香港罕見疾病聯盟
RARE DISEASE HONG KONG

2024 會務報告

ANNUAL REPORT

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
2024 年會務報告
(2025 年 2 月)

Rare Disease Hong Kong Limited
Annual Report 2024
(February 2025)

回首十年，無忘初衷。香港罕見疾病聯盟（下稱「罕盟」）在 2024 年踏入十周年，繼續堅守使命，持續推動政策倡導、提升公眾對罕見疾病的認知，並加強病友支援與能力建設。透過與政府及醫療機構的緊密合作，罕盟在罕病政策與醫療可及性方面取得顯著進展。同時，藉著多元化的社區活動、教育計劃及境外交流，進一步推動社會對罕病群體的關注與支持，為更多患者爭取福祉。

Looking back on the past decade, we remain true to our original aspiration. Entering its 10th anniversary in 2024, Rare Disease Hong Kong (RDHK) adhered to its mission by continuously promoting policy advocacy, enhancing public awareness of rare diseases, and strengthening patient support and capacity building. Through close cooperation with the government and medical institutions, RDHK has made remarkable progress in rare disease policies and healthcare accessibility. At the same time, through diverse community activities, educational programmes and overseas exchanges, it has further promoted social attention and support for the rare disease community, striving for the betterment of more patients.

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）建立恆常溝通機制，分別於 2024 年 4 月、9 月和 12 月進行會議，推動多項與罕病相關的政策及措施落實，取得階段性進展。

a. **病人名冊計劃：**醫管局逐步擴展病人名冊，採取新的數據收集方法，使涵蓋的罕病種類有所增加。與此同時，醫管局計劃以商業模式提供疾病數據，讓研究機構和藥廠參考，以促進罕病治療的發展。

b. **跨專科團隊一站式覆診：**雖然目前僅於香港兒童醫院設有跨專科一站式覆診，但醫管局計劃在九龍中區試行針對表皮溶解水皰症（EB）及神經纖維瘤（NF）的成人跨專科團隊服務。若試行成功，未來將考慮加入其他病種。

c. **罕病藥物可及性：**醫管局持續優化藥物名冊的審批程序，集中處理藥物申請以加快審批流程。此外，針對脊髓肌肉萎縮症 (SMA) 患者，當局正在審視擴展相關用藥資助範圍的可能性。

d. **罕病患者的特殊醫療需求：**罕盟積極推動醫管局將「特殊醫療用途配方食品」（特醫食品）納入恆常服務，以減輕患者家庭的負擔。醫管局雖尚未全面改變政策，但已處理部分個案，並考慮透過捐助或基金資助有需要的病人。

e. **推動政策用語統一：**醫管局確認將逐步把「不常見疾病」改為「罕見病」，以配合政府及國際標準的用語，並著手處理相關行政調整。

1. Policy advocacy

1.1 Promote the implementation of rare disease policies

RDHK has established a regular communication mechanism with the Hospital Authority (HA). Meetings were held in April, September and December 2024, promoting the implementation of a number of policies and measures on rare diseases and achieving progress in stages.

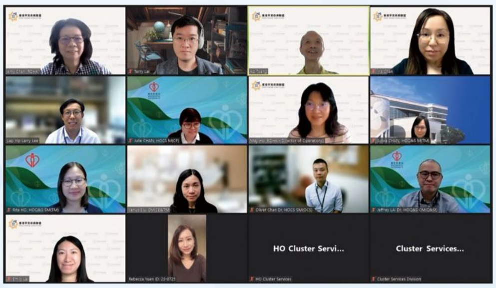
a. **Patient registry programme:** HA gradually expanded the patient registry and adopted new data collection methods, increasing the types of rare diseases covered. Meanwhile, HA is planning to provide disease data in a commercial model for reference by research institutions and pharmaceutical companies to promote the development of rare disease treatments.

b. **One-stop multidisciplinary follow-up consultations:** Currently, only the Hong Kong Children's Hospital offers one-stop multidisciplinary follow-up consultations. However, HA is planning to launch a pilot scheme on multidisciplinary team for adult patients with epidermolysis bullosa (EB) and neurofibromatosis (NF) in Central Kowloon. If the pilot scheme is successful, such service may be extended to other diseases in the future.

c. **Accessibility of rare disease drugs:** HA has been optimising the approval process of the drug formulary. Centralising the handling of drug inclusion applications helps to accelerate the approval process. In addition, for patients with spinal muscular atrophy (SMA), the authorities are examining the possibility of expanding the scope of subsidy for the relevant drug.

d. **Special medical needs of rare disease patients:** RDHK actively promoted the inclusion of "food for special medical purposes" (medical foods) in the regular services of HA to reduce the burden on families with rare disease patients. Although HA has not yet fully changed its policy, it has handled some cases and considered providing assistance to patients in need through donations or funds.

e. **Promote the unification of policy terminology:** HA confirmed that the term "uncommon disorders" would be gradually replaced by "rare diseases" to align with the government and international standards, and relevant administrative adjustments would also be made.



