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香港兒童醫院
Hong Kong Children's Hospital

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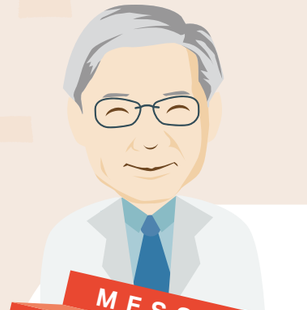


虛心求知 Learning

同理關懷 Caring

會心微笑 Smiling

院長的話

加強交流 Strengthen exchanges

香港兒童醫院成立以來，一直朝着卓越醫療中心的目標邁進。然而，本地兒童人口始終較小，我們必須汲取其他地方治療奇難雜症的經驗和最新醫學發展，擴闊視野，與世界接軌。因此，我向來強調對外交流是非常重要的。

隨着疫情過去，阻隔消除，我樂見不同專科團隊都很積極地建立對外聯繫，重啟交流活動。例如，早前放射科和心臟科的同事分別有份籌辦相關專業學會在港召開的大型亞太區醫學會議，並邀請出席的國際專家前來醫院參觀，介紹我們的服務。上個月，血液及腫瘤科舉辦了一場病例研討會。深圳市兒童醫院、中山大學附屬第一醫院等接近20間醫院總共70多位專家雲集本院，一起討論複雜難治的病例，交換專業意見，互相借鏡。

自七月開始，模擬培訓中心每個月舉行hospital grand round，由本院跨專科團隊分享不同兒科病症的治理經驗。除本地公立和私家醫護人員外，更陸續有內地和澳門的同業透過視像加入。

我鼓勵同事把握機遇，加入相關網絡和平台，與各地同業加強實體和非實體的聯繫。這絕對有助推動臨床、培訓和科研各方面的合作，互惠雙贏，最後當然是病人得益。

醫院行政總監
李子良醫生

Becoming a centre of excellence has always been the goal of HKCH. As the local children population is relatively small, it is crucial for us to learn from other places about their experience in treating complicated diseases and the latest medical advancement. I cannot emphasize enough the importance of exchanges in broadening our horizon and raising our standards.

With the lifting of barriers after the pandemic, it is encouraging to see that our clinical teams are proactively building external relations and resuming exchange activities. Earlier, our Radiology and Cardiology colleagues took part in organizing Asia-pacific medical conferences with their respective professional societies. Some speakers and participants also took the opportunity to visit HKCH while they were in town. Last month, Haematology & Oncology held a seminar where over 70 experts from nearly 20 hospitals in the Greater Bay Area gathered in HKCH to share practices and insights in tackling some difficult patient cases.

The Simulation Training Centre now organizes a monthly hospital grand round, a platform for introducing our multi-disciplinary management of different paediatric diseases. Apart from healthcare professionals from the local public and private sectors, Mainland and Macau counterparts are also joining via live streaming.

I encourage colleagues to seize every opportunity to join relevant networks, and maintain close ties and communication with counterparts around the world. These connections will definitely facilitate collaboration in the clinical, training and research aspects, and create mutual benefits towards a better patient service.

Dr Lee Tsz-leung
Hospital Chief Executive

雲集兒童血液及腫瘤專家的病例研討會在香港兒童醫院舉行。

A seminar on haematology and oncology was held in HKCH. It was attended by over 70 experts in the Greater Bay Area.

遺傳及基因組醫學帶來治療曙光

香港兒童醫院配備次世代定序儀，能進行大規模基因測序，為病人查找病因。

HKCH is equipped with next generation sequencers which can perform massive DNA sequencing to look for the cause of diseases.

GENETICS & GENOMICS

quest for answers and cures

一些不常見的遺傳病並無明確病徵，部份患者歷盡漫長診斷過程的煎熬，錯過了治療時機，亦有人突然發病，才發現情況嚴重。遺傳及基因組醫學是全球大勢所趨，政府近年大力推動，冀讓市民受惠於更精準的診斷和更有效的治療。

Some uncommon genetic disorders do not have distinctive clinical features, leading to diagnostic odyssey and delayed treatment, while some patients do not realize they are affected until severe symptoms suddenly surface. The government is determined to promote genetic and genomic medicine to benefit the community with more precise diagnosis and effective treatment.

