香港罕見疾病聯盟
RARE DISEASE HONG KONG

2025 ANNUAL REPORT
會務報告

因罕而聚，以愛同行

機構使命：尊重差異・確保權利

核心業務：政策倡導・提升認知・能力建設

發展策略：廣結網絡・共謀協作

管理哲學：善用資源・成效為本

管治原則：開放透明・誠信問責

Our mission:
To respect differences & ensure rights

What we do:
Policy advocacy, public awareness & capacity building

Development strategy:
To maintain and enhance networks and collaborative partnerships

Management philosophy:
Optimal resource utilisation & outcome-oriented approach

Governance principles:
Openness, transparency, integrity & accountability

香港罕見疾病聯盟有限公司
2025 年會務報告
(2026 年 2 月)

Rare Disease Hong Kong Limited
Annual Report 2025
(February 2026)

香港罕見疾病聯盟 (下稱「罕盟」) 在十周年的根基上始終秉持初心，繼續深耕罕見病權益。2025 年，罕盟在政策倡導與醫療優化上實現多項突破，不僅成功推動藥物資助範圍擴張，更透過發布指標性的研究報告，為社群需求提供堅實的數據實證。

過去一年，罕盟致力連結政府、醫療體系及社會各界，除落實多項人性化照護與跨專科機制外，亦透過公眾教育及境外交流，深度展現病友面對挑戰的堅毅，並凝聚醫患間的信任與連繫。罕盟將持續監察政策落實，優化診療路徑，為罕病群體共創具包容性的社會環境。

Building upon its decade-long foundation, Rare Disease Hong Kong (RDHK) remains steadfast in its mission to champion rare disease rights. In 2025, RDHK achieved breakthroughs in advocacy and healthcare, notably expanding drug subsidies and publishing landmark research to provide solid, data-driven evidence for community needs.

Over the past year, RDHK bridged the government, medical sector, and society to implement human-centric care and multidisciplinary mechanisms. Through education and exchanges, RDHK showcased patient resilience and strengthened clinician-patient trust. RDHK will continue monitoring policy and optimizing clinical pathways to co-create an inclusive environment for the rare disease community.

1. 政策倡議

1.1 推動罕病政策落實

罕盟與醫院管理局 (醫管局) 延續高效的恆常溝通，分別於 2025 年 4 月及 9 月會面，就多項核心政策達成實質共識，為罕病社群勾勒出更完善的醫療藍圖。

a. **脊髓肌肉萎縮症 (SMA) 藥物資助範圍擴張：**醫管局正式落實放寬脊髓肌肉萎縮症 (SMA) 成人口服藥的資助範圍，並持續由專家小組審視其他治療藥物的臨床數據，保障 SMA 成人患者的用藥選擇權。

b. **跨專科門診與社區化支援：**九龍中聯網試行成人跨專科一站式門診，首階段涵蓋結節性硬化症。局方亦積極研究將抽血等程序下放至普通科門診，目標 2026 年底前達成全港聯網通用，減輕患者跨區覆診之苦。

c. **特醫食品管理與資助銜接：**醫管局已完成逾百項特醫食品評審，並計劃發布首份名冊，加強管理並指引臨床使用。

d. **人性化照護與探訪安排：**局方認同照顧者對失去自理能力患者的重要性，將研究更具彈性的陪護方案。

e. **臨床試驗資訊通報機制：**醫管局透過「中央臨床研究及創新辦公室」與病友組織建立聯繫，分享罕病相關的臨床測試招募資訊。旨在增加醫療資訊透明度，讓合適的患者能及早接觸前沿藥物與治療機會。

1. Policy advocacy

1.1 Promote the implementation of rare disease policies

RDHK maintained productive dialogue with the Hospital Authority (HA) through meetings in April and September 2025, reaching consensus on several core policies to enhance the medical blueprint for the rare disease community.

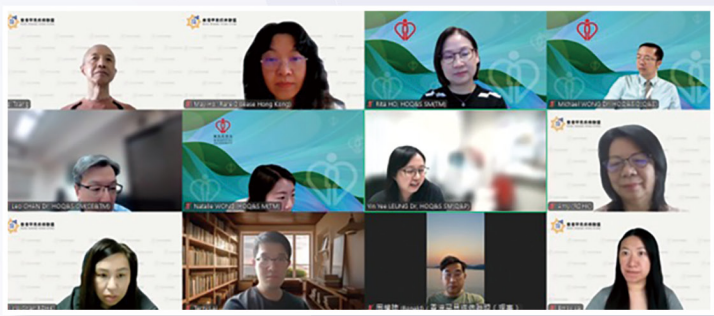
a. **Expanded SMA Drug Subsidy:** HA officially expanded subsidy coverage for adult oral medications for Spinal Muscular Atrophy (SMA), with expert panels continuously reviewing clinical data of other therapies to ensure treatment options for adult SMA patients.

b. **Multidisciplinary & Community Support:** A one-stop adult multidisciplinary clinic was piloted in the KCO Cluster, initially covering Tuberous Sclerosis Complex. HA is also decentralizing procedures like blood tests to primary clinics, aiming for city-wide implementation by late 2026.

c. **Medical Food Management:** Following reviews of over 100 specialized medical food items, HA plans to publish its first formulary to strengthen clinical guidelines and management.

d. **Human-Centric Care:** Recognizing the vital role of caregivers for patients with limited self-care ability, HA is exploring more flexible accompaniment and visitation schemes.

e. **Clinical Trial Information Sharing:** Through the Central Clinical Research and Innovation Office, HA now shares rare disease trial recruitment data with patient groups, increasing transparency and access to cutting-edge treatments.



1.2 就《施政報告》公眾諮詢提交意見書

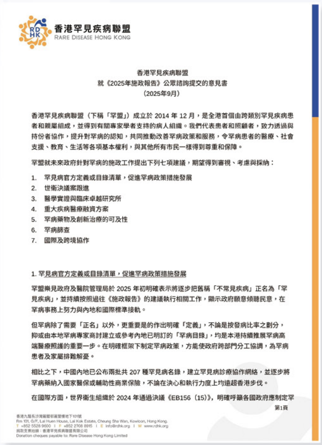
罕盟就《2025 年施政報告》提交公眾諮詢意見書，圍繞罕病群體的長遠發展提出七大核心建議。促請政府正視患者在診療路徑、藥物資助及社會福利保障等方面的實際訴求，致力倡導更完善的精準醫療與支援政策。

a. 罕見病官方定義或目錄清單，促進罕病政策措施發展
b. 世衛決議案跟進
c. 醫學實證與臨床卓越研究所
d. 重大疾病醫療融資方案
e. 罕病藥物及創新治療的可及性
f. 罕病篩查
g. 國際及跨境協作

1.2 Submission on the 2025 Policy Address public consultation

RDHK submitted recommendations for ***the 2025 Policy Address***, outlining seven core proposals for the rare disease community. The submission urges the Government to address critical gaps in clinical pathways, drug subsidies, and social welfare, advocating for enhanced precision medicine and robust support policies:

a. **Official Definition or List of Rare Diseases** to facilitate policy development.
b. **Follow-up on WHO Resolutions** regarding rare diseases.
c. **Institute for Medical Evidence and Clinical Excellence**.
d. **Healthcare Financing Schemes** for major diseases.
e. **Accessibility of Rare Disease Drugs and Innovative Therapies**.
f. **Rare Disease Screening** and early diagnosis.
g. **International and Cross-Border Collaboration**.