透過這些會議和持續的倡議工作，罕盟與醫管局的合作為罕病患者帶來實質改善，並為未來的政策優化奠定基礎。

Through these meetings and continuous advocacy efforts, the cooperation between RDHK and HA has brought tangible improvements for rare disease patients and laid the foundation for future policy optimisation.

1.2 就《施政報告》公眾諮詢提交意見書

罕盟向政府提交《***2024 年施政報告***》公眾諮詢意見書，提出八項建議，並向醫衛局局長及立法會衛生事務委員會反映罕病患者的醫療、藥物及福利需求。該八項建議如下：

a. 履行「第一責任人」的角色，主導組成跨界別罕病策略協調機制
b. 緊貼內地的罕見病目錄，制訂符合香港的綜合計劃
c. 為罕病患者提供人性化的臨床服務
d. 解除對脊髓肌肉萎縮症患者資助用藥的年齡限制
e. 將提供特殊醫療用途配方食品作為常規服務
f. 執行醫療科技早期預警，及早規劃資源分配的優先順序
g. 就罕病診治開展與大灣區協作
h. 將帶狀疱疹疫苗納入疫苗接種資助計劃

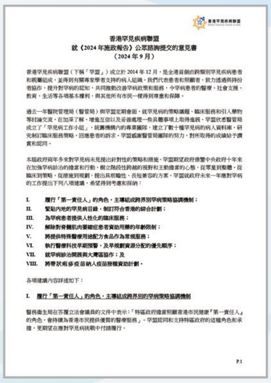
請掃瞄二維碼閱讀全文。

1.2 Submission on the 2024 Policy Address public consultation

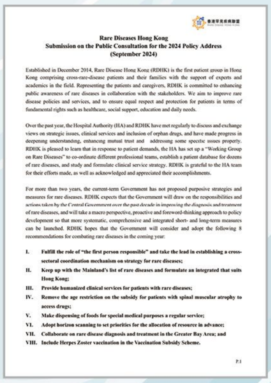
RDHK submitted a written submission on ***the 2024 Policy Address*** public consultation to the government, putting forward 8 recommendations, and reflecting the medical, drug and welfare needs of rare disease patients to the Secretary for Health and the Legislative Council Panel on Health Services. The 8 recommendations are as follows:

a. Fulfill the role of "the first person responsible" and take the lead in establishing a cross-sectoral coordination mechanism on strategy for rare diseases
b. Keep up with the Mainland's list of rare diseases and formulate an integrated that suits Hong Kong
c. Provide humanized clinical services for patients with rare diseases
d. Remove the age restriction on the subsidy for patients with spinal muscular atrophy to access drugs
e. Make dispensing of foods for special medical purposes a regular service
f. Adopt horizon scanning to set priorities for the allocation of resource in advance
g. Collaborate on rare disease diagnosis and treatment in the Greater Bay Area
h. Include herpes zoster vaccination in the Vaccination Subsidy Scheme

Please scan the QR code to read the full submission.


香港罕見病聯盟
就《2024年施政報告》公眾諮詢提交的意見書
（2024年9月）

中文版


Rare Diseases Hong Kong
Submission on the Public Consultation for the 2024 Policy Address
(September 2024)

English version

1.3 持續倡議　促進罕病患者醫療與福利改善

罕盟分別致函中央人民政府駐香港聯絡辦事處（中聯辦）、醫管局及衞生署，提出多項政策優化建議，以改善罕病患者的醫療與生活支援。倡議內容包括：

a. 促請中聯辦為回內地就醫的罕病病友外傭提供簽證便利；

b. 就鬆皮症 (Ehlers-Danlos syndromes) 患童受傷到急症室求醫的不快經歷，建議當局加強前線醫護的罕病培訓，以提升患者的護理及心理支援；

c. 向醫管局提供內地的特醫食品可及性研究報告及相關參考資料，並要求當局跟進苯丙酮尿症患者對配方奶粉的需求；

d. 建議醫管局將治療「阿拉吉歐症候群」的藥物Maralixibat儘快納入《藥物名冊》及安全網；

e. 促請藥物建議委員會（DAC）通過把 Ravulizumab 治療「全身型重症肌無力症」適應症納入《藥物名冊》及安全網；

f. 倡議醫管局為罕見病成人患者設立多專科聯合門診，以提升診療效果；及

g. 提議衞生署將「威爾遜氏症」篩查納入學童保健計劃，以加強早期診斷。

1.3 Continuous advocacy to improve the healthcare and welfare of rare disease patients

Advocacy letters were sent to the Liaison Office of the Central People's Government in the Hong Kong SAR (LOCPG), HA and the Department of Health (DoH) respectively, which aims to put forward a number of policy optimisation suggestions to improve the healthcare and life support of rare disease patients. The initiatives include:

