香港罕見疾病聯盟
RARE DISEASE HONG KONG

會務報告 2022
ANNUAL REPORT 2022

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-based approach

Governance principles:
Openness, transparency, integrity & accountability

香港罕見疾病聯盟有限公司 2022 年會務報告

(2023 年 2 月)

Rare Disease Hong Kong Limited Annual Report 2022

(February 2023)

2022 年是充滿變化的一年。新冠疫情起伏反覆，醫療系統遭受前所未有的衝擊，最終在防疫大鬆綁的寬鬆氣氛中畫上句號；治港班子全面更替，新一屆立法會和第六屆特區政府先後在年初和年中就職。面對不斷變化的環境，香港罕見疾病聯盟（下稱「罕盟」）過去一年積極把握各種新機遇，順勢而為，以持續推進各項核心業務為目標，取得多方面成果。

2022 is a year of change. The ups and downs of the COVID-19 epidemic have caused an unprecedented impact on the healthcare system, and finally the year ended in a relaxed atmosphere as most of the anti-epidemic measures were lifted. The governing team of Hong Kong has been completely changed, while the new-term Legislative Council and the sixth term SAR Government took office at the beginning and middle of the year respectively. Over the year, amidst the ever-changing environment, Rare Disease Hong Kong (hereinafter referred to as "RDHK") has actively seized the new opportunities and leveraged the trends to achieve fruitful results with the goal of making progress in different aspects of work.

1 政策倡議

1.1. 在新的政治氛圍下開始與新班子接觸

特區政府在去年七月換屆，罕盟於八月份在立法會葛佩帆議員協助下，與醫務衞生局（醫衞局）副局長李夏恩醫生短暫會面，提出對新政府在應對罕病政策的期望和建議；九月份向政府提交施政報告意見書；十月份周年研討會蒙李夏恩副局長應邀主禮並致詞；十二月向盧寵茂局長提交親啟信，請求與政府商討未來五年的罕病策略。罕盟還主動約見新一屆立法會議員，先後與林哲玄議員（醫療衞生界）、

1 Policy advocacy

1.1. Approaching the new governing team in the new political climate

Since the new HKSAR Government took office in July last year, RDHK has started to approach different policy makers. In August, with the assistance of the Legislative Council member Hon Elizabeth Quat, RDHK met briefly with Dr Libby Lee, the Under Secretary for Health, to put forward expectations and suggestions for the new Government's policy in dealing with rare diseases. In September, a submission to the Government on the policy address was made. In October, being invited, Dr Libby Lee, the Under Secretary for Health, attended the annual Rare Disease Symposium as the officiating guest and delivered a speech. In December, a letter was submitted to the Secretary for Health, Professor Lo Chung-mau, requesting to discuss with the

陳恒鑌議員（新界西南）、梁熙議員（香港島東）、陳沛良議員（選舉委員會）和陳健波議員（保險界）會面交流，請求他們在議會推動罕病議題的討論，訂定策略和規劃。

Government the strategy for the next five years. RDHK also took the initiative to meet with the newly elected Legislative Council members including Dr Hon David Lam (Medical and Health Services), Hon Ben Chan (New Territories South West), Hon Edward Leung (Hong Kong Island East), Hon Chan Pui-leung (Election Committee) and Hon Chan Kin-po (Insurance), requesting them to promote discussions on rare disease issues in the Council, formulate strategies and plans.

1.2. 施政報告意見書

罕盟自 2015 年起，每年均會就政府施政報告的公眾諮詢提交意見書。2022 年我們就現屆政府應對罕病的工作提出下列五項建議：

i. 政府主導，成立「罕病策略督導委員會」
ii. 就訂立本地罕病定義展開研究
iii. 制訂和落實五年行動計劃
iv. 拆牆鬆綁，優化和完善罕病藥物可及性機制
v. 就罕病診治開展與大灣區協作

1.2. Policy address submission

Since 2015, RDHK has made submissions on the policy address public consultation every year. In 2022, we made the following 5 recommendations for dealing with rare diseases to the current-term Government:

i. To set up a government-led "Steering Committee on Strategy for Rare Diseases"
ii. To commence a study on the local definition of rare diseases
iii. To develop and implement a five-year action plan
iv. To optimise and improve the access mechanism for orphan drugs by cutting the red tape
v. To collaborate on rare disease diagnosis and treatment in the Greater Bay Area

