Hong Kong Genome Project

Our Life-changing Stories

香港基因組計劃

改變生命的故事

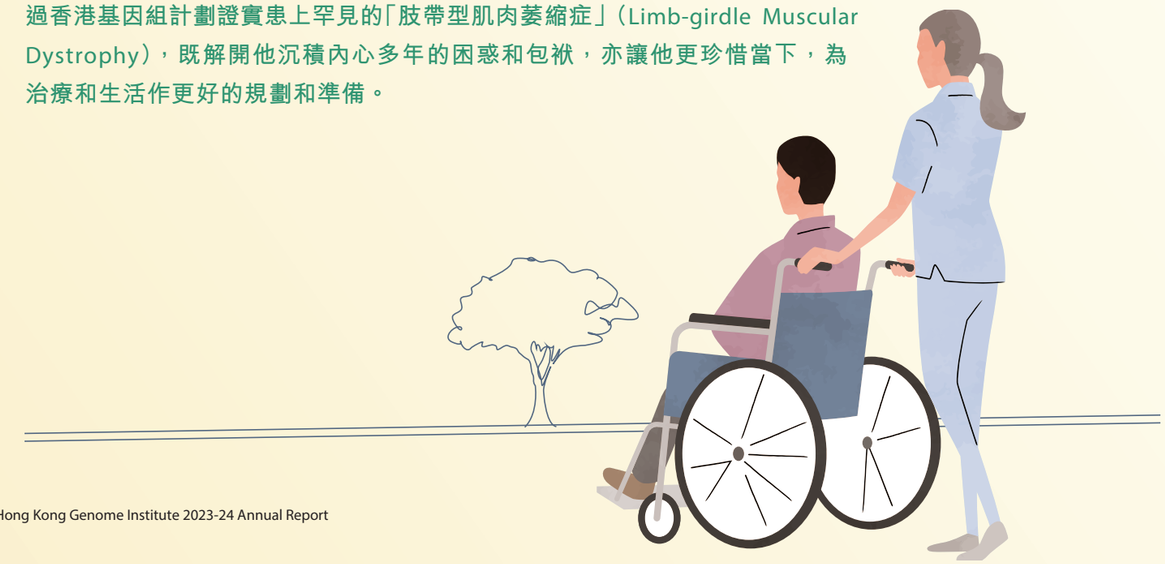
Case 個案 1

Unlocking the Mystery and Triumphing over Life's Trials

解開病因之謎 跨越人生障礙賽

Life has a way of challenging our resolve with unexpected twists and turns. Ka-ho (a pseudonym), a sports enthusiast, was thrown a curveball when he developed muscle weakness. Suddenly, the simple joys of running and jumping faded into distant memory, and he spent over a decade in search of answers. Fortunately, Ka-ho's physical limitations are no match for his inner strength. The resilience he has developed from sports training means he does not easily give in. Ultimately, the Hong Kong Genome Project (HKGP) confirmed he is suffering from a rare disease known as Limb-girdle Muscular Dystrophy (LGMD), putting an end to the deep-seated confusion and burden he has been carrying around for years. Now, he is able to seize every moment and plan his treatment and future with renewed determination.

老天總愛以生命的無常磨煉我們的意志。很多事情，總是意料之外。家豪（化名）熱愛運動，卻患上肌肉無力的頑疾，跑跳動彈成了他遙不可及的夢，更花上超過十年尋找患病原因。幸而身體上的囿限及不上他內心的強大，由運動鍛鍊出來的堅韌令他不輕易屈服，最後透過香港基因組計劃證實患上罕見的「肢帶型肌肉萎縮症」（Limb-girdle Muscular Dystrophy），既解開他沉積內心多年的困惑和包袱，亦讓他更珍惜當下，為治療和生活作更好的規劃和準備。



Ka-ho has always been a sports enthusiast, with a passion for sports that began at a young age, defining a life filled with dynamic movement. However, as he approached his 40th year while pursuing his ambitions, an unexpected health challenge crept up. At the age of 40, Ka-ho started experiencing weakness in his legs and pain in his back, while his walking pace gradually slowed. He dismissed these symptoms as muscle fatigue from exercise, but it was not until standing also became a struggle that he recognised the seriousness of his condition and sought medical attention. This marked the start of a decade-long quest to uncover the mysteries behind his condition.

家豪是一名運動健將，從小活潑愛動，沒有一刻定下來。然而，當他步入不惑之年，努力實踐抱負之際，頑疾卻悄然降臨。40歲的家豪起初發覺腿部無力、背部疼痛，走路越來越慢，本以為是運動後的肌肉勞損，並沒有特別在意；直至連站立也開始變得困難，他才意識到問題嚴重，需要求醫，自此展開了一場持續十數載的尋找病因之旅。

Whole Genome Sequencing Unravels a Decade Mystery

Over the years, Ka-ho continuously sought medical advice and underwent numerous tests. Despite completing all relevant examinations, a definitive diagnosis remained elusive. Fortunately, Ka-ho remained undeterred. In 2021, 12 years after his symptoms first appeared, he was referred by his hospital to the newly launched HKGP. The dedicated HKGP team conducted whole genome sequencing (WGS), which finally revealed a genetic mutation, confirming that Ka-ho has a rare disease known as LGMD Type 2A.

This rare condition is a subtype of LGMD. The onset of the disease is unpredictable, with muscle weakness and atrophy affecting the shoulders, hips and torso, progressively worsening over time. Patients typically experience tightening of the Achilles tendon, leading to challenges in mobility.