香港兒童醫院在本港的遺傳及基因組醫學發展中扮演重要角色。作為全港不常見遺傳病的轉介中心，我們匯聚相關範疇的專家和先進化驗技術，負責促進這些疾病的診治、研究、專才培訓和認知。

本院自今年七月起接辦了原屬衞生署的醫學遺傳服務（包括遺傳輔導診所、遺傳篩選診所、初生嬰兒普檢化驗、遺傳化驗），集中為遺傳病患者和家庭成員提供一站式的跨專業支援，涵蓋診斷、治療、後續跟進、輔導及預防，是醫療服務的重要里程碑。

The HKCH plays a key role in the development of genetics and genomic medicine in Hong Kong. It is the territory-wide referral centre for uncommon genetic disorders. It gathers expertise of related fields and advanced testing technologies to enhance the treatment, research, talent pool and knowledge of these diseases.

A new milestone has been reached recently, as HKCH has taken up the Clinical Genetic Service previously under the Department of Health, including its genetic counselling clinic, genetic screening clinic, neonatal screening programme, and genetic laboratory. Patients with genetic diseases and their families could obtain one-stop multi-disciplinary support, including diagnosis, treatment, subsequent management, counselling and prevention.

醫學遺傳科

一站式支援不常見病患者家庭

Clinical Genetics Service

offers one-stop care for families with uncommon disorders

香港兒童醫院的醫學遺傳科是醫管局首個提供臨床遺傳服務的單位。團隊成員包括遺傳學及基因組學專科（兒科）醫生、護士、遺傳輔導員和生物信息學家，為患有或懷疑有各種遺傳病的病人提供評估、檢驗、診斷、治療、輔導及預防的一站式服務。

團隊製作了單張和影片，增加大眾對醫學遺傳服務的認識。病人就診和做基因檢測前亦可預先消化資料，做好準備。

Educational leaflets and videos are produced to enhance public understanding of clinical genetics services. They also prepare patients for their upcoming consultation and genetic tests.

The HKCH Clinical Genetics Service Unit (CGSU) is the first such unit in the Hospital Authority. The team consists of specialists in Genetics and Genomics (Paediatrics), nurses, genetic counsellors and bioinformaticians. Serving patients with suspected or confirmed genetic diseases, it provides one-stop care including assessment, investigation, diagnosis, treatment, counselling and prevention.

醫學遺傳科顧問醫生陸浩明表示，有別於本院其他專科，該科服務所有年齡層的病人：「任何懷疑或確診遺傳病的病人或其家人都是我們的服務對象。我們最常見的是小朋友有特別病徵，兒科醫生擔心是遺傳綜合病，也有孕婦產前檢查發現胎兒異常或有家族史。此外，成年病人如有一些非常見的病徵，或很早發病的情況，都應該考慮轉介給我們進行評估。」

以家庭為本的遺傳輔導

遺傳輔導是醫學遺傳科的重點服務。醫生初次見病人時會詢問其病歷及家族健康史，按照有關資料繪畫譜系圖，所謂「查三代」，然後再做臨床檢查。醫生評估後，如懷疑是遺傳病便會建議病人做基因檢測以作診斷。陸醫生說：「到出了基因檢測報告，也不是正常或不正常這麼簡單。我們要詳細向病人講解，例如由哪個基因引致、潛在病徵和併發症、遺傳模式、預防方法、生育風險等，如有需要，會轉介病人到其他專科跟進。最特別的是，我們不只看一個病人，而是以整個家庭為單位。如擔心病症會同時影響其家人，便會邀請他們前來一起接受檢查和遺傳輔導。」

帶患者走出診斷迷宮

「有病人窮一生都找不到病因。雖然很多遺傳病目前沒有根治方法，但患者經我們確診了，能走出診斷迷宮，始終是一件好事，找到答案亦有助生育計劃。」曾有父母連續兩個孩子都有同一不常見病症，後來經檢測找到基因變異。當他們準備生第三個時，便透過試管嬰兒方法，選擇無遺傳病的胚胎植入，增加健康孩子出世的機會。

跨專科會診 集思廣益

醫學遺傳科亦設有跨專科診所，跟皮膚科、眼科、內分泌科、骨科、腦神經科等合作。陸醫生說：「沒有一個醫生會認識所有病症。遇上與遺傳相關兼病情複雜的病人，我們需要與不同專科一起跟進治理。」

進行遺傳輔導時，醫護人員會用上簡明圖像，向患者解釋其疾病在家族裡的遺傳方式和復現率。

During a genetic counselling session, simple diagrams are often used to illustrate the mode of inheritance and recurrence risk within the family.

醫學遺傳科顧問醫生陸浩明指，有些病人多年來不斷求醫，最後通過醫學遺傳科的幫助而確診，紓解了心靈困擾，亦有助往後的治療及預防。

Dr Luk Ho-ming, consultant in-charge of the Clinical Genetics Service Unit remarks that some patients have been seeking for answers for years. Through the team's help in reaching a diagnosis, patients and their families could find psychological relief. It also facilitates subsequent disease management and prevention.