1.3. 倡議改善罕藥審批及用藥機制

罕盟於去年四月向有關部門提交《拆牆鬆綁，優化和完善罕病藥物可及性機制》意見書，提出以下七項具體建議：

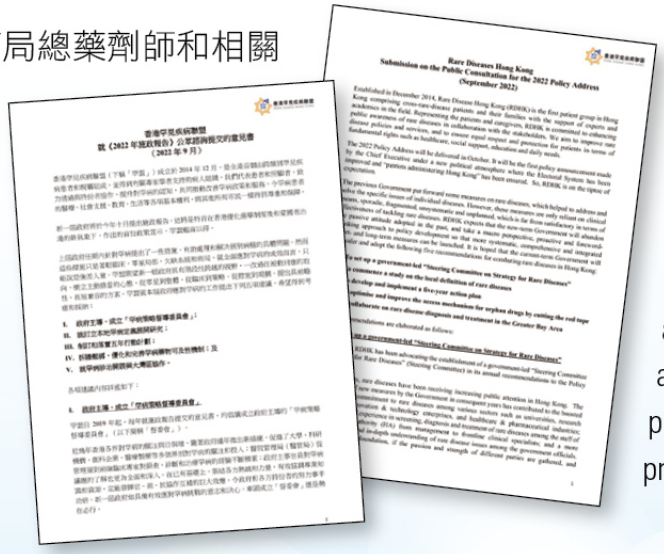
i. 優化罕病藥物註冊申請文件的要求和審批程序；
ii. 加強醫管局「總藥劑師辦事處」與藥劑業界的協作；
iii. 省略「藥事委員會」在入藥過程的非必要工作；
iv. 調整「藥事管理委員會」的會議時間和模式；
v. 走出「先有雞還是先有蛋」的因果困境，協調審理醫療檢測和入藥的兩個機制；
vi. 執行醫療科技早期預警，及早規劃資源分配的優先順序；及
vii. 推行「罕見病藥物試用期」，有系統地收集和應用真實世界數據，為加快病人使用罕見病藥物提供更多實證。

1.3. To advocate improving the approval mechanism for orphan drugs

Last year, a submission on "Optimising and Improving Access Mechanism for Orphan Drugs by Cutting the Red Tape" was made to the departments concerned with 7 recommendations as follows:

i. Optimise the documentation requirements for registration and registration process of orphan drugs;
ii. Strengthen collaboration between the Chief Pharmacist's Office of the Hospital Authority (HA) and the pharmaceutical industry;
iii. Omit the non-essential work of the Drug and Therapeutics Committee in the drug inclusion process;
iv. Adjust the schedule and mode of the Drug Management Committee meetings;
v. Solve the chicken-and-egg dilemma by coordinating the two mechanisms of medical testing and drug inclusion;
vi. Adopt early awareness and alert systems (also known as horizon scanning) to set priorities for the allocation of resource in advance; and
vii. Adopt "Orphan Drug Trial" policy and systematically collect and use real-world data to generate empirical evidence which helps to accelerate patient access to orphan drugs.

其後於十一月與醫管局總藥劑師和相關代表會面，具體討論上述建議，會面氣氛十分坦誠、務實和有建設性。罕盟會繼續主動接觸各持份者，跟進各項建議的推展進度。


香港罕見病聯盟
就《2022 年施政報告》公眾諮詢我們的意見書
（2022 年 9 月）

Rare Diseases Hong Kong
Submission on the Public Consultation for the 2022 Policy Address
(September 2022)

This was followed by a meeting with the Chief Pharmacist and relevant representatives of HA in November to discuss the above recommendations in a frank, pragmatic and constructive atmosphere. RDHK will continue approaching various stakeholders proactively to follow up on the progress of each recommendation.

1.4. 推動把不同罕病藥物和相關適應症納入名冊和安全網

罕盟一年來竭力推動把一些罕病藥物和相關適應症納入醫管局《藥物名冊》和安全網資助範圍，使相關患者早日得到適切治療，包括：

1.4. Promote the inclusion of various rare disease drugs and related indications in the Drug Formulary and safety net

Over the past year, RDHK has made every effort to promote the inclusion of the following rare disease drugs and related indications in the HA Drug Formulary and safety net, so that relevant patients can receive appropriate treatment as soon as possible:

藥物名稱	適應症
• 布羅索尤單抗	性聯遺傳低磷酸鹽佝僂
• 尼達尼布	硬皮症併發間質性肺病、惡化性纖維化間質性肺病
• Ravulizumab	陣發性夜間血紅素尿症、非典型性尿毒溶血症候群
• 利奧西呱	慢性血栓栓塞性肺高壓