a. Urge LOCPG to provide visa facilitation for foreign domestic helpers of families with rare disease patients who need to return to the Mainland for medical treatment;

b. In view of the unpleasant experience of a child with Ehlers-Danlos syndrome who was injured and sought medical treatment in the Accident and Emergency Department, it is recommended that HA should strengthen the training of frontline medical staff on rare diseases to improve patient care and psychological support;

c. Provide HA with a study report on the accessibility of medical foods in the Mainland and relevant reference materials, and request the authorities to follow up on the demand for phenyl-free food powder for phenylketonuria patients;

d. Urge HA to include maralixibat, a drug for the treatment of Alagille syndrome, in the Drug Formulary and safety net as soon as possible;

e. Request the Drug Advisory Committee (DAC) to include the indication of ravulizumab for the treatment of generalized myasthenia gravis in the Drug Formular and safety net;

f. Suggest that HA should provide multidisciplinary ambulatory services for adult patients with rare diseases to enhance the effectiveness of diagnosis and treatment; and

g. Recommend that DoH should include Wilson's disease screening in student health programmes to enhance early diagnosis.

1.4 脊髓肌肉萎縮症 (SMA) 患者生命質量調查

罕盟於 2021 年委託香港中文大學進行 SMA 患者生命質量調查，研究範圍包括量性及質性調查。質性調查已於 2023 年年中完成，而量性調查則於 2024 年年底完成數據收集，初步分析報告已完成，待校修工作完成後，將會適時公佈。

1.4 Survey on the quality of life of patients with Spinal Muscular Atrophy (SMA)

RDHK commissioned the Jockey Club School of Public Health and Primary Care in 2021 to conduct a survey on the quality of life of SMA patients. The scope of the study includes both quantitative and qualitative surveys. The qualitative one was completed in the middle of 2023, and the data collection for the quantitative survey was completed at the end of 2024. The preliminary analysis report has been completed then and will be released in due course upon completion of polishing and proofreading.

1.5 好「磷」社 XLH 病人治療需要及生活質素調查計劃

罕盟於 2024 年 2 月開展「好『磷』社」治療需要及生活質素調查，透過問卷及訪談收集「性聯遺傳型低磷酸鹽症」（XLH）患者、家屬及醫護人員的意見，涵蓋診斷、病徵、治療、病情監察、情緒壓力及病友支持等方面。調查分為兩部分，第一部分的問卷調查共收回 15 份有效問卷，並於同年 5 月完成分析報告。第二部分的個別訪談成功訪問 4 個家庭及兩位醫生，並於年底完成分析報告初稿，為未來的政策倡議提供參考依據。

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

In February 2024, RDHK launched a survey on the treatment needs and quality of life of patients with X-linked hypophosphatemia (XLH). XLH patients, their families, and medical staff were invited to complete questionnaires and/or be interviewed. Data about diagnosis, symptoms, treatments, disease management, emotional stress and patient support were collected. The survey was divided into two parts. The first part was questionnaire survey with 15 valid questionnaires collected, and the analysis report was completed in May 2024. The second part was individual interviews. A total of 4 families and 2 doctors were successfully interviewed. The first draft of the analysis report was completed at the end of the year, providing a reference for future policy advocacy.

1.6 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 2024 年 2 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)，以供持份者查閱和參考。

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

To continuously 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 February 2024 and uploaded them to RDHK’s website (https://rdhk.org/others) for stakeholders’ reference.

請掃瞄二維碼查閱及下載兩份藥物清單。
Please scan the QR code to read and download the two drug lists.

2. 提升認知

2.1 罕病研討會 2024

2024 年 11 月 30 日，罕盟舉辦「罕病研討會 2024」，主題為「『罕』建網絡，『罕』建未來」。研討會以實體形式進行，共吸引超過 250 名參加者，出席率接近九成，並得到參加者的高度評價。

研討會共有多達 41 位來自中國內地、瑞士、英國、法國、澳洲、韓國及香港的講者，包括醫學專家、學者、政策倡導者和病友組織代表。罕盟榮幸邀得香港醫務衞生局局長盧寵茂教授、中國罕見病聯盟執行理事長李林康先生及香港大學副校長（健康）劉澤星教授在早上的開幕式致辭。會議討論涵蓋中國罕見病診療研究體系建設、本地及香港大學深圳醫院的罕病服務發展、區域在推動世界衛生大會罕病決議中的角色及科學研究轉化應用等議題。

下午的研討會特設分論壇，主題分別為「罕見神經系統疾病」、「骨骼發育異常與骨科疾病」，以及「罕病患者組織分享」。歡迎到罕盟網站的「影視廊」（https://bit.ly/RDHK-videos）重温研討會精華片段。

2. Public awareness

2.1 Rare Disease Symposium 2024

The Rare Disease Symposium 2024 was held in-person on 30 November with the theme of “Living Connected, Brightens Living with the Rare”. The event attracted over 250 participants, with an attendance rate of nearly 90%, and was highly praised by the participants.