香港罕見疾病聯盟
RARE DISEASE HONG KONG

香港罕見疾病聯盟
就《2025年施政報告》公眾諮詢提交的意見書
（2025年9月）

香港罕見疾病聯盟（下稱「罕盟」）成立於 2014 年 12 月，是全港首個由跨類別罕見疾病患者和親屬組成，並得到有關專家學者支持的病人組織。我們代表患者和照顧者，致力透過與持份者協作，提升對罕病的認知，共同推動改善罕病政策和服務，令罕病患者的醫療、社會支援、教育、生活等各項基本權利，與其他所有市民一樣得到尊重和保障。

罕盟就未來政府針對罕病的施政工作提出下列七項建議，期望得到審視、考慮與採納：

1. 罕見病官方定義或目錄清單，促進罕病政策措施發展
2. 世衛決議案跟進
3. 醫學實證與臨床卓越研究所
4. 重大疾病醫療融資方案
5. 罕病藥物及創新治療的可及性
6. 罕病篩查
7. 國際及跨境協作

1. 罕見病官方定義或目錄清單，促進罕病政策措施發展

罕盟樂見政府及醫院管理局於 2025 年初明確表示將逐步把舊稱「不常見疾病」正名為「罕見疾病」，並持續按照過往《施政報告》的建議執行相關工作，顯示政府願意傾聽民意，在罕病事務上努力與內地和國際標準接軌。

但罕病除了需要「正名」以外，更重要是的作出明確「定義」，不論是按發病比率之劃分，抑或由本地罕病專家商討建立或參考內地已明訂的「罕病目錄」，均是本港持續推展罕病高端醫療照護的重要一步。在明確框架下制定罕病政策，方能使政府跨部門分工協調，為罕病患者及家屬排難解憂。

相比之下，中國內地已公布兩批共 207 種罕見病名錄，建立罕見病診療協作網絡，並逐步將罕病藥納入國家醫保或輔助性商業保險，不論在決心和執行力度上均遠超香港步伐。

在國際方面，世界衛生組織於 2024 年通過決議《EB156（15）》，明確呼籲各國政府應制定罕

第1頁

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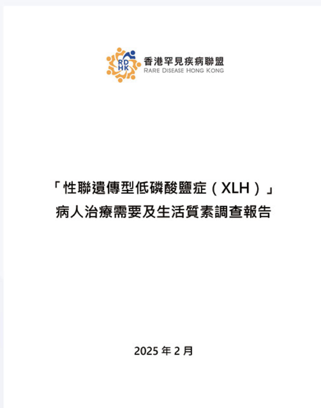
Donation cheques payable to: Rare Disease Hong Kong Limited

1.3 好「磷」社 XLH 病人治療需要及生活質素調查計劃

正式發布「好『磷』社」XLH 治療需要及生活質素調查結果。是次研究共收回 20 份有效問卷，對象包括 7 名成年及 5 名兒童患者，以及 8 名照顧者。透過量化數據與專科醫生、患者及家屬的深度分享，報告針對「發病至確診、治療副作用、情緒社交、照顧者身心狀況」四大範疇進行剖析，揭示患者家庭面對的困難。調查結果不僅反映社群訴求，更為優化診療與政策提供實證。

1.3 Survey on the treatment needs and quality of life of XLH patients

RDHK released survey results on treatment needs and quality of life for X-linked hypophosphatemia (XLH) patients. The study collected 20 valid responses from 7 adults, 5 pediatric patients, and 8 caregivers. Combining quantitative data with in-depth interviews, the report analyzes four key areas: diagnosis journey, side effects, social well-being, and caregiver stress. These findings reveal the challenges families face and provide data-driven evidence to optimize clinical care and policy.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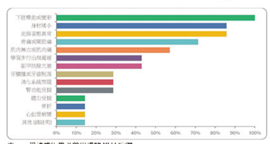
RARE DISEASE HONG KONG

「性聯遺傳型低磷酸鹽症（XLH）」
病人治療需要及生活質素調查報告

2025 年 2 月



表一：受訪成年患者曾出現的 XLH 病徵

至於對受訪成年患者的日常生活影響最大的病徵，逾七成人直言是「骨痛或關節痛（71.4%）」，其次是「肌肉無力或肌肉痛」、「下肢彎曲或變形」及「身材矮小」，分別佔 57.1%。

是次調查亦詳細了解患者骨痛或關節痛的情況，發現許多受訪者因骨痛或關節痛問題而生活質素備受影響。在受訪前的一星期，接近四成三受訪患者（42.9%）「連續 7 天」都有骨痛或關節痛，疼痛「3 至 4 天」或「1 至 2 天」者各佔 28.6%。在受訪前的一個月，逾五成七患者（57.1%）曾因骨痛或關節痛而失眠；約四成三人（42.9%）曾因疼痛而告病假；一成四人（14.3%）則曾因這問題而要取消社交活動。

骨科專科鄺宇翎醫生接受訪問時表示，骨骼成長在出生後首十多年最關鍵，體內需有正常水平的磷酸鹽及鈣質，若磷及鈣的水平失去平衡，會影響骨骼生長。因此，持續出現磷酸鹽流失的 XLH 患者，普遍較健康人士矮小。

XLH 病人治療需要及生活質素調查報告 P.8

1.4 脊髓肌肉萎縮症 (SMA) 成人患者用藥安排

經過多年不懈的耕耘與倡議，脊髓肌肉萎縮症 (SMA) 的藥物政策於本年度迎來重要進展，罕盟成功爭取 25 歲或以上的成年患者納入藥物治療保障範圍。並於 2025 年 12 月，聯同香港肌健協會及癌症策略關注組與醫院管理局代表會面，就優化成人患者的藥物可及性達成共識。局方於會上積極回應社群訴求，明確了用藥銜接機制：在符合臨床理據的前提下，已用藥患者可申請轉藥一次。此項機制不僅為患者提供了更靈活的治療規劃，亦是落實「公平用藥」原則的關鍵里程碑。罕盟將持續透過反饋真實個案數據，協助優化銜接流程，與醫管局並肩完善治療保障，確保 SMA 患者的醫療權利能與時並進。

1.4 Medication Arrangements for Adult SMA Patients

Following years of persistent advocacy, a breakthrough in Spinal Muscular Atrophy (SMA) policy successfully extended treatment coverage to adults aged 25 or above. In December 2025, RDHK joined HKNMDA and the Cancer Strategy Concern Group to reach a consensus with the Hospital Authority (HA) on enhancing drug accessibility.

HA established a proactive transition mechanism: patients may apply for a one-time medication switch based on clinical evidence. This milestone enables flexible treatment planning and upholds the principle of equitable access. RDHK will continue sharing real-world case data to refine these processes, ensuring SMA care evolves alongside medical advancements.

1.5 更新香港罕病及罕癌藥物清單

為持續提供最新的藥物資訊，罕盟於 2025 年 5 月更新《香港罕見疾病藥物參考清單》及《香港罕見癌症藥物參考清單》，並上載至罕盟網頁 (https://rdhk.org/others)，方便持份者查閱和參考。

1.5 Updating the drug lists for rare diseases and rare cancers in Hong Kong

To provide the latest drug information, RDHK updated the "Drug List for Rare Diseases in Hong Kong" and "Drug List for Rare Cancers in Hong Kong" in May 2025. These are available on the RDHK website (https://rdhk.org/others) for stakeholders' easy access and reference.