Dr Luk Ho-ming, consultant in-charge of CGSU stated that unlike other HKCH clinical departments, they serve patients of all ages, "Any patient or family suspected or confirmed to have genetic diseases are potentially our clients. Our clinic mostly provides consultation for children who are referred by paediatricians, worrying that they may have genetic syndrome. There are also pregnant women with abnormal prenatal exam results or related family history. Some adults have very unique symptoms or abnormally early age of onset. They should also be considered for referral to us for assessment."

Family-based genetic counselling

Genetic counselling is one of the key services of CGSU. During the first appointment, the doctor will ask patients about their medical and family history and perform physical examinations. A family tree will be drawn for pedigree analysis. After assessment, if the patients are suspected to have genetic disease, the doctor will advise them to undergo appropriate genetic tests. Dr Luk said, "The genetic report is not just a simple positive or negative. We have to explain its meanings to the patients thoroughly, such as the disease-causing gene, potential symptoms and complications, mode of inheritance, means of prevention, and reproductive risk. If needed, we would refer the patients to other specialties for follow-up. We serve and counsel the whole family, not just individual patients. If there is concern that the problem may affect other family members, we would invite them all to come for assessment and genetic counselling."

Ending the diagnostic odyssey

"Some patients spend their entire life looking for the cause of their disease, but to no avail. Even though many genetic diseases have no cure now, if we are able to make a diagnosis, it still means a lot to them by putting an end to the odyssey, and could assist their clinical management and family planning," Dr Luk said. He shared that a couple had given birth to two children with the same uncommon disease. Subsequent test identified a genetic mutation in both. When the parents decided to have their third child, they underwent in vitro fertilization and were able to select a normal embryo to increase the chance of carrying a healthy baby.

醫學遺傳科處理的病症獨特罕有，因此團隊經常進行病人個案會議，共商對策，例如尋求最新的診斷方法。

The CGSU handles genetic diseases that are rare and unique. Regular meetings are held to discuss how to tackle cases.

Multi-disciplinary management for the best results

CGSU had set up cross-specialty clinics in collaboration with dermatology, ophthalmology, endocrinology, orthopaedics and neurology etc. Dr Luk remarked, "No single doctor knows all the diseases in the world. Patients with complex genetic conditions require joint management by different specialties."

醫學遺傳科的成立，造就了遺傳輔導員和生物信息學家這兩個新職位的產生。

The establishment of CGSU has created two new positions, namely genetic counsellor and bioinformatician.

生物信息學家 — 數據解讀者

檢測的原始數據如漫天繁星，如何理出頭緒？生物信息學家姚永聰解釋：「我們會選取適當的生物信息工具及數據庫，去整理及解讀化驗室輸出的結果，為醫生提供診斷依據。我們亦要管理大量數據，快而準地找到和臨床決策相關的資訊。」

這份工作亦需要偵探頭腦。「之前我們的數據庫內，有同一個基因變異多次被分類為臨床意義不明。醫生發現這些記錄全部來自同一家族，於是交給我們分析，最終成功重新歸類為可致病的變異，可採取適切治療。」

雖然姚永聰多數對著電腦，但能夠參與診斷及治療過程卻使他感到責任重大：「因為每一個決定都有可能影響病人的治理和生活質素。」

姚永聰的本科是生物化學，之後攻讀基因組學及生物信息學和醫務化驗科學碩士，希望將自己的專業技能在臨床環境中實踐。

After graduating from biochemistry, Yiu Wing-chung pursued further studies in genomics and bioinformatics, as well as medical laboratory science. He wishes to apply his professional skills in clinical setting.

Bioinformatician: the data interpreter

How to look for clues from the galaxy of raw genetic data? Bioinformatician Yiu Wing-chung revealed, “We select appropriate bioinformatics tools and database to collate and analyze the data generated in the lab. Our interpretation can facilitate doctors in making diagnosis. We also need to maintain massive data effectively, so that useful information can be promptly retrieved as a basis for clinical decisions.”

A bioinformatician needs to think like a detective. Said Yiu, “Previously in our database, there was a genetic mutation repeatedly classified as 'uncertain significance'. One doctor noticed that all entries actually came from the same family, and asked us to look into it. Eventually, we successfully re-classified it as a disease-causing mutation, which enabled doctors to make suitable treatment plan.”

Though Yiu spends most time with the computer, he feels great responsibility through taking part in the diagnosis and treatment process, "Every decision may influence the disease management of patients and their quality of life.”