There were 41 speakers from Mainland China, Switzerland, the United Kingdom, France, Australia, South Korea and Hong Kong, including medical experts, scholars, policy advocates and representatives of patient organisations. RDHK was honored to have Professor LO Chung-mau, Secretary for Health of Hong Kong, Mr. LI Linkang, Executive Director, the Chinese Alliance for Rare Diseases, and Professor LAU Chak-sing, Vice-President & Pro-Vice-Chancellor (Health) and Dean of Medicine, The University of Hong Kong, attend the opening ceremony in the morning and deliver speeches. The morning session covered topics such as rare disease diagnosis and treatment research in China, rare disease service development in Hong Kong and HKU-Shenzhen Hospital, the region’s role in shaping the impact of the World Health Assembly Resolution on Rare Diseases, and the translation and application of scientific research, etc.

In the afternoon session, there were 3 sub-forums with the themes of “Rare Neurological Disorders”, “Rare Skeletal Dysplasia & Orthopaedic Disorders” and “Sharing by Rare Disease Patient Organisations”. The highlight video of the symposium can be viewed at the “Galleries” of RDHK’s website (https://bit.ly/RDHK-videos).

2.2 罕病公眾教育項目

罕盟去年繼續透過多元化的活動來推動罕病的公眾教育與宣傳，聚焦於不同罕見疾病的提升認知。各類病症的宣傳活動包括新聞發佈會、專題訪問、專題講座、製作小冊子及短片、豐富專題網站內容、社交媒體推廣、病友聚會等活動，具體病類涵蓋間質性肺病 (ILD)、膠質母細胞瘤 (GBM)、希佩爾 - 林道病 (VHL)、阿拉吉歐症候群 (ALGS)、轉甲狀腺素蛋白類澱粉沉積心肌病變 (ATTR-CM)、法布瑞氏症 (Fabry)、性聯遺傳型低磷酸鹽佝僂症 (XLH) 及杜興氏肌肉營養不良症 (DMD)。

罕盟亦於 2024 年世界罕病日啟動了「罕見病公眾教育計劃 2024」，以加強公眾對罕見病的認知，目的是提升診斷率、促進政策改進及提高醫療服務的可及性。計劃內容包括製作十周年紀念短片《讓罕見被看見》，出版文集《罕見病・同理心》，以及於罕盟網站的「罕病小百科」增添 20 種罕病的資料。透過這些活動，罕盟不僅希望減少患者和照顧者的孤立感，亦力求推動罕病群體在社會中的關注與支持。活動詳情請瀏覽罕盟網站（https://bit.ly/RDed2024c）。

中文版

2.2 Public education programmes on rare diseases

Last year, RDHK continued to promote public education and publicity of rare diseases. With a focus on enhancing awareness of different rare diseases, diverse activities such as press conferences, featured interviews, subject talks, production of leaflets and short videos, enriching the content of the thematic website, social media promotion, patient gatherings, etc. were rolled out for various rare diseases including interstitial lung disease (ILD), glioblastoma multiforme (GBM), von Hippel-Lindau disease (VHL), Alagille syndrome (ALGS), transthyretin amyloid cardiomyopathy (ATTR-CM), Fabry disease, x-linked hypophosphatemia (XLH) and Duchenne muscular dystrophy (DMD).

RDHK also launched the “Rare Disease Awareness Campaign 2024” on World Rare Disease Day in 2024 to strengthen public awareness of rare diseases, with a view to improving the diagnosis rate, promoting policy improvement and enhancing the accessibility of healthcare services. The campaign included producing a 10th anniversary commemorative short video themed “Making the Unseen Seen”, publishing a compilation book titled 罕見病・同理心 *(Rare Diseases・Empathy)*, and adding information on 20 rare diseases to the “Rare Disease Wiki” on RDHK’s website. Through these activities, RDHK not only hopes to reduce the sense of isolation of patients and caregivers, but also strives to promote the social attention and support of the rare disease community. Please visit RDHK’s website (https://bit.ly/RDed2024e) for campaign details.

English version

2.3 真人圖書館

「真人圖書館」讓罕病患者和照顧者透過親身分享，推動社會對罕病的認知和關注，同創共融社會。2024 年，罕盟為 12 間機構舉辦講座和工作坊，觸及人數達 2,361 人，包括中小學生、醫護學生及社區團體。活動涵蓋 16 種罕病，讓公眾更了解患者的生活挑戰與醫療需求。

2.3 Human library

The “Human Library” is a project that enables rare disease patients and caregivers to raise public awareness of rare diseases and create an inclusive society together through personal sharing. In 2024, RDHK held talks and workshops for 12 institutions, reaching 2,361 people, including primary and secondary school students, medical and nursing students, and community groups. 16 rare diseases were introduced in the project, allowing the public to better understand the patients’ challenges in life and healthcare needs.