請掃瞄二維碼，以查閱及下載
《香港罕見疾病藥物參考清單》
《香港罕見癌症藥物參考清單》

2. 提升認知

2.1 罕病公眾教育項目

罕盟致力開拓全方位的宣傳渠道，藉此拓展大眾對罕見病的認知邊界。本年度，我們透過專題訪問、專題講座、製作小冊子及短片、更新專題網頁、社交媒體推廣、病友聚會等活動，為杜興氏肌肉營養不良症 (DMD)、脊髓肌肉萎縮症 (SMA)、地中海貧血症、法布瑞氏症 (Fabry)、性聯遺傳型低磷酸鹽佝僂症 (XLH)、特發性多發性卡斯爾曼氏症 (iMCD) 及希佩爾 - 林道病 (VHL) 等病類爭取社會關注。

「罕見病公眾教育計劃 2025」旨在突顯患者在逆境中的堅毅，喚起社會的理解與尊重。項目包括製作《超越您想像》短片系列，藉罕病孩童的純真視角映照生命力；同時透過《愛 · 不罕見感恩分享會》，由病友及家屬向醫護團隊致意，肯定其專業貢獻，藉此深化醫患間的信任與連繫。歡迎到罕盟網頁閱讀：
https://rdhk.org/post/show?id=950&mid=43

2. Public awareness

2.1 Promote the implementation of rare disease policies

RDHK leverages diverse channels to broaden public understanding of rare diseases. This year, through interviews, seminars, brochures, videos, and social media, we advocated for conditions including Duchenne Muscular Dystrophy (DMD), Spinal Muscular Atrophy (SMA), Thalassemia, Fabry, x-linked hypophosphatemia (XLH), Idiopathic Multicentric Castleman Disease (iMCD), and von Hippel-Lindau disease (VHL).

The *Rare Disease Awareness Campaign 2025* highlights patient resilience to foster social inclusion. Key initiatives included the *More than you can imagine* video series, showcasing life's vitality through children's perspectives, and the *Love is Not Rare* sharing session. The latter allowed patients and families to honor medical teams, deepening clinician-patient trust. Visit our website to learn more:
https://rdhk.org/post/show?id=950&mid=43

2.2 真人圖書館

罕盟藉著「真人圖書館」的親身對話，讓大眾在故事分享中建立對罕見病的同理心。2025 年，罕盟連同罕病患者和照顧者走進 9 間不同的機構，透過講座與工作坊形式，向中小學生、醫護學生及社群成員等 1,227 位參與者，分享了 8 種罕見疾病的生命故事。這些交流，讓不同年齡和界別的受眾，從中體會如何將平等尊重與共融的意識，轉化為日常實踐，從而為罕病社群構建一個更具友善溫度的社會環境。

2.2 Human library

Through the "Human Library" project, RDHK fosters empathy for the rare disease community through personal dialogue. In 2025, patients and caregivers visited nine institutions, sharing stories of eight different rare diseases with 1,227 participants, including students and healthcare professionals. These exchanges empower audiences to translate respect and inclusion into daily practice, fostering a more compassionate and supportive environment for the rare disease community.

2.3 網頁

2025 年，罕盟持續優化社群連結，透過精準內容導引，將官方網站打造成為罕病患者與大眾獲取實用資訊的關鍵平台。本年度獨立瀏覽人數達 145,978，總瀏覽次數為 211,253，頁面瀏覽量更創下 469,141 次新高。

2.3 Website

In 2025, RDHK continued to optimize community engagement by transforming its official website into a vital information hub for patients and the public. This year, the platform reached a new high of 469,141 page views, with 145,978 unique visitors and 211,253 total sessions.

2.4 社交媒體平台

罕盟積極經營多元社交媒體平台，成效顯著，讓罕病資訊傳播至更廣泛的受眾。過去一年，在臉書 (Facebook) 共發布 194 則貼文，憑藉強大的社群傳播力，總觸及人數提升至 3,477,257 人，追蹤者亦成長至 7,433 人。Instagram 則發布了 162 則貼文，觸及人數達 2,476,249 人，追蹤人數亦穩定增至 1,474 人。此外，在 LinkedIn 平台發布了 164 則貼文，總曝光次數達 44,292 次，關注人數增至 696 人。這些數字反映罕盟在社交媒體平台影響力的持續擴張，有助於進一步凝聚社會大眾對罕病的關注。

2.4 Social media platforms

RDHK's active presence across multiple social media platforms has significantly expanded the reach of rare disease information. Over the past year, our Facebook reach surged to 3,477,257 across 194 posts, with followers growing to 7,433. On Instagram, 162 posts reached 2,476,249 individuals, while followers increased to 1,474. Additionally, LinkedIn generated 44,292 impressions from 164 posts, with followers rising to 696. These figures reflect RDHK's growing digital influence and its success in mobilizing public attention for rare diseases.

2.5 罕情

罕盟致力透過多元化渠道，構建透明且高效的資訊平台。而《罕情》季刊正是重要的渠道之一。於 1、4、7 及 10 月出版的《罕情》全方位報導國內外罕病政策、組織動向及醫藥科技等前瞻資訊。為響應數位化趨勢，《罕情》自 2025 年 4 月起轉為全面線上發布，讓醫護人員、罕病社群、各持份者及公眾人士能更便捷地閱覽。透過靈活的網絡傳播，我們期望進一步凝聚社會對罕病群體的關切，持續發揮資訊橋樑的作用。歡迎隨時於罕盟網頁瀏覽及下載：https://rdhk.org/RareCare

2.5 RareCare

RDHK remains committed to building a transparent and efficient information platform through diverse channels. The quarterly publication, *RareCare* (published in January, April, July, and October), provides comprehensive coverage of rare disease policies, organizational news, and medical advancements. To embrace digitalization, *RareCare* transitioned to a fully online format in April 2025, offering stakeholders and the public more convenient access. By leveraging digital distribution, this initiative strengthens social awareness and continues to serve as a vital information bridge. Read and download at: https://rdhk.org/RareCare

3. 能力建設

3.1 社交及消閒活動

促進社群成員間的交流與互助，是罕盟舉辦活動的核心目標。2025 年，罕盟透過 7 場形式多樣的活動，為大家提供建立連繫與舒展身心的空間，包括：罕在一起・動手迎新春、鮮花貼畫工作坊、紙境・心聲工作坊、竹編扇子 DIY、流沙畫工作坊、精神健康講座及藝術治療工作坊，以及第 3 屆全港傷健福音日營。同時，罕盟亦協助會員獲取不同類型的門票，包括盧冠廷紅館演唱會、香港書展、電影《無名指》、國際薩克斯管工作坊音樂會及港樂「星夜・交響曲」，藉此增加會員參與社交活動的機會。全年參加人數超過 420 人，反映會員對社群連繫及豐富社交生活的積極支持。

3. Capacity building

3.1 Social and leisure activities

Fostering mutual support and connection remains the core objective of RDHK's community activities. In 2025, RDHK organized 7 diverse events to provide spaces for relaxation and connection, including Lunar New Year workshop, Flower art workshop, Paper art workshop, Bamboo fan workshop, Sand art workshop, Mental health and art therapy seminar, and the 3rd Gospel Day Camp for the disabled.