香港基因組計劃夥伴中心

政府2021年開展香港基因組計劃，涵蓋未能確診病症、遺傳相關癌症，及與精準醫學有關的個案，旨在為病人提供準確診斷和個人化治療，同時建立本地基因組數據庫，促進科研及基因組的臨床應用。

本院是計劃其中一間夥伴中心，負責招募合適的患者，自願接受全基因組排序。陸浩明醫生指出：「一般的臨床測試有侷限性，但全基因組測序非常複雜費時，不會是首重選擇。我們會挑選合適的病人和家屬，邀請他們參與這個研究項目，希望為他們診斷。」

本院至今已招募了二千多個家庭，部分已收到報告，當中有些更成功找出病因。院方未來數年會繼續轉介合適的病人參加計劃。

建立不常見疾病資料庫

本院在2021-22年度開始，逐步為個別不常見疾病建立資料庫，冀為醫療服務規劃和臨床診斷治療提供參考。

編製資料庫的方法是首先擷取醫管局臨床系統的現存紀錄，再經醫護人員核實並進行分析。資料庫會列出每種疾病的病人總數、年齡組別、性別，及在世或死亡狀況。

現時資料庫涵蓋25種先天性代謝病、四種腦科疾病和一種內分泌疾病，如典型苯丙酮尿症、先天性腎上腺增生症、脊髓肌肉萎縮症及結節性硬化症等，未來會繼續增加更多病種資料。

本院參與香港基因組計劃的團隊。

HKCH team members who are involved in the Hong Kong Genome Project.

遺傳輔導員 — 不只是傾偈

進行基因檢測前，遺傳輔導員會與病人詳談，以取得其知情同意，新症一般要花超過一小時。遺傳輔導員胡鈞惠說：「我們會解釋檢測目的和侷限，並解答種種疑慮，例如有些人擔心一旦發現身體有其他疾病風險，會買不到保險，影響升學就業。」完成檢測後，他們會講解報告，包括是否找到問題所在、會否影響家人、下一步如何跟進等。她坦言工作極具挑戰性：「有些病連我也未聽過，所以每次見病人前都要做足功課。」

對於患者而言，心靈可能比身體更痛。胡鈞惠分享初入職時，一位媽媽帶著有發展遲緩及智力問題的孩子前來：「她哭訴多年來一直自責，擔心懷孕時的飲食影響了孩子。經過輔導後，她終於決定為孩子進行檢測。離開時，她感謝我傾聽她的經歷。此案例讓我體會到這份工作不單要有知識， 還要有同理心及輔導技巧去處理病人和家屬的情緒。」

Genetic counsellor: not just chatting

A genetic counsellor's role starts at obtaining patients' consent to undergo genetic testing, and a new case often takes more than an hour. Genetic counsellor Phoebe Wu said, “We inform patients the purpose and limitation of the test, and answer all queries they have. Some people worry the findings will affect their insurance, job and education prospect.” After the test, she will explain the report in detail, including whether a problem is found, impact on family members, and management plan. Phoebe admitted her job is challenging, “There are diseases which I have never heard of. I have to study hard before meeting my patients.”

For many families, the psychological pressure is greater than physical pain. Phoebe once counselled a mother who came with a child with developmental delay and intellectual problem. "She cried the whole time, fearing that her dietary choices during pregnancy had affected her child. After counselling, she finally decided to have her child undergo medical tests. As she left, she thanked me for listening to her story. This case made me realize that this job requires not only expertise, but also empathy and counselling skills to manage the emotional needs of patients and their families."

胡鈞惠本身為護士，修讀醫學遺傳學碩士課程後成為遺傳輔導員，希望教育大眾認識遺傳病，減少歧視。

Once a nurse, Phoebe Wu obtained a master's degree in medical genetics and became a genetic counsellor. Through public education, she hopes to eliminate discrimination towards patients with genetic diseases.

Partnering centre of Hong Kong Genome Project

The government launched the Hong Kong Genome Project (HKGP) in 2021. It covers cases with undiagnosed diseases, hereditary cancers, and those related to precision health, aiming to benefit patients with more precise diagnoses and personalized treatment. At the same time, the data collected will be used to build a genome database of the local population to facilitate the clinical application of genomic medicine and scientific research.

The HKCH is one of the partnering centres of HKGP to recruit suitable patients for receiving whole genome sequencing on a voluntary basis. Dr Luk Ho-ming said, “Regular clinical tests have their own limitations. However, whole genomic sequencing will not be the first choice because it is very complex and time-consuming. We target to invite suitable patients and their families to join this research project, hoping to give them an answer.”