全基因組測序解開十年心結

這些年來，家豪不停見醫生，做檢查；只是，即使做遍所有相關測試和檢查，仍然無法確認他所患何病。幸而家豪未有輕言放棄，在發病後的第12個年頭，即2021年，他在醫院轉介下，參加了當時剛推行的香港基因組計劃。經計劃團隊安排，家豪進行全基因組測序，終於找到基因變異之處，證實患上名為「肢帶型肌肉萎縮症2A型」的罕見病。

此罕見病屬「肢帶型肌肉萎縮症」亞型，發病時間不固定，肌肉無力和萎縮情況集中出現在肩帶、腰帶肌肉群以及軀幹，並會隨著病情發展而逐步加重，病人一般會出現腳跟肌腱收縮變形，導致不良於行。

Ka-ho vividly recalled the moment when he received the explanation of the sequencing results. After the initial shock and intense emotion, he soon felt a profound sense of relief washing over him. At long last, he could put his inner turmoil behind him, emerging from the fog of doubt. Understanding the trajectory of his illness, Ka-ho had a clear direction for planning his future.

醫生講解測序結果的一幕，家豪至今記憶猶新，一陣錯愕和激動過後，更多的是如釋重負，因為他終於可以解開多年心結，走出迷霧，知道自己病情的走向，對規劃未來也有方向和準備。

When it came to clinical care, with the analysis results from WGS, healthcare professionals were able to develop a personalised disease management plan for Ka-ho. Their primary goal was to slow the progression of his conditions while focusing on improving and preserving his mobility. In addition to physiotherapy, Ka-ho follows his therapist's advice by performing daily home exercises, such as leg stretches, to maintain muscle strength. He has also gained a better understanding of his disease's progression and potential challenges he may face. He proactively embraces the early adoption of various assistive tools, such as canes and wheelchairs, ensuring that his condition does not limit his future quality of life.

在臨床護理上，有著基因組測序的分析結果，醫護專業人員得以為家豪安排個人化的病情管理方案，助他延緩病情，以改善及保存行動力為首要目標。除了積極接受物理治療，家豪亦按照治療師的建議，每天進行居家運動，例如伸展腿部訓練肌力。同時，他亦深入了解到這個病的進程，以及自己將要面對的挑戰，因而及早了解和學習使用各種輔助生活的工具，例如拐杖和輪椅，不讓頑疾限制日後的生活。

A Rocky Road, Yet Unshaken with an Unbreakable Spirit

Ka-ho's willpower and perseverance honed through his years of sports training carried him through his decade-long quest for answers. With the help of HKGP, he finally found the explanation he was looking for, allowing him to find peace and lift a heavy burden off his shoulders. He is now tackling his condition head-on, adhering to a personalised disease management plan designed by HKGP's team, in the hope of slowing the progression of his symptoms.

It is said: "When the mountain won't yield, the path must change. When the path is unyielding, one must adapt. And when adaptation seems impossible, it's time to shift one's mindset." With this change in perspective, Ka-ho has chosen to live fully in the present, plan for his future, and resolutely face life's hurdles.

路縱崎嶇　心更堅毅

從運動鍛鍊出來的意志和毅力，助家豪走過十多個寒暑的尋找病因之旅，最後在香港基因組計劃協助下覓得答案，得以釋懷，放下心中大石。他亦積極面對，努力配合計劃團隊為他設計的個人化病情管理方案，期望延緩病情惡化。

「山不轉路轉，路不轉人轉，人不轉心轉。」轉念之間，家豪選擇好好活在當下，規劃未來生活，勇敢面對人生障礙賽。

Learn About Limb-girdle Muscular Dystrophy
認識肢帶型肌肉萎縮症

Disease Incidence

Limb-girdle Muscular Dystrophy (LGMD) can be classified into more than 20 subtypes, including Type 2A. On average, there are only about one to nine cases per 100,000 individuals. The condition is caused by mutations in the *CAPN3* gene on chromosome 15, resulting in deficiencies in the calcium-activated protease (Calpain-3) within muscle cells, which affects the normal function of skeletal muscle.

發病率

「肢帶型肌肉萎縮症」可細分為20多種亞型，而「肢帶型肌肉萎縮症2A型」是其中之一，平均每10萬人中只有大約一至九個案例。致病原因為位於第15號染色體上的「*CAPN3*基因」發生突變，導致肌肉細胞內鈣離子活化的蛋白分解酶（Calpain-3）出現異常，進而影響正常骨骼肌肉功能。

Symptoms

Different types of LGMD may present various symptoms. However, a common feature among patients is weakness in the lower limb muscles, which causes frequent falls and difficulties in running, stairs climbing, and standing. As the disease progresses, mobility may become increasingly difficult, necessitating the use of wheelchairs and other assistive devices.

Currently, there is no specific treatment for LGMD, and care needs to be personalised to each patient's circumstances. The primary objectives of treatment focus on extending life expectancy and improving quality of life. These goals are achieved through weight management, physiotherapy, and stretching exercises to reduce the risk of muscle tightening, along with the use of supportive medications, respiratory support, and heart health monitoring.

病徵

不同類型的「肢帶型肌肉萎縮症」可能會呈現不同的臨床表徵。然而，病人的共同特徵是下肢肌肉無力，導致他們經常跌倒，難以跑步、上落樓梯或站立。隨著病情進展，病人行走可能變得更困難，需要依賴輪椅及助行工具。

「肢帶型肌肉萎縮症」目前尚未有特定的治療方案，而需要根據每位病人的情況度身訂做。治療的主要目標是延長壽命並提升生活質素，方法包括控制體重、透過物理治療和伸展運動減少攣縮的風險、使用輔助藥物、呼吸輔助器，以及監測心臟健康。

Case 個案 2

From Diagnosis to a Legacy of Love and Hope

病患以外 代代相傳的守護和盼望

Every mother hopes that her children will grow up healthy and happy. For Lai-kuen (a pseudonym), however, who endures the prolonged suffering caused by various symptoms of a rare disease, there is a lingering concern that her children may also encounter similar health challenges.

After years of living with an undiagnosed disease, Lai-kuen's life reached a turning point when she participated in the Hong Kong Genome Project (HKGP) in 2022. The results of whole genome sequencing (WGS) finally revealed that she has Fabry disease, a rare genetic disease. This long-sought diagnosis not only facilitates personalised treatment for her but also allows her asymptomatic daughter to take proactive preventive measures.