Additionally, RDHK facilitated access to various cultural events, such as Lo Kwun Ting's Hong Kong Coliseum concert, the Hong Kong Book Fair, the film *My First of May*, international saxophone concerts, and the HK Phil's *Swire Symphony Under the Stars*, broadening opportunities for social participation. With over 420 participants throughout the year, these initiatives reflect members' active desire for community connection and a vibrant social life.

3.2 義工培訓

罕盟在 2025 年為 4 名港大碩士生及 1 名中大學生提供實習機會，合計完成逾 900 小時實習。實習生更深入參與個案支援，協助脊髓肌肉萎縮症 (SMA) 及希佩爾 - 林道病 (VHL) 患者向醫療單位查詢用藥安排。

在實習生與義工團隊的鼎力支持下，全年共協助 86 場次罕盟活動，總參與人次達 245 人，展現高效的支援能量。此外，透過持續運作「罕盟醫科」群組，罕病資訊得以在準醫護群體中有效傳遞，確保社群訴求在未來醫療專業領域中獲得長遠關注。

3.2 Volunteer training

In 2025, RDHK provided internships for 4 HKU and 1 CUHK medical students, totaling over 900 services hours. Interns engaged deeply in case advocacy, assisting SMA and VHL patients with medication inquiries. The intern and volunteer team participated in 86 activity sessions throughout the year, with a total of 245 participants involved in these support efforts. Furthermore, through the ongoing *RDHK Medical* WhatsApp group, rare disease insights are effectively shared with future healthcare professionals, ensuring the community's voice gains long-term attention within the medical field

3.3 本地交流活動

罕盟積極連結本地及海內外專家，透過多元對話提升罕病支援的廣度。罕盟代理會長受邀擔任由香港基因組中心、國際罕見疾病聯盟 (RDI) 及 RDI - Lancet Commission 共同主辦之「國際基因醫學研討會」主禮嘉賓。是次盛會匯聚全球專家，探討基因醫學從早期篩查到個人化治療的突破。會中專家強調「以人為本」的科研初心，主張科學成果應轉化為解決患者迫切需求的臨床方案，這份理念為罕盟籌備中的「世界罕病日研討會 2026」提供了關鍵啟發。

此外，罕盟亦接待北京兒童醫院罕見病中心代表團，探討創新科技與教育經驗。代表團分享了利用 AI 遙距技術掌握患者狀況的成果，為跨境醫療交流提供借鑑；同時對「真人圖書館」表示認同，期望將此教育模式引入內地校園。透過參觀香港復康會設施，雙方對「助人自助」理念深感共鳴，為未來跨地域合作奠定基礎。

3.3 Exchange activities in Hong Kong

RDHK actively engages with local and international experts to broaden the scope of rare disease support. The Acting Chairman of RDHK was invited as an officiating guest at the *International Conference on Genomic Medicine*, co-organized by the Hong Kong Genome Centre, Rare Diseases International (RDI), and the RDI–Lancet Commission. This global summit explored breakthroughs from early screening to personalized medicine. Experts emphasized a patient-centered approach, advocating for the translation of scientific research into clinical solutions that address urgent patient needs—a philosophy that serves as key inspiration for the upcoming *World Rare Disease Day Symposium 2026*.

Additionally, RDHK hosted a delegation from the Rare Disease Center of Beijing Children's Hospital to discuss innovation and education. The delegation shared achievements in using AI remote technology for patient monitoring, providing a valuable reference for cross-border medical exchange. They also expressed strong interest in the Human Library model, envisioning its introduction to schools in Mainland China. A visit to the Hong Kong Society for Rehabilitation facilities further reinforced a shared commitment to the self-help and mutual aid philosophy, laying a foundation for future regional collaboration.

3.4 境外交流活動

2025 年，罕盟持續透過參與國際及內地交流，將本地病友的聲音帶往更廣闊的平台。罕盟會長代表本港遠赴瑞士，出席國際罕見疾病聯盟 (RDI) 於第 78 屆世界衛生大會期間舉辦的邊會 (WHA Side Event) 並發表專題演說，向全球分享香港的倡議經驗及對國際罕病決議案的期盼。除了政策層面的發聲，罕盟亦獲邀往杭州參與罕見病公益音樂會，會長向八百多名兩岸三地的嘉賓介紹罕盟理念；罕盟會員更獲邀擔任表演嘉賓，以才藝為盛會揭開序幕，展現生命韌力。

在人才培育和知識接軌方面，罕盟傳訊主任獲 RDI 贊助前往西班牙參與 EURORDIS 藥物研發培訓課程，與三十多位全球罕病領袖交流最前瞻的藥研實務。同時，理事何正琛先生成功入選 RDI 青年領袖計劃，致力於將世衛決議案落實為具體的本地執行項目。此外，罕盟亦資助香港好磷社協會領袖前往新竹參與性聯遺傳型低磷酸鹽佝僂症病友醫療座談，強化區域性的病友組織協作。這些多元化的國際參與，不僅提升了團隊的專業素養，更引進全球智慧，為優化本地罕病政策注入了源源不絕的動力。

3.4 Exchange activities outside Hong Kong

In 2025, RDHK amplified local patient voices on international and Mainland China platforms. The Chairman delivered a keynote at a *WHA Side Event* in Switzerland, sharing Hong Kong's advocacy experience and expectations for global resolutions. In Hangzhou, RDHK introduced its mission to over 800 guests at a charity concert, where members performed to showcase the community's resilience.

Capacity building also saw significant progress. The Communications Officer attended an EURORDIS drug research training in Spain, while Council Member, Sam Ho, joined the *RDI Young Leaders Program* to translate WHO resolutions into local action. Additionally, RDHK supported the XLH patient group leader's participation in a regional medical seminar in Hsinchu. These engagements bring global expertise to Hong Kong, driving the optimization of local rare disease policies.

4. 其他工作

4.1 周年大會

罕盟於 2025 年 6 月 14 日順利召開第六屆會員周年大會，共 76 人齊聚一堂。會上除通過各項議程，更補選兩位新成員加入第三屆理事會。今年亦增設新會員簡介短片，清晰說明權益與義務，助新朋友投入這個溫暖大家庭，並肩為罕病社群的未來編織新章。

4. Public awareness

4.1 Annual general meeting (AGM)

The 6th AGM was successfully held on June 14, 2025, with 76 participants in attendance. In addition to passing the agenda, two new members were elected to the 3rd Council. To improve member experience, a new introductory video was launched to clearly outline rights and responsibilities, helping new members integrate into the community and collaborate on future advocacy.

4.2 護理物資及服務資助先導計劃

罕盟積極推動「護理物資及服務資助計劃」，為正處於公共醫療與社福銜接期的患者提供適時支援。年度內共批出 6 宗申請，針對丙酮酸鹽脫氫酵素缺乏症、苯丙酮尿症、戊二酸血症第 1 型及表皮溶解水皰症患者，提供所需的特別奶粉或敷料資助。此舉旨在減輕患者的日常負擔，並透過收集實際個案經驗，協助推動更全面的罕病支援措施。

4.2 The Pilot Scheme of Subsidy for Healthcare Needs

RDHK actively promoted the *Pilot Scheme of Subsidy for Healthcare Needs*, providing timely support to patients navigating the gaps between public healthcare and social welfare systems. Throughout the year, six applications were approved for patients with Pyruvate Dehydrogenase Deficiency (PDH), Phenylketonuria (PKU), Glutaric Aciduria Type 1 (GA1), and Epidermolysis Bullosa (EB). The scheme provided essential subsidies for specialized formulas and medical dressings, aiming to alleviate daily burdens while gathering case data to advocate for more comprehensive rare disease support policies.