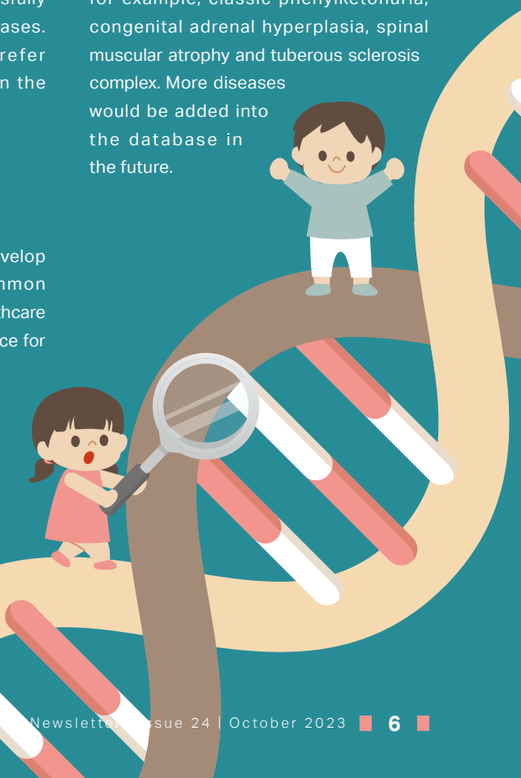
More than 2,000 families have been recruited through HKCH so far. Some have already received their reports, and a proportion of them have successfully identified the cause of their diseases. The hospital will continue to refer eligible patients to the project in the next few years.

Uncommon disorders database

In 2021-22, HKCH has begun to develop a database for individual uncommon disorders in phases to facilitate healthcare service planning and provide reference for diagnosis and treatment.

The data is derived from existing records in the HA clinical information system, which is then verified and analysed by healthcare professionals. The database lists the total patient number, age group, gender, and alive / death status of each disease.

At present, the database covers 25 metabolic diseases, four neurological diseases and one endocrine disease, for example, classic phenylketonuria, congenital adrenal hyperplasia, spinal muscular atrophy and tuberous sclerosis complex. More diseases would be added into the database in the future.



科技打破診斷框框 拯救生命

Technologies transform diagnostics and save lives

工欲善其事，必先利其器。為對付不常見複雜病症，本院配備先進儀器，提供多種精準化驗，為患者偵查病因，預視及監測病情發展，協助臨床團隊制訂合適的治療和預防方案。

In the war against uncommon and complex diseases, HKCH is armed with an arsenal of advanced equipment to provide a wide spectrum of tests for precise diagnosis, prognostication and monitoring, enabling clinicians to formulate appropriate treatment and preventive plans.

本院的病理學部設有醫管局唯一的「遺傳及基因組學」分部，為不同專科轉介的病人以至他們的家族成員進行遺傳病和癌症相關化驗，亦提供全港性的無創性胎兒染色體篩查，針對唐氏綜合症等疾病。

病理學部顧問醫生袁月冰表示，遺傳及基因組學的臨床應用在世界上越趨廣泛。本院率先引入次世代定序儀，能進行大規模基因測序，作全面的診斷分析。她說：「檢測方法各有優勢與侷限，要考慮成本效益和數據量，最重要是病人需要。如病人有典型病徵，或涉及單基因遺傳病，用傳統方法如桑格定序去檢視目標基因片段會比較直接省時。至於茫無頭緒的個案，如抽筋、發展遲緩可以有很多解釋，廣泛測序則更有機會找出病因。」

The HKCH Department of Pathology set up the only genetic and genomic division in the Hospital Authority. It conducts tests for various inherited diseases for patients and family members referred by different clinical specialties, as well as childhood cancers. Another important service is the territory-wide non-invasive prenatal testing for chromosomal disorders like Down syndrome.

HKCH is equipped with next generation sequencers (NGS) which can perform massively parallel sequencing of DNA to yield comprehensive analysis results. Dr Liz Yuen, Consultant (Pathology) said, "Different technologies have their own strengths and limitations. For diseases caused by single gene mutation or when there are classic symptoms, we may advise using conventional methods like Sanger sequencing to look at the targeted DNA fragments. But for cases without leads, such as patients presented with seizures and developmental delay, NGS has a better chance to find out what's wrong with them."

袁月冰醫生說：「化驗項目越來越複雜，我們的知識和技術都要快速增長去配合，不可閉門造車，要留意新科技發展，適時引入。」

Dr Liz Yuen said, "As modern tests have become more and more complicated, there is a constant need for us to catch up with the latest knowledge and technologies."