每一位母親，都希望孩子能夠健康快樂地成長，奈何患上罕見頑疾，身體長期承受各種病徵所帶來的苦楚，作為母親的麗娟（化名）難免擔憂家人會走上同一條患病崎嶇路。

經歷多年病因未明，她在2022年參加了香港基因組計劃，透過進行全基因組測序，終被診斷罹患罕見遺傳病「法布瑞氏症」（Fabry Disease）。這個尋覓多年的答案，除了讓她得以接受針對性治療，亦助其未發病的女兒及早預防。

As Lai-kuen entered the second half of her life, she remained dedicated to her family's well-being. However, having witnessed both her father and younger brother battle rare inherited disorders, a sense of uneasiness lingers in the back of her mind that she might not be spared.

Upon turning 50, Lai-kuen's body began to show signs of trouble, with elevated levels of protein, potassium, and urine creatinine. As time passed, her condition gradually deteriorated. She found herself facing end-stage renal disease, further complicated by blood clotting in her lower legs.

20 Years Later – Whole Genome Sequencing Unlocks Diagnosis

From her 50s to her 70s, Lai-kuen spent two decades searching for the cause of her illness. At the age of 71, under the attending doctor's referral, she joined the HKGP in 2022. Through the collaborative efforts of a multi-disciplinary team of healthcare professionals, scientists, and researchers, Lai-kuen underwent WGS. Following a thorough assessment and analysis, it was confirmed that Lai-kuen was diagnosed with the rare condition known as Fabry disease.

Fabry disease is primarily caused by mutations in the gene responsible for producing the enzyme α-galactosidase (a-GAL). Patients with this condition lack this crucial enzyme, which prevents the breakdown of fatty substances called "glycolipids" in the body, resulting in an accumulation of these substances in various organs, such as nerves, skin, kidneys, and heart, causing widespread dysfunction throughout the body.

Following the precise molecular diagnosis, Lai-kuen's doctors promptly arranged for her to begin Enzyme Replacement Therapy (ERT), supplementing the deficient enzyme and helping to alleviate her symptoms.

進入人生下半場，麗娟努力為家庭打拼，惟目睹父親及弟弟先後患上罕見遺傳病，她內心一直忐忑不安，擔心自己亦無法倖免。

果不其然，到「入五」之年，她的身體開始出現警號，蛋白質、鉀及尿液肌酐酸等指數水平升高。隨著時日轉移，她的身體狀況持續惡化，已進入腎病末期，小腿亦出現血管栓塞的跡象。

尋醫廿載 全基因組測序拆解患病因由

麗娟從半百人生，走到古稀之年，歷經20年，始終無法找到患病原因。及至71歲，她在主診醫生轉介下，於2022年參加了香港基因組計劃。計劃團隊的跨學科醫護專業人員、科學家和研究員等通力合作，為麗娟進行了全基因組測序和分析，終於確認她患上罕見病「法布瑞氏症」。

此病症主要是由身體內負責製造α-galactosidase（a-GAL）酵素的基因突變所引起，病人身體缺少此酵素，造成體內的「醣脂質」無法進行分解，堆積在神經、皮膚、腎臟及心臟等器官，導致身體各處功能出現問題，無法正常運作。

確定病因後，醫護人員隨即安排麗娟接受酵素替代療法（Enzyme Replacement Therapy），補充身體缺少的特定酵素，助她紓緩病情。

A Beacon for Family Health: Paving the Way for Prevention and Care

Given the hereditary nature of Fabry disease, Lai-kuen's daughter also participated in HKGP and underwent WGS. The analysis confirmed that she, like her mother, has the same rare disease. Fortunately, she has not yet developed symptoms, allowing healthcare professionals to intervene early with ERT to reduce the risk of heart and kidney damage. For Lai-kuen's family members who are considering pregnancy, this finding also underscores the importance of targeted prenatal testing. It can assist in early diagnosis and help mitigate potential disease risk.

Lai-kuen's experience has enlightened her daughter and future generations of her family about their health risks, strengthening their vigilance over their health and enabling them to make informed family planning decisions. Although Lai-kuen was diagnosed in her later years, she can still manage her health through personalised treatment and make the most of her time with her cherished family.

The efforts of the HKGP team have brought significant changes for Lai-kuen, her daughter, and their entire family. Not only did it help Lai-kuen find answers to her own medical condition, but it also made her a guiding light for her family. Her journey has become a legacy of love and hope, protecting her loved ones' health for generations to come.

為家人健康引路 及早預防和治療

考慮到「法布瑞氏症」帶有遺傳性質，麗娟的女兒亦一同參與香港基因組計劃，接受全基因組測序，分析結果確認她與母親一樣患上相同罕見病，猶幸尚未發病，讓醫護專業人員得以提前介入，以酵素替代療法，減低其心臟及腎臟因病受損的風險。若麗娟的家族成員於懷孕時進行針對性的產前檢查，亦將有助及早診斷和預防患病。

麗娟的經歷，讓女兒以至往後每一代的家族成員了解到自身的患病風險，從而加強監察個人健康，及早為家庭計劃作好打算。雖然麗娟於晚年才確診患病，但仍可透過針對性治療，以最大努力做好健康管理，珍惜與家人共聚的時光。

香港基因組計劃團隊的努力，為麗娟以至她的女兒和整個家族帶來重大改變，不但助麗娟找到患病原因，亦讓她成為家人最明亮的引路燈，將經歷化作代代相傳的愛和盼望，守護着摯愛的健康。

Learn About Fabry Disease
認識法布瑞氏症

Disease Incidence

Fabry disease is a rare genetic disease that affects approximately one in 40,000 to 60,000 males. While it is more prevalent in males, females can also inherit the genetic mutations associated with this disease and generally experience milder symptoms.