4.3 與其他病友組織交流

深化跨組織的連結是推動罕病權益的核心。2025 年，罕盟先後與 8 個本地及 4 個外地罕病組織交流，將合作視野拓展至境外。並特別舉辦《我們的足跡—罕病病友團體交流會》，邀請多位領袖分享營運組織的挑戰與展望，讓病友們集思廣益，互相學習。同時，罕盟亦積極走進不同社群活動，發放實用資訊，與各界夥伴攜手提昇罕病患者的福祉。

4.3 Interchange with other patient organisations

Deepening cross-organizational ties is central to advancing rare disease rights. In 2025, RDHK exchanged insights with eight local and four overseas organizations, expanding its collaborative reach beyond Hong Kong. A highlight was the Rare Disease Patient Group Leader Workshop, where leaders shared organizational challenges and visions to foster mutual learning. Additionally, RDHK actively participated in various community events to disseminate practical information and partner with diverse stakeholders to enhance patient well-being.

附錄一、會員及所涵蓋病種

截至 2025 年年底為止，會員總人數為 998，其中：

1. 普通會員（關注及支持罕病患者人士）
   數目：135
2. 基本會員（罕病患者或家屬）
   數目：863
   a) 個人會員數目：854
   b) 團體會員數目：9

Appendix I – Members and types of diseases

As at the end of 2025, the total number of members was 998, among which:

1. Number of ordinary members (those who care about and support rare disease patients): 135
2. Number of full members (rare disease patients or families): 863
   a) Number of individual members: 854
   b) Number of organisational members: 9

(按英文名稱排序)
(In alphabetical order)

- 牽手同行協會
  Hand in Hand Association
- 香港平滑肌肉瘤基金會
  Hong Kong Leiomyosarcoma Foundation
- 香港神經纖維瘤協會
  Hong Kong Neurofibromatosis Association
- 香港雷特氏症協會
  Hong Kong Rett Syndrome Association
- 香港小腦萎縮症協會
  Hong Kong Spinocerebellar Ataxia Association
- 香港威爾遜氏症協會
  Hong Kong Wilson's Disease Patient Group
- PNH 病人權益關注組
  PNH Concern Group
- 勉逆歷協進會
  Primary Immunodeficiency League
- 香港視網膜病變協會
  Retina Hong Kong

會員所涵蓋的病種 *：259

Types of diseases among members*: 259

1 10q26 Microdeletion Syndrome (Genetic Disorder)
2 14q Deletion Syndrome (Genetic Disorder)
3 16q11.2 & q22.2 Deletion Syndrome (Genetic Disorder)
4 18q21.2 Duplication Syndrome (Genetic Disorder)
5 1p36 Deletion Syndrome (Genetic Disorder)
6 1q44 Deletion Syndrome (Genetic Disorder)
7 2q37 Microdeletion Syndrome (Genetic Disorder)
8 3q23 & 3q25 Deletion Syndrome (Genetic Disorder)
9 8p Inverted Duplication/Deletion Syndrome (Invdupdel(8p)) (Genetic Disorder)
10 Achondroplasia (軟骨發育不全症)
11 Acromegaly (肢端肥大症)
12 ACTA1-related Congenital Myopathy (Genetic Disorder)
13 Alagille Syndrome (阿拉吉歐氏症)
14 Albinism (白化病)
15 Allan-Herndon-Dudley Syndrome (AHDS) / MCT8 Deficiency (MCT8 缺乏症)
16 Alstrom Syndrome (AS) (阿爾斯特倫綜合症)
17 Alveolar Soft Part Sarcoma (肺泡狀軟組織肉瘤)
18 Amyloidosis (AL) (類澱粉沉積症)
19 Amyotrophic Lateral Sclerosis (ALS) (肌萎縮性脊髓側索硬化症)
20 Angelman Syndrome (AS) (天使綜合症)
21 Angiosarcoma (血管肉瘤)
22 Anomalous origin of the Left Coronary Artery from Pulmonary Artery (ALCAPA) (肺動脈左冠狀動脈異常症)
23 Anti-MOG Associated Encephalomyelitis (抗 MOG 相關腦脊髓炎)
24 Anti-NMDA Encephalitis (自體免疫性腦炎)
25 Antisynthetase Syndrome (抗合成酶綜合症)
26 Apert Syndrome (亞伯氏症)
27 Aplastic Anemia (再生不良性貧血症)
28 Aromatic L-amino Acid Decarboxylase Deficiency (AADC) (芳香族 L- 胺基酸類脱羧基酶缺乏症)
29 Arthrogryposis Multiplex Congenita (AMC) (先天性多發性關節攣縮症)
30 ATAD3A Syndrome (Genetic Disorder)
31 ATR-X Thalassemia Syndrome (ATRX) (α 地中海貧血 X- 連鎖智力障碍症)
32 Atypical Hemolytic Uremic Syndrome (aHUS) (非典型溶血性尿毒症)
33 Autoimmune Insulin Syndrome (胰島素自身免疫綜合症)
34 Autosomal Recessive Polycystic Kidney Disease (ARPKD) (常染色體隱性多囊腎)
35 Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (ARSACS) (Charlevoix-Saguenay 型隱性痙攣性共濟失調症)
36 Beckwith-Wiedemann Syndrome (BWS) (貝克威思－威德曼症候群)
37 Behcet's Disease (BD) (貝賽特氏症)
38 Bilateral Perisylvian Polymicrogyria (BPP) (雙側西耳維厄斯周圍多小腦回畸形症)
39 Burning Mouth Syndrome (BMS) (口腔灼熱症侯群)
40 Cardiofaciocutaneous Syndrome (CFC) (CFC 症侯群)
41 CDKL5 deficiency disorder (CDD) (CDKL5 缺乏症)
42 Central Core Disease (CCD) (肌中央軸空症)
43 Charcot-Marie-Tooth Disease - Type 2 (CMT2) (進行性神經性腓骨肌萎縮症第 2 型)
44 Chronic Hemolytic Anemia (慢性溶血性貧血症)
45 Chronic Inflammatory Demyelinating Polyneuropathy (CIPD) (慢性脱髓鞘性神經炎)
46 Chronic Suppurative Osteomyelitis (慢性化膿性骨髓炎)
47 Chronic Thromboembolic Pulmonary Hypertension (CTEPH) (慢性血栓栓塞性肺高壓)
48 CNS Vasculitis (腦血管炎)
49 Cockayne Syndrome (柯凱因氏症侯群)
50 Coenzyme Q10 Deficiency-7 (Genetic Disorder)
51 Coffin Siris Sydrome (Coffin Siris 綜合症)
52 Collagen XII Myopathy (膠原蛋白 XII 缺失肌肉萎縮症)
53 Complex Regional Pain Syndrome (CRPS) (複雜性局部疼痛症) / Reflex Sympathetic Dystrophy Syndrome (RSD) (交感神經失調症)
54 Cone Dystrophy (錐細胞失養症)
55 Cone-rod Dystrophy (視幹細胞營養不良症)
56 Congenital Bile Acid Synthesis (CBAS) (胆汁酸合成障碍)
57 Congenital Glaucoma (先天性青光眼)
58 Congenital Insensitivity to Pain with Anhidrosis (CIPA) (先天性痛覺不敏感症合併無汗症)
59 Congenital Muscular Dystrophy (CMD) (先天性肌營養不良症)
60 Cornelia De Lange Syndrome (CdLS) (狄蘭氏症侯群)
61 Costello Syndrome (克斯提洛氏彈性蛋白質缺陷症)
62 Cri-Du-Chat Syndrome (貓哭症)
63 Crohn's Disease (克隆氏症)