麥苗醫生說：「香港大多數家庭只有一兩個孩子，十分矜貴，真的一個也不能少。新生兒篩查是政府、醫護人員和父母共同給孩子的禮物，為迎接人生做好準備。」

Dr Chloe Mak said. "With the low birth rate in Hong Kong, every child is precious. Newborn screening is a gift from the government, healthcare workers and parents to prepare for their life journey."

兒童為本的化驗服務

另一邊廂，病理學部顧問醫生麥苗的團隊亦不斷推出新的化驗項目。看似冰冷的化驗室，原來為病童灌注了不少心思。麥醫生說：「小朋友怕打針抽血，我們特意採購只需靠少量化驗樣本的儀器，減輕他們的痛苦。另外，化驗結果的參考值往往以成人為基準，套用在橫跨不同年齡的兒童身上不太合適，於是我們對比不同文獻並一點一滴建立自己的本地數據，希望更準確判斷病童的結果是否有異常。」

本院也負責支援全港性的初生嬰兒篩查計劃，涵蓋在公立醫院出生的所有嬰兒，讓患兒能盡早接受治療，有機會健康成長。麥醫生預期篩查範圍會繼續擴大，加入更多病種，盼跟國際看齊。

Children-centred pathology services

Meanwhile, the team led by Dr Chloe Mak, Consultant (Pathology) also works tirelessly to roll out new lab tests, and special consideration is given to meet children's need. She explained, "No child likes needles. To ease their pain and anxiety, we bought analyzers that require only a little sample volume. Moreover, the reference intervals of lab results are mainly set for adults. Therefore, we make references to various literature and build up our own local data to adjust the values accordingly, so we could really tell if a child's result is normal or not."

HKCH also shoulders the task of screening all babies born in public hospitals, so they could receive timely treatment when problem is detected. Dr Mak expects more diseases to be added to the screening panel in the future to meet international standards.

香港兒童醫院的初生嬰兒篩查項目

Newborn screening by HKCH

- 26種先天性代謝病
  26 inborn errors of metabolism
- 嚴重聯合免疫缺陷病
  Severe combined immune deficiency
- 脊髓肌肉萎縮症（先導）
  Spinal muscular atrophy (pilot)
- 葡萄糖六磷酸去氫酵素缺乏症（蠶豆症）
  Glucose-6-phosphate dehydrogenase deficiency
- 先天性甲狀腺功能不足
  Congenital hypothyroidism

新生兒篩查救一命

「女兒出生數天我們接到電話，說她免疫力偏低，要立即入院進一步檢查。我們覺得很感恩和幸運，能夠在她未受感染前發現患病，而我的骨髓亦正好吻合，可以捐給她。她現已康復，跟普通小朋友沒分別。」年約半歲的 Aaira 是嚴重聯合免疫缺陷病新生兒篩查計劃推出以來第一個確診個案，她爸爸 Javed 對這段經歷仍歷歷在目。

這病症由基因突變引起，患者免疫系統非常弱，即使普通傷風，甚至接種減毒活疫苗，也可引發反覆感染。如未經治療，病童通常活不過兩歲。免疫科榮譽顧問醫生李珮華表示：「未有篩查時，病童由轉介到第一次治療可能要三至四個月，大大增加嚴重感染的風險。現在整個篩查計劃及跟進在兒童醫院進行，制訂了清晰流程，加上不同專科緊密合作，省卻轉介，更有效率。」

A life saved by newborn screening

"We received a phone call a few days after our daughter was born, telling us to bring her to the hospital immediately for further check-up. We feel very grateful and lucky that she was diagnosed before any infection occurred, and my bone marrow matched so I could donate it to her. Now she has recovered, and she is just like any other normal child." Baby Aaira was the first confirmed case picked up by the newborn screening programme for severe combined immune deficiency (SCID). Her father, Javed, still vividly remembers this extraordinary journey.

SCID is caused by gene mutation. Patients' immune system is very weak, so even a common cold or live-attenuated vaccine can induce repeated infections. Without treatment, infants usually die within the first two years. Dr Pamela Lee, Honorary Consultant in the Immunology team said, "Before screening is available, it would take three to four months from a patient's referral to the first treatment, which increases the risk of severe infection. Now the programme is hosted in HKCH with well-established workflow and multidisciplinary teamwork, referral time is minimized and the whole process becomes much more efficient."

Aaira 正接受造血幹細胞移植，這是嚴重聯合免疫缺陷病的根治性治療。

Aaira receiving haematopoietic stem cell transplant, the definitive treatment for SCID.