發病率

「法布瑞氏症」是一種罕見的遺傳性疾病，發病率約為每40,000至60,000名男性中便有一名病人。男性發病率較女性高，而即使基因變異發生在女性身上，其症狀一般較輕微。

Symptoms

Clinical symptoms of Fabry disease include periodic pain in the hands or feet, unexplained gastrointestinal discomfort, and decreased sweating. Most patients experience kidney complications, including declining kidney function that may necessitate dialysis. The disease can also cause irregular heartbeats or heart failure. Angiokeratomas may develop on the skin of the lower abdomen and thighs in patients, and cornea verticillata may be observed.

病徵

「法布瑞氏症」臨床症狀包括四肢末端出現間歇性疼痛、不明原因的腸胃不適、出汗能力減少等。大部份病人的腎臟都會受影響，出現腎功能退化，甚或需要洗腎，亦有機會心律不正或心臟衰竭。此外，病人的下腹、大腿等皮膚上或會出現血管角質瘤，眼睛亦可能出現角膜濁斑。

Currently, there is no complete cure for Fabry disease. However, various treatment options are available, including Enzyme Replacement Therapy and targeted treatments specific to the affected organs and symptoms.

目前「法布瑞氏症」仍無法徹底根治，但有各種治療方法，包括酵素替代療法，以及針對各器官及症狀而進行不同治療。

Case 個案 3

Enduring with Courage, Embracing Stern Challenges

以等待的勇氣 迎戰生命硬仗

Waiting can sometimes be an excruciating process, especially for those with end-stage illnesses who require an organ transplant. The uncertainty, with no end in sight, creates a unique burden that few can fully appreciate. Wai-ling (a pseudonym), a patient with Polycystic Kidney Disease (PKD), understands this plight all too well. Not only must she endure the pain of kidney failure, but she is also in a race against time to receive an organ transplant.

With help from the Hong Kong Genome Project (HKGP) team, Wai-ling received a precise diagnosis and gained access to targeted medication, which slowed the progression of her condition while affording her more time to wait for a suitable organ donor. At the same time, thanks to whole genome sequencing (WGS), the HKGP team was able to quickly and efficiently identify close relatives without the same disease-causing genes, thus increasing the chances of organ donation from family members.

等待，有時是一種難以形容的煎熬，尤其是對於等候器官捐贈的末期病人來說，看不見盡頭的無力感不足為外人道。慧玲（化名）不幸患上「多囊性腎病」（Polycystic Kidney Disease），深深明白等待的百般滋味。她既要承受腎衰竭所帶來的痛苦，亦要與時間競賽，等候器官移植。

香港基因組計劃團隊透過精準診斷，有助慧玲找到針對病因的特效藥延緩病情，為她爭取更多時間等待合適的器官捐贈者。同時，計劃團隊透過全基因組測序，為慧玲快速和有效地找出沒有致病基因的家人，提高近親捐贈器官的機會。

After experiencing frequent upper abdominal pain, Wai-ling, aged 36, underwent medical examinations and was diagnosed with hereditary Polycystic Kidney Disease (PKD). The disease was not new to her, as it had already claimed the lives of several family members. When Wai-ling learnt that she too had inherited this condition, the news deeply affected her, but she nevertheless summoned the strength to face the challenging journey ahead.

As time passed, Wai-ling's condition gradually deteriorated. Cysts first appeared in her kidneys, which then multiplied and began to erode her kidney tissue, ultimately leading to a decline in her kidney function. The cysts not only caused her abdomen to swell, but also led her enlarged kidneys to compress the surrounding organs. This compression made it difficult for her to sit, stand, eat, or even breathe. By the age of 54, her condition had deteriorated to kidney failure, and she needed dialysis to sustain her life. An organ transplant became her only hope.

36歲的慧玲，因經常感到上腹疼痛而接受檢查，被診斷出遺傳性的「多囊性腎病」。對於慧玲來說，這個病並不陌生，因為她家族中已有多位親人同患此病，不幸去世。當她得知自己亦難逃命運的安排，雖感到無奈，仍勇敢面對。

慧玲的病情隨著時間逐漸惡化，先是腎臟開始出現囊腫，其後囊腫日益增多並開始侵蝕腎臟組織，令她的腎功能逐漸下降。這些囊腫更使她的腹部脹大，而腫大的腎臟更擠壓其他器官，導致她坐立、進食，甚至呼吸都出現困難。及至54歲，她的病情已惡化至腎衰竭，需依賴洗腎維生，器官移植成為她重生的唯一希望。

Personalised Treatment Slows Disease Progression

Finding a suitable donor takes time. Amidst this uncertainty, WGS brought her hope. In 2022, at the age of 59, Wai-ling participated in the HKGP and underwent WGS. The analysis revealed a truncating mutation in her genes, confirming a diagnosis of Autosomal Dominant Polycystic Kidney Disease. With this critical information, the HKGP team determined that the targeted medication Tolvaptan could slow the progression of kidney cyst damage, thereby buying her more time to find an organ donor.

The HKGP team also recommended that Wai-ling's family undergo targeted examinations, such as kidney imaging, and identify relatives without the disease-causing gene, potentially increasing the chances of finding a suitable donor. On the other hand, it also allowed her family members to better understand their genetic risks, enabling them to proactively monitor their health, and engage in disease prediction and prevention.

Wai-ling is bravely facing the challenges ahead, finding hope along the way. Thanks to the collaborative efforts of the Hong Kong Genome Institute's professional teams and cutting-edge WGS technologies, Wai-ling was able to identify the cause of her illness and receive personalised, precision treatment that has effectively slowed the progression of her conditions.