2.4 網頁

罕盟在 2024 年採取新策略，透過社交媒體貼文引導讀者瀏覽官網，進一步提升網站流量。全年獨立瀏覽人數達 144,533 人，較 2023 年增加約 176%；總瀏覽次數達 207,111 次，增幅約 189%；頁面瀏覽量更攀升至 459,943，較去年增長 124%。這項策略有效提高公眾對罕病資訊的關注，進一步擴大罕盟的影響力。

2.4 Website

In 2024, RDHK adopted a new strategy, guiding readers to visit the official website through social media posts to further increase website traffic. The number of independent visitors throughout the year reached 144,533, an increase of approximately 176% compared with 2023; the total number of visits reached 207,111, an increase of approximately 189%; and the page views climbed to 459,943, a growth of 124% compared with last year. This strategy effectively increased public attention to rare disease information and further expanded the influence of RDHK.

2.5 社交媒體平台

罕盟社交媒體平台在過去一年持續增長，觸及更廣泛的公眾群體。全年在臉書發布259則貼文，總觸及人數達3,368,806人，追蹤者增至6,847人。Instagram則發佈202則貼文，觸及人數達2,349,775人，追蹤者增至1,321人。LinkedIn的關注人數增長亦相當顯著，全年發佈205則貼文，總曝光次數達328,081次，關注人數增至590人。

2.5 Social media platforms

RDHK's social media platforms continued to grow in the past year, reaching a wider public audience. Throughout the year, 259 posts were published on Facebook, with a total reach of 3,368,806 people, and the number of followers increased to 6,847. On Instagram, 202 posts were published, with a reach of 2,349,775 people, and the number of followers increased to 1,321. The growth of the number of followers on LinkedIn was also quite significant. 205 posts were published throughout the year, with a total exposure of 328,081 times, and the number of followers increased to 590.

2.6 罕情

《罕情》季刊續於1、4、7、10月出版，提供印刷版及網上版，內容涵蓋國際及本地罕病政策、罕盟及相關團體的動向、罕病診療與藥物資訊等。讀者群廣泛，包括政府官員、議員、醫護人員、罕病社群及公眾人士，進一步推動社會對罕病的關注與了解。歡迎到罕盟網頁下載閱讀（https://rdhk.org/RareCare）。

2.6 RareCare

The quarterly newsletter *RareCare* continued to be published in January, April, July and October, providing both print and online versions. The content covered international and local policies on rare diseases, the trends of RDHK and relavant organisations, and information on rare disease diagnosis, treatments and orphan drugs. The readership is extensive, including government officials, District or Legislative Council members, healthcare professionals, rare disease communities and the general public, further promoting social attention and understanding of rare diseases. The public is welcome to download the newsletters from RDHK's website (https://rdhk.org/RareCare).

3. 能力建設

3.1 社交及消閒活動

罕盟積極舉辦各類會員活動，讓患者及照顧者在交流互動中建立支持網絡，並且舒展身心。同時，亦積極邀請義工參與，透過接觸罕病社群，加深對患者生活和需求的認識。全年共舉辦 12 場活動，包括：

3. Capacity building

3.1 Social and leisure activities

RDHK actively organised various member activities, enabling patients and caregivers to build support networks through communication and interaction, and to relax. At the same time, volunteers were invited to participate actively in these activities, deepening their understanding of the lives and needs of patients through contact with the rare disease community. A total of 12 activities were held throughout the year, including:

苦中一點甜親子甜品工作坊
Parent-child dessert workshop

龍年動物繪畫工作坊
Animal painting workshop

春日甜蜜蜜鬆餅與水果芭菲烹飪班
Muffin and fruit parfait cooking class

SMA 病友龍的約會
SMA patient gathering

端午艾葉香囊工作坊
Mugwort sachets workshop

樂聚罕盟慶中秋
Mid-autumn festival party

齊齊做彩沙瓶工作坊
Colour sand art in bottle workshop

花籃製作體驗班
Flower basket workshop

看見・罕見
Seeing the rare

理財講座及交流午宴
Financial management talk and networking lunch

DMD 醫療專題講座暨午餐聚會
Healthcare talk on DMD and lunch gathering

紅山 x 罕盟聖誕歡樂派對
Christmas party

此外，罕盟亦協助會員獲得《音樂・人生 ~ 我們都是唱這歌》慈善演唱會、《遠在遠方的風》音樂會及電影《把幸福拉近一點》的門票，進一步豐富社交與文化體驗。全年活動總參加人數超過 520 人，較 2023 年顯著增長，反映出會員對各類活動的積極參與和熱烈支持。

In addition, RDHK helped members obtain tickets for a charity concert, a musical concert and the movie “Wish Comes True”, further enriching their social and cultural experiences. The total number of participants in the activities throughout the year exceeded 520, a significant increase compared with 2023, reflecting the active participation and enthusiastic support of members for various activities.