64 CTNNB1 Syndrome (Genetic Disorder)
65 Cushing's Syndrome (CS) (庫欣氏症候群)
66 Cystic Fibrosis (囊腫性纖維化)
67 Dandy-Walker Syndrome (Dandy-Walker 氏症候群)
68 Ddx3x Syndrome (Genetic Disorder)
69 Dermatomyositis (皮肌炎)
70 DiGeorge Syndrome (DGS) (迪喬治症侯群)
71 DNM1-L Syndrome (Genetic Disorder)
72 Dravet Syndrome (DS) (卓飛症候群)
73 Duchenne Muscular Dystrophy (DMD) (杜興氏肌肉營養不良症)
74 DYRK1A Syndrome (Genetic Disorder)
75 Ectodermal Dysplasia (外胚層發育不良症)
76 Ehlers-Danlos Syndrome (埃勒斯 - 當洛二氏症)
77 Emery-Dreifuss Muscular Dystrophy (EDMD)
78 Eosinophilic Granulomatosis with Polyangiitis (EGPA) (嗜酸性肉芽腫性多血管炎)
79 Epidermolysis Bullosa (EB) (表皮溶解水皰症)
80 Epithelioid Hemangioendothelioma (EHE) (上皮樣血管內皮瘤)
81 Erythromelagia (紅斑性肢痛症)
82 Fabry Disease (法布瑞氏症)
83 Facioscapulohumeral Muscular Dystrophy (FSHD) - Type 1 (面肩肱型肌肉營養不良症第 1 型)
84 Familial Amyloid Polyneuropathy (FAP) (家族性澱粉樣物多發性神經病變)
85 Familial Hypertriglyceridemia (FH)
86 Fibrodysplasia Ossificans Progressiva (FOP) (進行性肌肉骨化症)
87 Floating-Harbor Syndrome (FHS) (Floating-Harbor 綜合症)
88 Fragile X Syndrome (FXS) (X 染色體脆弱症)
89 Gitelman syndrome (吉特曼症候群)
90 Glioblastoma Multiforme (GBM) (膠質母細胞瘤)
91 Glutamate Receptor, Ionotropic, N-Methyl D-Aspartate 1 (GRIN 1)
92 Glutaric Acidemia Type 1 (GA1) (戊二酸血症第 1 型)
93 Glutaric Acidemia Type 2 (GA2) (戊二酸血症第 2 型)
94 Glycogen Storage Disease (GSD) - Type 1，G6PC Variants (肝醣儲積症第 1 型，G6PC 病變異)
95 Glycogen Storage Disease (GSD) - Type 1A (肝醣儲積症第 1A 型)
96 Glycogen Storage Disease (GSD) - Type 1B (肝醣儲積症第 1B 型)
97 Glycogen Storage Disease (GSD) - Type 2 (肝醣儲積症第 2 型) / Pompe Disease (龐貝氏症)
98 Glycogen Storage Disease (GSD) - Type 6 (肝醣儲積症第 6 型)
99 Glycogen Storage Disease (GSD) - Type 9A (Pathogenic Variant) (肝醣儲積症第 9A 型)
100 Guillain-Barré Syndrome (GBS) (格林 - 巴利綜合征)
101 Hereditary Angiodema (HAE) (遺傳性血管性水腫症)
102 Hereditary Spastic Paraplegia (HSP) (遺傳性痙攣性下身麻痺症)
103 Hirschsprung's Disease (HD) (先天性巨結腸症)
104 Huntington's Disease (HD) (亨丁頓舞蹈症)
105 Ichthyosis (斑色魚鱗癬症)
106 Idiopathic Intracranial Hypertension (IIH) (特發性顱內壓增高症)
107 Idiopathic Multicentric Castleman Disease (iMCD) (特發性多中心卡斯特曼病)
108 Idiopathic Pulmonary Fibrosis (IPF) (特發性肺纖維化)
109 Idiopathic Thrombocytopenic Purpura (ITP) (自發性血小板缺乏紫斑症)
110 IgG4-related Chronic Sclerosing Sialadenitis
111 Interstitial Lung Disease (ILD) (間質性肺病)
112 Intravenous Leiomyomatosis (IVL) (靜脉內平滑肌瘤)
113 Jacobsen Syndrome (JS) (雅各森症候群)
114 Kabuki Syndrome - Type 2 (KDM6A) (歌舞伎症候群 - 第 2 型)
115 Kallmann Syndrome (KS) (卡爾曼氏症)
116 KCNB1 Syndrome (Genetic Disorder)
117 KCNQ2 Syndrome (Genetic Disorder)
118 Kennedy Disease (甘迺迪氏症) / Spinal and Bulbar Muscular Atrophy (SBMA) (脊髓延髓性肌肉萎縮症)
119 KID Syndrome (heterogenous variant in GJB2) (綜合症魚鱗病)
120 KIF1A (Genetic Disorder)
121 KIF5C Syndrome (Genetic Disorder)
122 Kleefstra Syndrome (KS) (Kleefstra 綜合症)
123 Klippel-Trénaunay Syndrome (KTS) (靜脈畸形骨肥大綜合症)
124 Langerhans Cell Histiocytosis (LCH) (蘭格罕細胞組織球增生症)
125 Larsen Syndrome (顎裂 - 先天性脱位症候群)
126 Leber's Congenital Amaurosis - Type 1 (LCA1) (Genetic Disorder) (先天性黑蒙症 - 第一型) (基因突變)
127 Leber's Congenital Amaurosis - Type 3 (LCA3) (Genetic Disorder) (先天性黑蒙症 - 第三型) (基因突變)
128 Leigh Syndrome (Leigh 氏症候群)
129 Leiomyosarcoma (LMS) (平滑肌肉瘤)
130 Lennox-Gastaut syndrome (LGS) (雷葛氏症候群)