個人化治療 延緩病情惡化

尋找合適的捐贈者需時，在充滿未知的等待中，全基因組測序為她帶來了希望。2022年，59歲的慧玲參加了香港基因組計劃，進行全基因組測序，分析結果揭示她的基因出現截斷突變，確診為「常染色體顯性多囊性腎病」（Autosomal Dominant Polycystic Kidney Disease）。根據這個關鍵資訊，計劃團隊確定了針對「多囊性腎病」的特效藥物「Tolvaptan」能夠助慧玲控制病情，延緩腎臟囊腫的破壞速度，為她爭取更多時間尋找器官捐贈者。

此外，計劃團隊亦建議慧玲的家人接受針對性的檢查，例如腎臟造影檢查，一方面嘗試尋找沒有致病基因的近親，期望提高覓得合適捐贈者的機會；另一方面，她的家族成員亦可了解自身的遺傳風險，及早監察身體狀況，進行疾病預測及防控。

慧玲勇敢面對生命的硬仗，在等待中尋找希望。有賴香港基因組中心專業團隊共同努力，透過嶄新的全基因組測序技術，讓慧玲最終可確定病因，得到個人化的精準治療，有效延緩病情惡化。

Learn About Polycystic Kidney Disease
認識多囊性腎病

Disease Incidence

Polycystic Kidney Disease (PKD) is the most common hereditary kidney disease, with Autosomal Dominant Polycystic Kidney Disease (ADPKD) accounting for approximately 90% of all cases. If either of the parents has the disease, their children have a 50% chance of inheriting it. Globally, one in every 400 to 2,500 individuals suffers from ADPKD. Based on Hong Kong's population of approximately 7.5 million, it is estimated that there are between 3,000 and 18,750 ADPKD patients.

發病率

「多囊性腎病」是最常見的遺傳性腎病，約90%病例屬於「常染色體顯性多囊性腎病」（ADPKD）。若父或母患病，其子女有50%機會遺傳此病。全球每400至2,500人中就有一人患有ADPKD。根據香港約750萬人口估算，約有3,000至18,750名ADPKD病人。

Symptoms

PKD is a condition characterised by the development of proliferative cysts in the kidneys, accompanied by high blood pressure and declining kidney function. PKD can affect both kidneys, and as the disease progresses, these cysts continue to grow, gradually replacing the normal kidneys and blood vessel tissues, leading to a deterioration in kidney function. Eventually, this can result in end-stage renal failure. PKD can also cause problems in other organs, such as the liver and pancreas. In some cases, it may even lead to bleeding in the brain, which can be fatal.

Currently, there is no cure for this rare disease; however, early diagnosis, coupled with effective blood pressure control, can significantly slow its progression. Medications are now available for clinical use that, when administered early, can help decelerate the growth of the cysts.

病徵

「多囊性腎病」又俗稱「泡泡腎」，病人腎臟會出現增生性囊腫，伴隨高血壓和腎功能衰退。此病可影響兩側腎臟，隨著囊腫不斷增大，更會逐漸取代正常的腎臟、血管組織，令腎功能逐漸衰退，最終導致末期腎衰竭。這個病亦會影響肝臟、胰臟等，更有機會引發腦出血，有致死風險。

這個罕見病目前尚無治癒方法，但及早診斷，加上有效控制血壓，可減慢病情惡化。現時已有藥物作臨床應用，如病人及早服用，有助減慢囊腫增長。

Case 個案 4

The Strength of a Mother Overcoming Breast Cancer for Her Children

為母則強 為子女奮戰癌魔

Love is the most powerful force in life. As she gazed at her children, Wing-sze (a pseudonym) was determined to defeat cancer for their sake, bravely embracing the necessary choices for their future, even if it meant she might never give birth again. A happy mother of two young children approaching her 40, she never expected that breast cancer would come uninvited during the prime of her life, forever altering her life. Wing-sze found the resilience to face the challenge head-on for her children. She joined the Hong Kong Genome Project (HKGP) and benefitted from whole genome sequencing (WGS) to identify disease-causing genetic variations. Upon learning that she was at risk of other cancers, she made the difficult but necessary decision to undergo surgery, removing her fallopian tubes and ovaries. This proactive measure allowed her to avoid future cancer risks and regain control of her health.

At the same time, WGS provided Wing-sze with valuable insights into the hereditary risks associated with her cancer, and she is now doing everything she can to protect her children from the threat she faces head-on.

愛，是人生最強大的後盾。穎詩（化名）凝視著子女的那一刻，已下定決心要為家人戰勝癌魔，即使以後無法生育，她亦勇於抉擇取捨。即將步入四十歲，擁有一對年輕子女的穎詩是一名幸福媽媽。她從沒想過，竟在人生的黃金階段遇上不速之客—乳癌，從此改寫了她的人生。為了孩子，穎詩積極面對，參加了香港基因組計劃，並透過全基因組測序精準地找到致病的變異基因。當她知道自己有患上其他癌症的風險時，便果斷地接受了輸卵管及卵巢切除手術，預防癌魔再襲，重拾健康主導權。

同時，穎詩亦了解到自己罹患的癌症的遺傳風險，盡其所能保護子女遠離威脅。

With no family history of cancer, Wing-sze never imagined she would face such a serious threat to her health. At age 39, cancer came knocking, throwing her life into turmoil. She was diagnosed with Invasive Ductal Carcinoma, an aggressive type of breast cancer in which cancer cells penetrate from the milk ducts into surrounding fatty tissue. It required prompt treatment to prevent the cancer spreading through her lymphatic system and blood vessels.

Understanding Unveils Precise Prevention and Treatment

The diagnosis came as a shock, but Wing-sze quickly found her footing and resolve. She decided to stay strong for her family, determined to witness her children's growth and to be there for their milestones. Alongside actively pursuing treatment, Wing-sze delved into her condition and its hereditary risks, hoping that this knowledge might help her children take preventive measures early on for a healthy future.