3.2 義工培訓

罕盟在 2024 年共為 6 名來自港大及中大的醫科生和公共衛生碩士課程學生提供實習機會，合共完成超過 1,400 小時的實習時數。此外，根據活動需要，安排了 13 次培訓，培訓對象包括 14 名醫科生及公共衛生碩士課程學生，以及 4 名罕盟義工。義工們積極參與會員活動，共協助 80 場次，總參與人次達 147 人，並協助設計「香港杜興氏肌肉營養不良症協會」的會徽及 2025 年生日卡、聖誕卡和賀年卡。罕盟亦持續透過「罕盟醫科」WhatsApp 群組分享罕見病患者的活動資訊及外訪安排，以期讓歷屆實習生在未來的專業生涯中持續關注罕見病群體的需要。

3.2 Volunteer training

In 2024, RDHK provided internship opportunities for 6 medical students and public health students from the University of Hong Kong and the Chinese University of Hong Kong, with a total of over 1,400 internship hours completed. In addition, according to the needs of activities, 13 training sessions were arranged for 14 medical students and public health students, as well as 4 RDHK volunteers. The volunteers actively participated in member activities, assisting in 80 sessions, with a total of 147 participants, and helped design the logo for DMD Association of Hong Kong, as well as the 2025 birthday cards, Christmas cards and New Year cards. RDHK also continued to share the information on patient activities and overseas visits in a WhatsApp group comprising of the past and current interns, hoping that they will continue to pay attention to the needs of the rare disease community in their future professional careers.

3.3 境外交流活動

罕盟去年積極參與內地及海外的罕見病活動，以拓展視野並深化交流。會長與營運總監分別出席比利時舉行的「**第 12 屆歐洲罕見疾病及孤兒藥產品會議**」、西班牙舉行的「**2024 年世界孤兒藥大會（歐洲大會）**」，以及北京舉行的「**2024 罕見病合作交流會**」。此外，罕盟資助團體會員領袖、理事、基本會員及職員參加上海舉行的「**2024 第十三屆中國罕見病高峰論壇**」，並於會後舉行分享會，讓參與者報告所學，交流心得，進一步提升對罕病議題的認識。會長與服務統籌經理亦出席了北京舉行的「**第七屆 DMD 國際醫患交流大會暨 DMD 新技術創新論壇**」，與各地專家及病友組織交流最新醫療技術與創新發展。這些活動有助於促進罕盟與內地及國際夥伴的合作，並持續優化本地罕病支援與倡議工作。

3.3 Exchange activities outside Hong Kong

Last year, RDHK actively participated in different events on rare diseases in the Mainland and overseas to broaden its horizons and deepen exchanges. The Chairman and Director of Operations attended the **12th European Conference on Rare Diseases and Orphan Products 2024** held in Belgium, the **World Orphan Drug Congress Europe 2024** held in Spain and the **2024 Rare Disease Symposium on Collaboration and Communication** held in Beijing respectively. In addition, RDHK subsidised the group leaders of organsational members, council members, full members, and staff to participate in the **13th China Rare Disease Summit 2024** held in Shanghai, and held a sharing session after the summit, allowing participants to report what they had learned and exchange experiences, further enhancing their understanding of rare disease issues. The Chairman and Service Co-ordination Manager also attended the **7th DMD International Patient-Doctor Exchange Conference and DMD New Technology Innovation Forum** held in Beijing, communicating with experts and patient organisations from various places on the latest medical technologies and innovative developments. These activities have helped to promote the cooperation between RDHK and partners from the Mainland and other countries, and continuously optimise local support and advocacy for rare diseases.

4. 其他工作

4.1 周年大會

罕盟於 2024 年 6 月 15 日成功舉辦第五屆會員周年大會，吸引 70 名會員出席。會上同時舉行第三屆理事會選舉，共有 11 名候選人，由於未超出理事會上限人數，全部自動當選。這次大會為會員提供了交流的平台，加強彼此聯繫，並推動罕盟未來的發展。

4. Miscellaneous work

4.1 Annual general meeting

The 5th annual general meeting was successfully held on 15 June 2024, attracting 70 members to attend. The election of the 3rd council of RDHK was also held in the meeting. As the number of candidates did not exceed the upper limit of the Council, all the 11 candidates were automatically elected. The meeting provided a platform for members to interact and connect with each other, and promoted the future development of RDHK.