當發現 Aaira 的篩查結果異常，院方便即時安排她入院接受保護性隔離，並再次驗血，一天後確診。由於 Aaira 的 T 細胞數量低，可說是沒有抵抗力，所以首先將爸爸捐出的 T 細胞注入她體內，給予暫時的免疫保護，免受可致命的病原體侵害。

Aaira 最終在七周大時，移植了爸爸捐出的造血幹細胞，重建健康的免疫系統。李醫生說：「移植非常成功，現時她定期覆診，康復進度良好，可說是完全根治。」

When Aaira's screening result was found abnormal, she was immediately admitted into the Special Care Baby Unit of HKCH for protective isolation. Blood tests were performed and she was genetically confirmed to have SCID the day after. Since Aaira had a low T-cell count, she basically had no immunity. As an interim treatment, T-cells from her father were infused to help her develop basic resistance to life-threatening pathogens.

Aaira eventually received a haematopoietic stem cell transplant at seven weeks to rebuild a healthy immune system. Said Dr Lee, "The transplant was very successful. Now she comes back regularly for follow-up and is making good recovery progress. We can say she has a complete cure."

李珮華醫生感謝各團隊的合作，讓 Aaira 能夠極速無縫地接受所需檢查和治療。她指有關篩查在先導階段獲香港弱能兒童護助會支持，至現在發展為政府資助的恆常服務。

Dr Pamela Lee attributes Aaira's efficient and seamless treatment process to the collaboration across different specialties. She also thanks the Society for the Relief of Disabled Children's support in the pilot phase of the SCID screening programme, which has now become a regular public service funded by the government.

SCID treatment journey

Aaira 的治療之旅

1 醫生將 T 細胞注入 Aaira 體內，為她建立第一道免疫屏障。

Doctors infuse T-cells into Aaira's body to give her the first immune barrier.

2 化驗室人員小心處理 Aaira 爸爸捐出的造血幹細胞，隨即供她移植。

Stem cells donated by Aaira's father are carefully processed in the laboratory before being transplanted.

3 Aaira 移植後約一個月出院。Javed 與太太感謝醫護團隊用心醫治女兒，並在覆診時安排翻譯，確保溝通順利。

Aaira was discharged about a month after the transplant. Javed and his wife thank the clinical teams for taking good care of their daughter. They are also pleased to have an interpreter during follow-up appointment to ensure smooth communication.

陳寶璘（右）與「石頭症」症患者泉欣（中）關係密切。專責護士的協調有助跨專科團隊制訂以病人為本的治療計劃。

Candy Chan (right) closely follows the needs of patients like Chuen Yan (middle). The coordination by designated nurses helps to develop patient-centred care plan across different services.

專責護士 一路相伴

Designated nurses guide patients through treatment journey

罹患嚴重不常見疾病的病童需集合眾人之力悉心照顧。香港兒童醫院現時於內分泌科及腦神經科設有專責護士，擔當病童家庭與不同醫院和跨專科團隊之間的橋樑，負責協調各項檢查、治療、復康訓練和病情監察，令他們獲得更簡便、全面、個人化的醫療服務和社區支援。

It takes a village to raise a child. This is especially true for children with serious uncommon diseases. In HKCH, there are now designated nurses under endocrinology and neurology to take care of these patients. They serve as a bridge between families and different hospitals / multidisciplinary teams to coordinate the patients' investigations, treatments, rehab training and disease monitoring, so they can receive accessible, holistic, personalized medical care and community support.

八歲的泉欣患有俗稱「石頭症」的進行性肌肉骨化症。此病十分罕有，由基因突變引發，患者的肌肉和筋腱受傷後有機會轉化成新骨頭，令骨骼變形，逐漸喪失活動能力，並影響身體其他功能。

陳寶璘是泉欣的專責護士，即第一線聯絡人。媽媽蕭太說：「女兒早前一直發燒，我打電話向陳姑娘求助。她耐心地教我怎樣應對，並安排提早覆診見醫生，令我放下心頭大石，真的非常感謝她。」

與病童的學校溝通亦是陳姑娘的工作之一。她說：「我們曾舉辦講座，向老師們講解泉欣的病情，例如預防受傷措施和突發狀況處理，盼為她提供安全環境和適當照顧。」

泉欣說：「體育老師特別關心我，總是在運動前問我能不能參與。」蕭太憶述：「有一次，泉欣被同學撞倒了。老師反應迅速，帶她到醫療室消毒傷口。我亦即時趕到，讓她服用醫生預先處方的類固醇以減輕炎症，幸運地患處事後沒有變成骨頭。」

專責護士計劃的另一個重要目標，是因應病童不同成長階段的需要提供相應支援，提升他們的生活質素和長遠自我管理疾病的能力，順利融入社區。

Eight-year-old Chuen Yan has fibrodysplasia ossificans progressiva, nicknamed "stone man syndrome", a rare condition caused by gene mutation. Patients' muscles, tendons and ligaments may grow into new bones after an injury, causing skeletal malformation and loss of mobility over time. Other bodily functions may be affected as well.