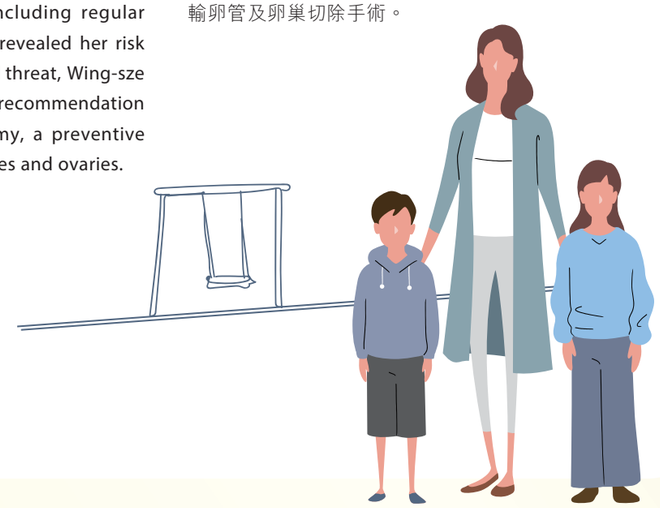
In 2022, just a year after being diagnosed with breast cancer, Wing-sze joined HKGP upon her attending doctor's referral. Through WGS and subsequent analysis, the HKGP team pinpointed a disease-causing genetic mutation related to breast cancer in her *RAD51C* gene. The result precisely explained the cause of her illness, allowing healthcare professionals to better understand her condition and develop a personalised treatment and management plan, including regular disease monitoring. The analysis also revealed her risk of ovarian cancer. Facing this potential threat, Wing-sze made the bold decision to follow the recommendation and undergo a salpingo-oophorectomy, a preventive surgery that removed her fallopian tubes and ovaries.

由於沒有家族癌症病史，穎詩從未想過自己會面臨如此嚴峻的疾病威脅，直至癌魔找上門，打亂了她的人生計劃。39歲那年，穎詩被證實患上「侵襲性乳管癌」（Invasive Ductal Carcinoma），癌細胞從乳管穿透管壁，侵入至四周的脂肪組織，她必須及時接受治療，以防癌細胞進一步通過淋巴系統和血管擴散至身體其他部位。

透徹了解病症 揭示精準防治

面對癌症，大部份人都無法坦然面對，穎詩亦不例外；然而，震驚過後，她深明為了家人，為了見證子女的成長和人生重要時刻，必須振作起來。因此，除了積極接受治療，穎詩亦努力深入了解這個病症和遺傳予下一代的風險，期望子女可及早預防，健康成長。

2022年，即確診乳癌翌年，穎詩在主診醫生轉介下參加了香港基因組計劃。透過全基因組測序和分析，計劃團隊發現她的「*RAD51C*基因」帶有乳癌相關的致病性基因變異。測序結果精準地解釋了穎詩患病的原因，讓醫護專業人員更透徹了解她的病況，為她制定針對性的乳癌治療和管理方案，包括定期監測病情。此外，分析結果亦顯示她有罹患卵巢癌的風險。面對未知的癌症威脅，穎詩果敢地決定接受計劃團隊建議，進行預防性輸卵管及卵巢切除手術。



Stepping Beyond the Unknown and Regaining Control of Health

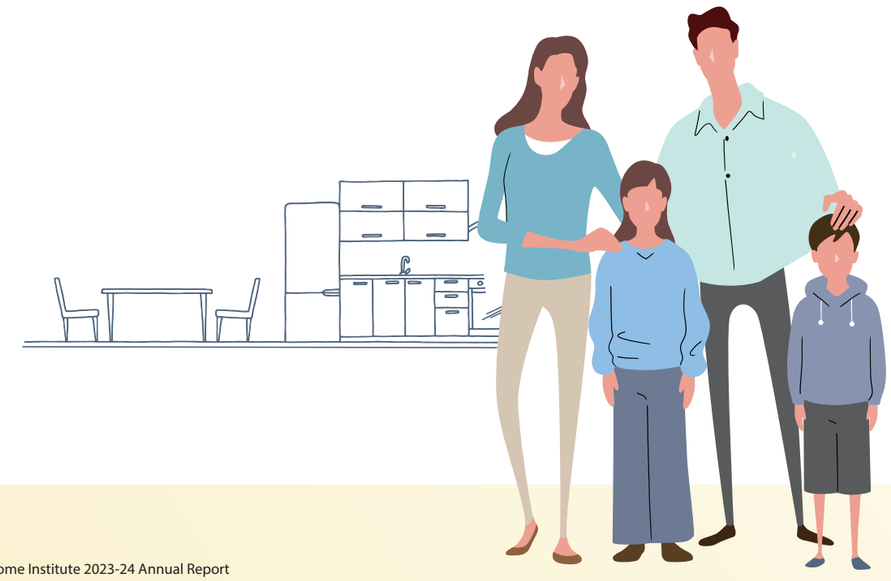
As the WGS results showed the hereditary nature of the genetic mutation, the HKGP team advised Wing-sze's daughter to take genetic testing once she reaches adulthood for early detection and prevention. They also recommended her son receive genetic counselling in the future to understand the hereditary risks and make informed family planning decisions.

A mother's strength knows no bounds. Despite facing the changes and impacts of cancer, this young mother fearlessly confronted the disease for the sake of her children and family. Through WGS, she regained control over her health and helped her family to understand their own health risks, allowing for more precise and effective disease prevention and management. With her positive attitude and courageous determination, Wing-sze has transformed her life story, ensuring her family can continue to live with love and happiness.