4.2 罕盟十周紀念活動

十年同行，承載無數關愛與支持，共同為罕病社群築起希望的橋樑。2024 年 11 月 29 日，罕盟舉辦十周年誌慶晚宴，與會員及各界夥伴回顧十年來的發展與成果。此外，特別安排海外嘉賓參觀香港兒童醫院，加深對本地罕見病醫療服務的了解。

為紀念這重要時刻，罕盟印製了十周年紀念特刊《**十載同舟十載情**》，以情誼和溫暖為主調，收錄患者、照顧者、醫護人員、科研專家、學者、服務機構及商界夥伴的祝福與分享，見證罕盟十年的成長與夥伴間的緊密聯繫。此外，罕盟特別製作「十周年限定版座枱月曆〈2025〉」，以會員的視覺藝術作品為插畫，展現罕病社群的創意與生命力。

4.2 The 10th anniversary commemoration of RDHK

A series of programmes were arranged to celebrate the 10th anniversary of RDHK. On 29 November 2024, RDHK hosted the 10th Anniversary Gala Dinner to review the development and achievements in the past ten years with members and partners from all walks of life. In addition, overseas guests were specially arranged to visit the Hong Kong Children's Hospital to deepen their understanding of local healthcare services for rare diseases.

To commemorate this important moment, RDHK published a 10th anniversary commemorative issue, namely ***A Decade of Love and Partnership***. With a focus on friendship and warmth, it features messages from patients, caregivers, healthcare professionals, researchers, scholars, service providers and business partners, which shows the growth of RDHK in the past ten years and the close connection among partners. Besides, a "10th Anniversary Limited Edition Desk Calendar (2025)" illustrated with members' visual arts work was produced to exhibit the creativity and vitality of the rare disease community.

4.3 護理物資及服務資助先導計劃

罕盟持續推行「**護理物資及服務資助先導計劃**」，為公共醫療及社會福利未能全面覆蓋的罕病患者提供過渡性支援。全年共收到6份申請，並全部獲批。透過此計劃，罕盟不僅能即時協助有需要的患者，亦能累積政策倡議的實證資料，推動更完善的支援措施。

4.3 The Pilot Scheme of Subsidy for Healthcare Needs

RDHK continued to implement the "**Pilot Scheme of Subsidy for Healthcare Needs**" which provides transitional funding for nursing supplies and services that are not covered by the current public healthcare and social welfare services. A total of 6 applications were received throughout the year, and all were approved. Through this scheme, RDHK can not only assist patients in need immediately but also collect empirical data for policy advocacy, promoting more comprehensive support measures.

欲了解資助計劃詳情，請掃瞄二維碼。
Please scan the QR code for details of the subsidy scheme.

4.4 與其他病友組織交流

建立緊密的病友網絡，有助促進資源共享與共同發展。2024年，罕盟與7個本地罕病組織代表會面，了解各組群的現況與需求，並就活動協作、病友支援及政策倡議等議題交換意見。此外，罕盟亦參與多個病友組織的活動，為社群提供實用資訊和資源，攜手推動罕病患者的福祉。

4.4 Interchange with other patient organisations

Establishing a close patient network helps to promote resource sharing and common development. In 2024, RDHK met with representatives of 7 local rare disease organisations to understand their current situations and needs, and exchanging views on issues such as activity cooperation, patient support and policy advocacy. In addition, RDHK participated in the activities of a number of patient organisations, providing them with practical information and resources, and jointly promoting the well-being of rare disease patients.

附錄一、會員及所涵蓋病種

截至 2024 年年底為止，會員總人數為 921，其中：

1. 普通會員（關注及支持罕病患者人士）
   數目：122
2. 基本會員（罕病患者或家屬）
   數目：799
   a) 個人會員數目：790
   b) 團體會員數目：9

Appendix I – Members and types of diseases

As at the end of 2024, the total number of members was 921, among which:

1. Number of ordinary members (those who care about and support rare disease patients): 122
2. Number of full members (rare disease patients or families): 799
   a) Number of individual members: 790
   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 *：243