Candy Chan is the designated nurse and first contact point for Chuen Yan. Her mother, Mrs Siu said, "Chuen Yan had a persistent fever recently, so I called Candy for advice. She taught me how to deal with it and arranged an early follow-up with the doctor. I am so grateful to have her guide me through all these."

Communicating with schools is also part of Candy's job. "We held a talk to explain Chuen Yan's situation to her teachers, such as injury prevention and emergency handling, so she could have a safe environment and appropriate care."

Chuen Yan said, "My PE teacher is especially caring. He always makes sure if I can join certain sports." Mrs Siu shared, "One time, Chuen Yan was knocked down by a classmate. The teacher reacted quickly and helped cleanse her wound. I rushed to the school and gave her steroid prescribed by the doctor to suppress the inflammation. Luckily, the injured area did not become bone afterwards."

The designated nurse programme also aims to support patients' changing needs at different life phases, enhance their quality of life, empower them for life-long self-care, and facilitate a smooth integration into the community.

基因治療 NOVEL GENE THERAPY brings new hope 燃點新希望

這項試驗的對象為確診杜興氏肌肉萎縮症，並符合相關臨床條件的四至七歲男童。陳凱珊醫生（右）正監察一名參加者按照研究的規定接受輸注。

Dr Sophelia Chan (right) watches a patient undergo infusion according to protocol. The study has recruited boys aged four to seven who are diagnosed with Duchenne muscular dystrophy and fulfill specific clinical criteria.

患杜興氏肌肉萎縮症的兒童因基因突變，身體無法製造一種重要蛋白，導致肌肉逐漸退化及受損。大多數病人最終要使用輪椅，並會在20至40歲因心臟或呼吸衰竭離世，現時並無治癒方法。香港兒童醫院正參與一項跨國性第三期臨床試驗，研究基因治療對杜興氏肌肉萎縮症的成效及安全性，冀尋求治療突破。

香港大學臨床助理教授兼兒童醫院腦神經科榮譽副顧問醫生陳凱珊領導本院這項研究。她解釋，這是香港首次進行基因治療的臨床試驗，方法是透過一次性靜脈輸注，利用病毒載體把經改造的基因傳遞到患者的肌肉細胞，指令它們製造一種較短且具功能性的蛋白，從而控制甚至逆轉病情。

Due to gene mutation, children with Duchenne muscular dystrophy (DMD) fail to produce an essential protein, causing progressive muscle degeneration and damage. No curative treatment is available at the moment. Most patients will end up in a wheelchair, and pass away between 20 and 40 because of heart or respiratory failure. HKCH is now part of a multinational phase III clinical trial to study the efficacy and safety of using gene therapy to treat DMD.

Dr Sophelia Chan, Clinical Assistant Professor of the University of Hong Kong (HKU) and Honorary Associate Consultant in the HKCH Neurology team is the principal investigator leading this gene therapy trial at HKCH, which is the first in Hong Kong. Through one-time intravenous infusion, a viral vector is used to deliver an engineered gene to the patients' muscle cells, instructing them to produce a shortened and functional version of the needed protein, which could potentially control and even alter the disease progression.

> 感謝院方和香港大學支持，亦有賴多個臨床和科研團隊通力合作，為病童爭取這次難得的機會。我期望印證基因治療的成效，幫助更多患者。
>
> This is a precious opportunity for our patients, made possible by the support of hospital management and the HKU, as well as the collaboration among various clinical and research teams. I hope it can prove the effectiveness of gene therapy and help more DMD patients.
>
> 陳凱珊醫生
> Dr Sophelia Chan

研究在全球九個地區超過40間中心進行，對人手、設施、病人篩選、製劑預備、注射安排、跟進評估和數據收集均有嚴格要求。本院團隊審慎計劃整個流程，並接受了相關培訓。

The clinical trial is conducted at over 40 centres in nine regions across the globe. There are high standards for manpower, facilities, patient selection, dose preparation, injection arrangement, follow up assessment and data collection. The teams at HKCH have mapped out the workflow carefully and underwent related training.

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