走出未知 重拾健康主導權

由於測序結果顯示相關的基因變異具有遺傳性，計劃團隊建議穎詩的女兒在成年後接受針對性的檢查，及早診斷和預防患病；而她的兒子未來亦可接受遺傳輔導，了解有關的遺傳風險，以助他有更好的家庭規劃。

為母則剛，這位年輕母親雖要奮力面對癌症所帶來的變化和影響，為著子女和家人，她無懼與癌魔一搏，透過全基因組測序的分析結果，重新掌握健康的主導權，並讓家人認識到自身的患病風險，從而更精準有效地預防管理。穎詩以她的正面積極、果斷勇敢，為自己的生命故事重新定調，亦讓一家人延續愛與幸福。



Learn About Invasive Ductal Carcinoma
認識侵襲性乳癌

Disease Incidence

Breast cancer has been the most common cancer among women in Hong Kong since 1994. It is estimated that one in every 13 women will face this disease in their lifetime, with Invasive Ductal Carcinoma (IDC) being the most common type, accounting for approximately 80% of all breast cancer cases. Early-stage breast cancer may develop without symptoms, and some patients are unaware of any changes in their breasts until a screening reveals the presence of cancer.

Research indicates that about 5-10% of breast cancer cases may be hereditary. Women with breast cancer gene mutations are 10 times more likely to develop breast cancer than the general population. As a result, understanding the hereditary risks of breast cancer and getting regular breast screenings can help in the early detection of hereditary breast cancer and increase the chances of a successful treatment.

發病率

乳癌自1994年起已成為香港女性中最常見的癌症，預計每13名女性就有一人在其一生中罹患此病。其中，「侵襲性乳癌」是香港女性最常見的乳癌，約佔所有病例的80%。早期乳癌可能毫無徵狀，有些病人從沒有察覺乳房有任何改變，直至接受乳房檢查才發現患上乳癌。

有研究顯示，約有5-10%的乳癌個案可能與遺傳有關。有乳癌基因變異的女性，罹患乳癌的機會比一般人高10倍。因此，了解乳癌的遺傳風險並定期進行篩查，有助於及早發現遺傳性乳癌，並增加治癒機會。

Symptoms

Current breast cancer treatments include surgery, radiotherapy, chemotherapy, hormone therapy, and immunotherapy. Patients with early-stage IDC typically undergo breast-conserving surgery, followed by radiation therapy or surgical removal of the entire breast. These treatments are effective, although some patients may require post-operative adjuvant chemotherapy or hormone therapy to prevent recurrence.

病徵

現時治療乳癌的方法包括外科手術、放射治療、化療、荷爾蒙療法、免疫治療等。「侵襲性乳癌」的早期病人通常會接受乳房保留手術及放射線治療，或乳房全切除手術，這些方法相當有效，部份病人則需接受術後輔助性化學治療或荷爾蒙治療，以預防復發。

Case 個案 5

Parents' Devotion Guides Their Angel through Adversity

父母以愛相伴 小天使逆境前行

Every child is a gift. For Ka-chun and Wing-sze (pseudonyms), the birth of their son was like an angel blessing their lives – a precious gift that they cherished dearly. As their son Chi-hin (a pseudonym) grew, the couple began to notice that his movements were not as agile as those of other children his age, and his speech development was somewhat delayed. They took Chi-hin to various doctors, yet even after years of consultation none could make a diagnosis. It was not until Chi-hin joined the Hong Kong Genome Project (HKGP) that the medical team, through whole genome sequencing (WGS), finally diagnosed him with Duchenne Muscular Dystrophy (DMD), a rare genetic disease. Confronted with life's uncertainties, Ka-chun and Wing-sze realised the importance of cherishing every moment. They wholeheartedly devoted themselves to Chi-hin's rehabilitation, joined a patient group, and with the encouragement and support of others in the same plight, they and their little angel faced this rare disease together, moving forward with determination and hope through adversity.

有人說，每個孩子都是天使。對家俊與詠思（化名）來說，兒子的出生便如天使下凡，是他們捧在手心的寶貝。隨著兒子梓軒（化名）日漸成長，兩人開始發現兒子日常走動不如同齡孩子般靈活，言語發展亦較遲緩。他們帶著梓軒四處求醫，輾轉數年仍然找不到病因；直至梓軒參加了香港基因組計劃，醫護團隊透過全基因組測序，終證實他患上罕見遺傳病「杜興氏肌肉萎縮症」（Duchenne Muscular Dystrophy）。面對世事無常，家俊與詠思明白珍惜當下的重要，盡心盡力陪伴梓軒進行復康訓練，並加入病友組織，與同路人相互扶持，以愛築路，帶領著梓軒在逆境中勇敢前行。

Chi-hin grew up in a loving and nurturing environment as the first child in the family. His first smile and cry were the most precious gifts for Ka-chun and Wing-sze. However, when Chi-hin took his first steps, his parents noticed a slight lack of co-ordination in his movements. His lower limbs appeared weak, particularly when he was playing in the park, leading to frequent falls. At the same time, Chi-hin's speech development was found to be delayed compared to his peers. Ka-chun and Wing-sze took Chi-hin through a series of medical consultations and tests, including genetic tests, yet they were only able to determine that he had a neuromuscular condition, without reaching a definitive diagnosis. When Chi-hin eventually turned five in 2022, his attending doctor referred him to HKGP. Following WGS and analysis, the HKGP team confirmed that Chi-hin has the rare disease DMD.

In the early stages of DMD, symptoms start with the deterioration of hip and thigh muscles, which makes standing and stair climbing challenging. As time progresses, muscle wasting continues, often resulting in significant mobility limitations and the need for a wheelchair by adolescence. Moreover, hand muscles may also gradually decline in function, affecting self-care abilities and potentially leading to life-threatening complications. Typically, patients with DMD have an average life expectancy of 20 to 30 years.

Whole Genome Sequencing Illuminates the Path Forward

Upon learning the details of their son's condition, Ka-chun and Wing-sze inevitably felt a profound heaviness in their hearts. They are now faced with a daunting reality: frequent hospital visits and ongoing treatments for Chi-hin, and the emotional preparation for his condition worsening over time. Most heart-wrenching is the possibility that they might outlive their son. Despite the immense pressure, the sight of Chi-hin's innocent joy fortifies their resolve. Ka-chun and Wing-sze know they must stay strong for him. They work hard to adjust their mindset, standing by Chi-hin's side as they confront this monumental challenge together, cherishing every single day.