Types of diseases among members*: 243

1 10q26 Microdeletion Syndrome (Genetic Disorder)
2 14q Deletion Syndrome (Genetic Disorder)
3 16q11.2 & q22.2 Deletion Syndrome (Genetic Disorder)
4 18q21.2x3 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 Albinism (白化病)
14 Alexander Disease (亞歷山大症)
15 Allan-Herndon-Dudley Syndrome (AHDS) / MCT8 Deficiency (MCT8 缺乏症)
16 Alstrome 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 Anomalous origin of Left Coronary Artery from Pulmonary Artery (ALCAPA) (肺動脈左冠狀動脈異常症)
22 Anti-MOG Associated Encephalomyelitis (抗 MOG 相關腦脊髓炎)
23 Anti-NMDA Encephalitis (自體免疫性腦炎)
24 Antisynthetase Syndrome (抗合成酶綜合症)
25 Apert Syndrome (亞伯氏症)
26 Aplastic Anemia (再生不良性貧血症)
27 Aromatic L-amino Acid Decarboxylase Deficiency (AADC) (芳香族 L- 胺基酸類脱羧基酶缺乏症)
28 Arthrogryposis Multiplex Congenita (AMC) (先天性多發性關節攣縮症)
29 ATAD3A Syndrome (Genetic Disorder)
30 ATR-X Thalassemia Syndrome (ATRX) (α 地中海貧血 X- 連鎖智力障碍症)
31 Atypical Hemolytic Uremic Syndrome (aHUS) (非典型溶血性尿毒症)
32 Autoimmune Insulin Syndrome (胰島素自身免疫綜合症)
33 Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (ARSACS) (Charlevoix-Saguenay 型隱性痙攣性共濟失調症)
34 Behcet's Disease (BD) (貝賽特氏症)
35 Bilateral Perisylvian Polymicrogyria (BPP) (雙側西耳維厄斯周圍多小腦回畸形症)
36 Burning Mouth Syndrome (BMS) (口腔灼熱症侯群)
37 Cardiofaciocutaneous Syndrome (CFC) (CFC 症侯群)
38 Central Core Disease (CCD) (肌中央軸空症)
39 Charcot-Marie-Tooth Disease (CMT) (進行性神經性腓骨肌萎縮症)
40 Chronic Hemolytic Anemia (慢性溶血性貧血症)
41 Chronic Inflammatory Demyelinating Polyneuropathy (CIPD) (慢性脱髓鞘性神經炎)
42 Chronic Suppurative Osteomyelitis (慢性化膿性骨髓炎)
43 CNS Vasculitis (腦血管炎)
44 Cockayne Syndrome (柯凱因氏症侯群)
45 Coenzyme Q10 Deficiency-7 (Genetic Disorder)
46 Collagen XII Myopathy (膠原蛋白 XII 缺失肌肉萎縮症)
47 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)
48 Cone-rod Dystrophy (視幹細胞營養不良症)
49 Congenital Bile Acid Synthesis (CBAS) (胆汁酸合成障碍)
50 Congenital Glaucoma (先天性青光眼)
51 Congenital Insensitivity to Pain with Anhidrosis (CIPA) (先天性痛覺不敏感症合併無汗症)
52 Congenital Muscular Dystrophy (CMD) (先天性肌營養不良症)
53 Cornelia De Lange Syndrome (CdLS) (狄蘭氏症侯群)
54 Costello Syndrome (克斯提洛氏彈性蛋白質缺陷症)
55 Cri-Du-Chat Syndrome (貓哭症)
56 Crohn's Disease (克隆氏症)
57 CTNNB1 Syndrome (Genetic Disorder)
58 Cushing's Syndrome (CS) (庫欣氏症候群)
59 Cystic Fibrosis (囊腫性纖維化)

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

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

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

會長：曾建平先生（患者，退休企業主管）
內務副會長：阮佩玲女士（患者家屬，產品代理）
外務副會長：黃耀文先生（患者，公司主管）
義務秘書：徐江琼女士（患者家屬，理財顧問）
義務司庫：梁七根先生（患者，自由工作者）
理事 *：陳振勝教授（科學家）
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）
賴家衡先生（患者，政策研究及倡議經理）
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

何翠薇女士 （營運總監）
陳淑雲女士 （服務統籌經理及護理顧問）
陳蔚斯女士 （項目主任）
賴佩琪女士 （傳訊主任）
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

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
- 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
- 香港視網膜病變協會

Appendix II – Members of the Council

Chairman: Mr TSANG Kin Ping (patient, retired business executive)
Vice-Chairman (Internal Affairs): Ms Rebecca YUEN (patient's family, product agent)
Vice-Chairman (External Affairs): Mr Davy WONG (Patient, company manager)
Honorary Secretary: Ms Helen TSUI (patient's family, financial advisor)
Honorary Treasurer: Mr Jackie LEUNG (patient, freelancer)
Council Members*: Professor Danny CHAN (scientist)
Mr Ronald CHOW (patient's family, nurse practitioner)
Mr Sam HO (patient, lab technician)
Mr Terry LAI (patient, policy research & advocacy manager)
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 May HO (Director of Operations)
Ms Amy CHAN (Service Coordination Manager & Nursing Advisor)
Ms Iris CHAN (Project Officer)
Ms Emily LAI (Communications 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-chung LUK, *JP*
Doctor Shirley PANG
Doctor Bun SHENG
Professor Ian WONG

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
- Retina Hong Kong

香港罕見疾病聯盟
RARE DISEASE HONG KONG

香港九龍長沙灣麗閣邨麗萱樓地下101號
電話: (852) 5528 9600　傳真: (852) 2708 8915
網頁: www.rdhk.org
臉書: https://www.facebook.com/RareDiseaseHK
Instagram: https://www.instagram.com/rdhk2014
領英: https://www.linkedin.com/company/rdhk

Room 101, G/F, Lai Huen House, Lai Kok Estate, Cheung Sha Wan, Kowloon, Hong Kong
Tel: (852) 5528 9600　Fax: (852) 2708 8915
Website: www.rdhk.org
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