131 Leopard Syndrome (豹皮症候群)
132 Limb-girdle Muscular Dystrophy (LGMD) - Type 2A (肢帶型肌肉失養症 - 第 2A 型)
133 Linear Scleroderma (線性硬皮症)
134 Lissencephaly (平腦症)
135 Lymphangioleiomyomatosis (LAM) (淋巴管平滑肌增生症)
136 Maple Syrup Urine Disease (MSUD) (楓糖尿症)
137 Marfan Syndrome (MFS) (馬凡氏症)
138 McCune-Albright Syndrome (MAS) (纖維性骨失養症)
139 Medulloblastoma (髓母細胞瘤)
140 Melanomae / Eye Ciliary Body Tumor (眼睫狀體黑色素瘤)
141 Mesothelioma (間皮瘤)
142 Methylmalonic Acidemia (MMA) (甲基丙二酸血症)
143 Mitochondrial Cardiomyopathy (線粒體心肌病變)
144 Mitochondrial Disease (線粒體病)
145 Mixed Connective Tissue Disease (MCTD) (混合性結締組織病)
146 Mixed Gonadal Dysgenesis (混合型性腺發育不良)
147 Moebius Syndrome (莫比斯症候群)
148 Monoclonal Gammopathies (單克隆免疫球蛋白沉積病)
149 Motor Neuron Disease (MND) (運動神經元病)
150 Mowat-Wilson Syndrome (MWS)
151 Moyamoya Disease (毛毛樣腦血管病)
152 Mucopolysaccharidoses (MPS) - Type 2 (黏多醣症第 2 型)
153 Mucopolysaccharidoses (MPS) - Type 3 (黏多醣症第 3 型)
154 Mucopolysaccharidoses (MPS) - Type 4 (黏多醣症第 4 型)
155 Mucopolysaccharidoses (MPS) - Type 4A (黏多醣症第 4A 型)
156 Mucopolysaccharidoses (MPS) - Type 6 (黏多醣症第 6 型)
157 Multiple Epiphyseal Dysplasia (MED) (多發性骨骺發育不全症)
158 Multiple Pituitary Hormome Deficiency (MPHD) / Anterior Pituitary Hypoplasia (腦下垂體發育不全症)
159 Multiple Sclerosis (MS) (多發性硬化症)
160 Multiple System Atrophy (MSA) - Pakinsonian Subtype (多重系統退化症 - 非典型巴金森症)
161 Multiple System Atrophy (MSA) - Type C (多重系統退化症 -C 型)
162 Myasthenia Gravis (MG) (重症肌無力症)
163 Myhre Syndrome (Myhre 綜合症)
164 NAA-10-related neurodevelopmental syndrome (NAA10 相關神經發育綜合症)
165 Nemaline Myopathy (桿狀體肌肉病變)
166 Neuroendocrine Tumor (NET) (神經內分泌腫瘤)
167 Neurofibromatosis (NF) - Type 1 (神經纖維瘤第 1 型)
168 Neurofibromatosis (NF) - Type 2 (神經纖維瘤第 2 型)
169 Neuromyelitis Optica (NMO) (視神經脊髓炎)
170 Nicolaides-Baraitser Syndrome (NCBRS)
171 Niemann-Pick Disease Type C (NPC) (尼曼匹克症 C 型)
172 Niemann-Pick Disease Type C1 (NPC1) (尼曼匹克症 C1 型)
173 Non-infectious Non-Anterior (NINA) Uveitis (非感染性非前段葡萄膜炎)
174 Noonan Syndrome (努南氏症候群)
175 Optic Neuropathy (視神經病變)
176 Osteogenesis Imperfecta (OI) (成骨不全症)
177 PACS1 Related Syndrome (Genetic Disorder)
178 Paroxysmal Kinesigenic Dyskinesia (原發性陣發性動作型運動不良症)
179 Paroxysmal Nocturnal Hemoglobinuria (PNH) (陣發性夜間血紅素尿症)
180 Pemphigus Vulgaris (天皰瘡)
181 Persistent Hyperplasia of Primary Vitreous (PHPV) (持續增生性原始玻璃體症)
182 Peutz-Jeghers Syndrome (PJS) (黑斑息肉症候群)
183 Phakomatosis Pigmentovascularis (PPV) (色素血管性母斑症)
184 Phenylketonuria (PKU) (苯丙酮尿症)
185 Phosphatase and Tensin Homolog (PTEN) (Genetic Disorder) (磷酸酯酶與張力蛋白同源物) (基因病變)
186 Pierre Robin Syndrome (PRS) (皮爾羅賓症侯群)
187 Pitt-Hopkins Syndrome (皮特 - 霍普金斯症候群)
188 Polymyositis (多發性肌炎)
189 Prader-Willi Syndrome (PWS) (小胖威利症)
190 Primary Ciliary Dyskinesia (PCD) (先天性纖毛運動障礙症)
191 Primary Hyperoxaluria Type 1 (PH1) (原發性高草酸尿症 1 型)
192 Primary Immunodeficiency (PID) (原發性免疫缺陷病)
193 Primary Pulmonary Arterial Hypertension (肺動脈高壓)
194 Progressive Pseudorheumatoid Dysplasia (PPD) (進行性假性類風濕性骨發育不良症)
195 Propionic Acidemia (丙酸血症)
196 PTEN Hamartoma Tumor Syndrome (PHTS) (PTEN 過誤腫症)
197 Pulmonary Alveolar Proteinosis (PAP) (肺泡蛋白質沉著症)
198 Pyruvate Dehydrogenase Deficiency (丙酮酸鹽脫氫酵素缺乏症)

199 Raynaud Phenomenon (雷諾氏現象)
200 Relapsing Polychondritis (RP) (復發性多軟骨炎)
201 ReNU Syndrome
202 Retinitis Pigmentosa (RP) (視網膜色素病變)
203 Rett Syndrome (RTT) (雷特氏症)
204 Rhabdomyosarcoma (RMS) (橫紋肌肉瘤)
205 Rubinstein-Tabyi Syndrome (RTS) (Rubinstein-Taybi 症候群)
206 Russell-Silver Syndrome (RSS) (羅素 - 西弗氏症)
207 Sacral Chordoma (骶骨脊索瘤)
208 Schaaf-Yang Syndrome (SYS) (Schaaf-Yang 症候群)
209 Schinzel Giedion Syndrome (SGS) (Schinzel Giedion 綜合症)
210 Sciatic Nerve Tumors / Schwannoma (坐骨神腫瘤)
211 SCN1A Syndrome (Genetic Disorder)
212 SCN2A Syndrome (Genetic Disorder)
213 SCN4A Myopathy (Genetic Disorder)
214 SCN8A Syndrome (Genetic Disorder)
215 Short Bowel Syndrome (短腸症)
216 Sialidosis Type 1 (涎酸酵素缺乏症第 1 型)
217 Smith-Magenis Syndrome (SMS) (史密斯 - 馬吉利氏症)
218 Spina Bifida (SB) (先天性脊椎裂症)
219 Spinal Muscular Atrophy (SMA) - Type 1 (脊髓肌肉萎縮症第 1 型)
220 Spinal Muscular Atrophy (SMA) - Type 2 (脊髓肌肉萎縮症第 2 型)
221 Spinal Muscular Atrophy (SMA) - Type 3 (脊髓肌肉萎縮症 3 型)
222 Spinalcerebellar Ataxia (SCA) - Type 2 (小腦萎縮症第 2 型)
223 Spinalcerebellar Ataxia (SCA) - Type 3 (小腦萎縮症第 3 型)
224 Spinalcerebellar Ataxia (SCA) - Type 35 (小腦萎縮症 - 第 35 型)
225 Spinalcerebellar Ataxia (SCA) - Type 40 (小腦萎縮症 - 第 40 型)
226 Spondyloepiphyseal Dysplasia Congenita (SEDC) (先天性脊椎骨骺發育不全症)
227 Stat1 Immunodeficiency (Genetic Disorder) (Stat1 免疫缺陷) (基因突變)
228 Stiff Person Syndrome (SPS) (僵硬人症侯群)
229 Succinic Semialdehyde Dehydrogenase Deficiency (SSADH) (琥珀酸半醛脱氫酶缺乏症)
230 Supernumerary Chromosome 8 Syndrome (Genetic Disorder)
231 Symbrachydactyly (先天手差異)
232 Takayasu Arteritis (TA) (高安氏症)
233 Tetratricopeptide Repeat, Ankyrin Repeat and Coiled-coil containing 2 (TANC2)
234 Thalassemia Major - Type B (地中海貧血症乙型) 重型
235 TRIO and F-actin Binding Protein Syndrome (TRIOBP) (Genetic Disorder)
236 Tuberous Sclerosis Complex (TSC) (結節性硬化症)
237 Usher Syndrome (尤塞氏綜合症)
238 Verrucous Venous Malformation (VVM) (尤狀靜脈畸形症)
239 Vitamin D-Resistant Rickets (抗維生素 D 性佝僂病)
240 Vogt-Koyanagi-Harada Disease (VKH Disease) (原田氏病)
241 von Hippel-Lindau Disease (VHL Disease) (希佩爾 - 林道病)
242 Waardenburg Syndrome (WS) (瓦登伯革氏症侯群)
243 Waldenstrom's Macroglobulinemia (WM) (華氏巨頭蛋白血症)
244 Walker-Warburg Syndrome (WWS) (Walker-Warburg 綜合症)
245 WDR45 (Genetic Disorder)
246 Wells Syndrome (韋爾斯綜合症)
247 West Syndrome-Infantile Spasms (韋氏症 - 嬰兒痙攣症)
248 Williams Syndrome (WS) (威廉氏症候群)
249 Wilson's Disease (WD) (威爾遜氏症)
250 Wolf-Hirschhorn Syndrome (WHS) (沃夫 - 賀許宏氏症候群)
251 Wolfram Syndrome (Wolfram 症候群)
252 Worster-Drought Syndrome (WDS)
253 WWOX Syndrome (Genetic Disorder)
254 Xeroderma Pigmentosum (XP) (著色性乾皮症)
255 X-linked Agammaglobulinemia (XLA) (X- 連鎖無丙種球蛋白血症)
256 X-linked Hypophosphatemic Rickets (XLH) (性聯遺傳型低磷酸鹽佝僂症)
257 X-linked Myotubular Myopathy (XLMTM) (肌小管病變)
258 X-linked Retinoschisis (XLRS) (Genetic Disorder)
259 X-linked Thrombocytopenia (XLT) (Genetic Disorder)