梓軒是家中第一個孩子，在充滿愛與關懷的環境中成長。他的第一個微笑，或是第一聲哭鬧，都是家俊與詠思最寶貴的禮物。然而，當梓軒開始蹣跚學步時，二人注意到孩子有點手腳不協調，在公園玩耍時，總是下肢無力，經常跌倒；而他在言語發展評估中，亦被評為發展較同齡兒童遲緩，二人遂帶著梓軒到處求診。數年間，雖然做遍不同檢查，包括遺傳疾病基因檢測，卻仍然無法斷症，只知道梓軒的情況與神經肌肉疾病有關。直至梓軒五歲時，即2022年，主診醫生轉介他參加香港基因組計劃，經全基因組測序分析後，計劃團隊終證實他患上罕見病「杜興氏肌肉萎縮症」。

此病初期病徵是臀部和大腿肌肉開始退化，導致病人站立和行樓梯變得困難。隨著年齡增長，肌肉會不斷萎縮；及至青春期開始，病人往往出現行動不便，需要依賴輪椅代步。同時，病人手部的肌肉活動能力亦會逐漸退化，因而影響自理能力，甚至可能出現致命的併發症。一般來說，此病病人的平均壽命大約為20至30歲。

全基因組測序 為未來覓得方向

家俊與詠思得知詳情後，內心難免沉重。他們除了要作好準備，照顧梓軒日常生活起居，不斷進出醫院和接受治療外，還要面對兒子病情將日漸惡化，甚或比他們更早離世的可能。縱然面對巨大壓力，看著天真爛漫的梓軒，家俊與詠思明白他們必須振作，於是努力調整心態，重排生活優次，決心堅強面對，珍惜每一天。

Based on the WGS results and clinical data, the HKGP team concluded that existing medications had limited efficacy for Chi-hin's condition. For this reason, they developed a personalised rehabilitation-focused treatment plan aimed at slowing the progression of his condition, enhancing his quality of life, and strengthening his self-care abilities. With the HKGP team's support, Ka-chun and Wing-sze devote themselves wholeheartedly to Chi-hin's exercise and rehabilitation training. Their efforts are rewarded by Chi-hin's determined smile, a beacon of warmth in these challenging times, inspiring them to press forward hand in hand with their child.

Advancing Together and Contributing to Medical Research

Ka-chun and Wing-sze realised how important it is to connect with others in similar situations, leading them to join a DMD patient group. There, they met other families navigating similar challenges, and found a community of kindred spirits. Sharing experiences with others not only provided comfort but also inspired hope. Aware of the current limitations of medical technology in treating DMD, they have decided to contribute their genomic data to support medical research, in the hopes that it will pave the way for breakthroughs that will benefit future DMD patients.

在香港基因組計劃下，醫護團隊根據全基因組測序結果以及臨床數據，仔細分析後認為現有藥物對梓軒的情況來說，治療成效有限，因此轉而為梓軒制定以復康為主的個人化治療方案，以延緩病情、提升生活質素和自理能力為目標。有了團隊的協助，家俊與詠思積極陪伴兒子進行運動和復康訓練，梓軒臉上堅定的笑容，便是二人在困難中最溫暖的回報，讓他們繼續奮力向前，與病同行。

同路人守望前行 貢獻醫學研究

家俊與詠思深感同路人支持的重要，於是加入了「杜興氏肌肉萎縮症」病友組織，認識了面對相同挑戰的家庭，大家互助互勉，保持信念和盼望。二人亦深明現今醫學技術對治療這個病的限制，因此期望透過提供個人相關資料和數據，支持醫學研究，日後幫助更多同路人。

Learn About Duchenne Muscular Dystrophy
認識杜興氏肌肉萎縮症

Disease Incidence

Duchenne Muscular Dystrophy (DMD) is a rare hereditary condition characterised by progressive muscle wasting that primarily affects males, occurring in approximately one in every 3,500 to 5,000 male newborns. The disease is caused by a genetic abnormality that impedes the body from producing a crucial functional protein called "Dystrophin", leading to the decay and eventual demise of skeletal muscle cells.

發病率

「杜興氏肌肉萎縮症」是一種罕見遺傳性肌肉萎縮疾病，主要影響男性，發病率約為每3,500至5,000名新生男嬰中有一名病人。疾病起因是由於基因異常，阻止身體製造一種重要的功能性肌肉萎縮(Dystrophin)蛋白，導致骨骼肌肉細胞萎縮及壞死。

Symptoms

Patients experience limb weakness due to muscle degeneration. The calf muscles often become abnormally enlarged as muscle tissue is replaced by fibrotic tissue. Symptoms such as an unsteady gait and frequent falls, typically appear in patients aged three to seven. As patients age, muscle degeneration and atrophy progressively worsen. By the ages of 10 to 12, patients usually require the use of a wheelchair. Abnormal muscle function can lead to joint deformities and impact heart and lung function. In severe cases, it can trigger irregular heartbeat and heart failure, necessitating ventilator support for breathing.

There is no cure for DMD. Treatment primarily focuses on physiotherapy and rehabilitation, which aim at strengthening muscles and preventing muscle tightening. These interventions seek to improve the quality of life for patients and slow the progression of the condition.

病徵

病人因肌肉病變而四肢無力，小腿肌肉通常會被硬化組織取代而異常腫大。病徵通常在病人三至七歲出現，如走路蹣跚和頻繁跌倒，隨著年齡增長，病人的肌肉病變和萎縮也變得越來越嚴重。10至12歲時通常需要坐輪椅。肌肉的異常活動能力也導致關節畸形，並影響心肺功能，嚴重情況下可引發心律不正和心臟衰竭，或需依賴呼吸器輔助呼吸。

此罕見病目前尚未有方法根治，治療以物理治療復健為主，透過增強肌肉力量和預防攣縮，以改善病人生活質素並延緩病情進展。