This not only reduces the patient's waiting time significantly but also provides holistic care to formulate the personalised management plan for complicated genetic diseases."

Dr Luk cites the case of a boy suspected to have a genetic disease for many years, he was finally diagnosed with Duchenne muscular dystrophy after extensive clinical and research testing. His mother was found to be a genetic carrier. With that, the mother now understands more her reproductive risk and options that she can undergo prenatal screening if she has another child in future. "Once they learned the diagnosis, the whole family was relieved," recalls Dr Luk. "Although there is currently no cure for most of the genetic diseases, it may have treatment in the future, who knows? At least there is a clear pathway for patient with accurate genetic diagnosis."

遺傳輔導服務短片
Genetic counselling service video

# 全方位透視基因
# Genetic testing for inherited diseases







（左起）鄭華哲醫生、蘇志釗醫生及麥苗醫生。
(From left) Dr Timothy Cheng, Dr Jason So and Dr Chole Mak.

新生兒篩查化驗室，與遺傳及基因組學病理化驗室分別於2018及2019年起在兒童醫院病理學部提供服務。至2023年，原屬衞生署的醫學遺傳化驗服務，亦合併至兒童醫院病理學部。為配合是次服務整合，病理學部增加人手和添置先進儀器，在天時地利人和下順利交接。部門主管**蘇志釗**醫生表示，遺傳化驗服務對病人及家人都非常重要，通過擴大檢測範圍，醫生可為病人提早作出診斷，制定合適的治療方案。

顧問醫生**麥苗**解釋，病人在三種情況下會進行遺傳測試，最常見是診斷式，當病人有病徵，醫生臨床懷疑是遺傳病；第二是預測性，病人有遺傳病家族史或病人服用的藥物需要藥物基因學數據，以作風險評估及調節劑量；第三是篩查，特別是初生嬰兒，讓他們未有病徵前進行測試，以防潛在嚴重甚至有生命危險的情況出現。

是否所有病因都可檢測得到？顧問醫生**鄭華哲**指出，疾病可由多種遺傳因素或環境因素所造成，遺傳測試無法完全解釋我們遇到的所有情況。隨著全基因組排序變得普及，加上我們對基因組的認識加深，有助我們更了解基因變異和不同疾病的連繫。

## 新增新生兒篩查範圍

現時兒童醫院新生兒篩查化驗室會為公立醫院出生的新生嬰兒，進行俗稱蠶豆症的葡萄糖六磷酸去氫酵素缺乏症，及先天性甲狀腺功能不足的篩查服務，加上26種先天性代謝病、嚴重聯合免疫缺陷病，以及脊髓肌肉萎縮症（先導）。麥醫生期望日後可引入更多病種作篩查，令計劃更全面。

The Newborn Screening Laboratory and the Genetic & Genomic Pathology Laboratory commenced services in the Department of Pathology of HKCH in 2018 and 2019 respectively. In 2023, the clinical genetic laboratory services of the Department of Health were also transferred to the Department. Manpower was increased in the Department and advanced equipment was installed to allow for a smooth transfer. The Department's Chief of Service Dr **Jason So** says genetic testing is vital for patients and families. Doctors are now able to obtain early diagnosis and draw up treatment plans through an expanded scope of genetic testing.

Consultant Dr **Chloe Mak** explains that genetic testing takes place in three scenarios. The most common is diagnostic, when doctors suspect a patient has a genetic disease because of their symptoms. The second scenario is predictive, when subjects have a family history of genetic disease or are taking drugs that may need pharmacogenetic data for risk assessment and dosage adjustment. The third is screening, especially for newborns, during which babies are given pre-symptomatic testing to check for potentially serious and even life-threatening conditions.

Can all diseases be diagnosed through genetic testing? Consultant Dr **Timothy Cheng** points out that diseases can arise from a combination of genetic factors and environmental factors. Genetic testing cannot fully explain all the conditions that we encounter. As whole genome sequencing becomes more common, and as our understanding of the human genome improves, we may have a better understanding of the links between genetic variants and different diseases.

## Increasing scope of newborn screening

The Newborn Screening Laboratory of HKCH screens all newborn babies delivered in public hospitals for glucose-6-phosphate dehydrogenase (G6PD) deficiency and congenital hypothyroidism as well as for 26 metabolism defects, severe combined immune deficiency, and spinal muscular atrophy (pilot). Dr Mak hopes more conditions will be included in future newborn screening to make the programme even more comprehensive.

## 新角色 New posts:

在提供遺傳科服務的過程中，出現兩個重要的新角色：遺傳輔導員和生物信息學家，他們分別為病人提供輔導，及協助醫生診斷。

Two essential posts are created in the provision of genetic service: Genetic Counsellor and Bioinformatician. They counsel patients and analyse data to help doctors to make a diagnosis respectively.

訪問短片
Interview video

## 遺傳輔導員：遺傳科解籤佬 Genetic Counsellor: The genetic fortune teller

**施芳瑩**的主要工作是根據病人家族病史繪成家族譜系圖，就風險評估提供輔導，獲得對方知情同意後進行測試，並向病人及家人解釋報告結果。「病人做測試好像求籤，報告內容有時跟古文一樣難解，遺傳輔導員就像解籤佬，用簡單易明的方法替病人解讀測試結果，讓他們明白古文的含義。」



遺傳輔導員大都有醫療相關背景，大學修讀遺傳輔導、生物醫學、護理學、社會工作、心理學、理科、醫療科學或相關學科。

**Connie Shih** looks into a patient's genetic future by studying their family backgrounds, provides counselling for risk assessments, carries out tests on patients after obtaining informed consent, and explains the results to patients and their family members. "Undergoing a genetic test is like drawing a fortune stick, and the report can sometimes be as obscure as the ancient fortune poems," Connie explains. "A genetic counsellor acts like a fortune teller and interprets the results for the patient so they can understand them and adapt to the implications."

Most Genetic Counsellors have a medical-related background and studied genetic counselling, biomedical sciences, nursing, social work, psychology, science, medical science or equivalent at the university level.

## 生物信息學家：數據解讀者 Bioinformatician: The data interpreter

大部分基因檢測只需抽血便可完成，基因排序後會產生大量原始數據，每名病人的數據可大至10G甚至100G。**蘇小茜**和**吳峰**博士說，生物信息學家的工作是使用生物信息的軟件和工具，系統化收集、儲存和檢索遺傳和基因組資料。他們會根據最新的臨床證據和指引分析基因組資料，並整合來自各種生物信息學資料庫和研究發現，以協助醫生診斷。此外，他們負責更新病人和基因資料庫，並與醫生和遺傳輔導員緊密溝通，討論病例，分享有關指引更新的知識。



生物信息學家大多是修讀生物信息學、臨床科學、生物醫學、計算機科學、統計學或相關學科，熟悉編寫和編碼程式。

Most genetic tests can be done by a simple blood test which provides a vast amount of raw data following gene sequencing. The data can be as large as 10G or even 100G for each patient. **Susie Su** and Dr **Feng Wu** say the role of a bioinformatician involves designing and implementing workflows for the systematic collection, storage, and retrieval of genetic and genomic data by using bioinformatics software and tools. They analyse and annotate genomic data with the latest clinical evidence and guidelines, integrating information from various bioinformatics databases and research findings to assist doctors in patient diagnosis. Additionally, they are responsible for maintaining up-to-date databases of patient and genetic information. They also closely communicate with clinicians and genetic counsellors for case discussions and share knowledge about guideline updates.

A bioinformatician usually studied bioinformatics, clinical science, biomedical science, computer science, statistics or related discipline, with expertise in programming and coding.