* 以上病種均由病人或其家屬於登記入會時提供。
* The above types of diseases were provided by patients or their families at the time of membership registration.

附錄二、理事會成員

暫代會長： 賴家衛先生 (患者，政策研究及倡議經理)
內務副會長：從缺
外務副會長：黃耀文先生 (患者，公司主管)
名譽秘書： 徐江琼女士 (患者家屬，理財顧問)
名譽司庫： 楊嘉儀女士 (患者，註冊會計師)
理事 * ： 翟佩茵女士 (患者家屬，行政人員)
周權棣先生 (患者家屬，執業護士)
何正琛先生 (患者，實驗室技術員)
梁培基先生 (患者，退休科企聯合創始人)
黃敏兒女士 (註冊社工)

*(* 按英文姓氏排序)*

附錄三、秘書處成員

阮佩玲女士 (義務總幹事)
陳淑雲女士 (服務統籌經理及護理顧問)
賴佩琪女士 (業務發展經理)
陳蔚斯女士 (項目主任)
黃桂婉女士 (活動助理)

附錄四、顧問

(按英文姓氏排序)
車錫英教授
陳麗雲教授 , 太平紳士
陳凱珊醫生
鍾一諾教授
許鍾妮醫生
陸志聰醫生 , 太平紳士
彭彥茹醫生
盛斌醫生
黃志基教授
黃文熙先生
楊雲峰先生

附錄五、科學及醫療諮詢委員會

主席： 鍾侃言醫生
委員 * ： 陳振勝教授
董咚教授
李雪教授
邵得志醫生
Dr. Sarah WYNN

*(* 按英文姓氏排序)*

附錄六、罕盟的本地及境外參與

罕盟是下列本地和境外機構的成員：
- 香港社會服務聯會
- 香港復康聯會
- 亞太罕見病聯盟
- 國際罕見病聯盟

鳴謝

(按英文字母排序)
- 所有支持者、捐贈者和贊助機構
- 義務公關顧問
  Ernest & Donald Marketing Communications Ltd
- 社會福利署 — 殘疾人士 / 病人自助組織資助計劃
- 罕盟顧問
- 罕盟理事會成員
- 罕盟科學及醫療諮詢委員會成員
- 罕盟秘書處成員及義工

Appendix II – Members of the Council

Acting Chairman (Mr. Terry LAI): Patient | Policy research & advocacy manager)
Vice-Chairman (Internal Affairs): Vacant
Vice-Chairman (External Affairs): Mr. Davy WONG (Patient | Company manager)
Honorary Secretary: Ms. Helen TSUI (Patient's family | Financial advisor)
Honorary Treasurer: Ms. Margaret YEUNG (Patient | Certified Public Accountant)
Council Members*: Ms. Fiona CHAK (Patient's family | Administration staff)
Mr. Ronald CHOW (Patient's family | Nurse practitioner)
Mr. Sam HO (Patient | Lab technician)
Mr. Raymond LEUNG (Patient | Retired corporate co-founder)
Ms. Amy WONG (Registered social worker)

(**In alphabetical order of last name*)

Appendix III – Staff of the Secretariat

Ms. Rebecca YUEN (Honorary Chief Executive)
Ms. Amy CHAN (Service Coordination Manager & Nursing Advisor)
Ms. Emily LAI (Business Development Manager)
Ms. Iris CHAN (Project Officer)
Ms. Pat WONG (Programme Assistant)

Appendix IV – Advisors

(In alphabetical order of last name)
Professor CHAIR Sek Ying
Professor Cecilia CHAN, JP
Doctor Sophelia CHAN
Professor Roger CHUNG
Doctor Joannie HUI
Doctor Che-chug LUK, JP
Doctor Shirley PANG
Doctor Bun SHENG
Professor Ian WONG
Mr. Stanley WONG
Mr. Martin YEUNG

Appendix V – Scientific & Medical Advisory Committee

Chairman: Doctor Brian CHUNG
Members*: Professor Danny CHAN
Professor Dong DONG
Professor Shirley LI
Doctor Byron SHIU
Doctor Sarah WYNN

(**In alphabetical order of last name*)

Appendix VI – RDHK's local and overseas involvements

RDHK is a member of the following local and overseas organisations:
- Hong Kong Council of Social Service
- The Hong Kong Joint Council for People with Disabilities
- Asia Pacific Alliance for Rare Disease Organisations
- Rare Diseases International

Acknowledgement

(In alphabetical order)
- All supporters, donors and sponsors
- Ernest & Donald Marketing Communications Ltd, honorary PR advisor
- Financial Support Scheme for Self-help Organisations of Persons with Disabilities / Chronic Illnesses, Social Welfare Department
- RDHK advisors
- RDHK council members
- RDHK Scientific & Medical Advisory Committee members
- RDHK staff and volunteers

香港罕見疾病聯盟
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Facebook : https://www.facebook.com/RareDiseaseHK
Instagram : https://www.instagram.com/rdhk2014/
Linkedin : https://www.linkedin.com/company/rdhk/

Website　Facebook　Instagram　LinkedIn

社聯 HKCSS 機構會員 AGENCY MEMBER