Drug name	Indication
• Burosumab	X-linked Hypophosphatemia
• Nintedanib	Interstitial Lung Disease associated with Systemic Sclerosis, Progressive Fibrosing Interstitial Lung Disease
• Ravulizumab	Paroxysmal Nocturnal Hemoglobinuria, Atypical Hemolytic Uremic Syndrome
• Riociguat	Chronic Thromboembolism Pulmonary Hypertension

同時，罕盟根據醫管局和 MIMS Hong Kong 網上提供的資料，編整了《香港罕見疾病藥物參考清單》和《香港罕見癌症藥物參考清單》，並上載於罕盟網頁（https://rdhk.org/others），供有需要的人士參考和下載。

Meanwhile, based on the information provided by the HA and MIMS Hong Kong, RDHK compiled the "Drug List for Rare Diseases in Hong Kong" and "Drug List for Rare Cancers in Hong Kong", which have been posted on the website of RDHK (https://rdhk.org/others) for public reference and download if needed.

《香港罕見疾病藥物參考清單》
List of Drugs for Rare Diseases in HK
http://bit.ly/RDdruglist

《香港罕見癌症藥物參考清單》
List of Drugs for Rare Cancers in HK
http://bit.ly/RCdruglist

1.5. 脊髓性肌肉萎縮症（SMA）患者生命質量調查

罕盟於 2021 年 11 月委託香港中文大學賽馬會公共衞生及基層醫療學院進行的SMA患者生命質量調查，原定可於 2022 年年底完成。由於支持調查的受訪者人數較預期多，以及受疫情的影響，調查期會延長一年，除了進行量性研究外，更會對第三型成人患者進行質性研究，務求更全面收集臨床和真實世界數據，以供醫院管理局審批相關藥物時作參考。預計研究調查的中期報告可於 2023 年年中完成，屆時請留意罕盟的消息發佈。

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

The survey on the quality of life of SMA patients commissioned by RDHK in November 2021 to the Jockey Club School of Public Health and Primary Care, CUHK was supposed to be completed by the end of 2022. But since the number of respondents supporting the survey was higher than expected, and due to the impact of the epidemic, the survey period is to be extended for one year. In addition to quantitative research, qualitative research on Type 3 adult patients will also be conducted to collect more comprehensive clinical and real-world data for HA's reference when it comes to review and approval of the relevant drug. The interim report of the survey is expected to be completed by mid-2023. Please stay tuned with RDHK for further updates.

2 提升認知

2.1. 罕病研討會 2022

罕病研討會乃罕盟的年度盛事，去年 10 月 29 日以線上線下模式同步舉行的研討會以「同為罕病開新篇」為主題，旨在反映罕病患者和照顧者的迫切需要，並與不同持份者一起探討新一屆政府應該就應對罕病定下什麼目標，以及建議能使罕病患者真正受惠的短、中、長期政策與措施。是次研討會不但邀請到醫務衞生局副局長李夏茵醫生親臨現場擔任主禮嘉賓，還有多位來自不同界別的重量級嘉賓擔任討論嘉賓，包括*（按英文姓氏排序）*陳恒鑌議員、陳志峰教授、高拔陞醫生、林哲玄議員、陸志聰醫生、田詩蓓博士和杜啟峻醫生。登記參與線上或現場研討會的人數接近 400 人，出席率達七成。

2 Public awareness

2.1. Rare Disease Symposium 2022

Rare Disease Symposium is an annual event of RDHK. The theme of the symposium held in hybrid mode on 29 October last year was “Turning a New Page for Rare Diseases Together”. It aimed to reflect the urgent needs of rare disease patients and caregivers, to explore with different stakeholders what goals the new-term Government should set to deal with rare diseases, and to recommend short, medium and long-term policies and measures that can truly benefit rare disease patients. We were honoured to have Dr Libby Lee, the Under Secretary for Health, as the officiating guest, and a number of heavyweight guests from different sectors as panelists, including (*in alphabetical order of last name*) Hon Ben Chan, Professor Godfrey Chan, Dr Tony Ko, Dr Hon David Lam, Dr Luk Chi-chung, Dr Pamela Tin and Dr Michael To. Nearly 400 people registered for the online or in-person symposium, with an attendance rate of 70%.

2.2. 罕病公眾教育項目

罕盟不時透過不同媒體渠道發放各種罕見疾病的資訊。去年的主題包括遺傳諮詢服務、膠質母細胞瘤、間質性肺病和肺動脈高壓，形式包括新聞稿、專欄文章、短片、傳媒小組訪問、患者分享、漫畫和電視特輯等，務求以淺白易明和生動有趣的方式引起公眾對罕病議題和罕病社群的關注。

2.2. Public education programmes on rare diseases

From time to time, RDHK disseminates information on various rare diseases through different media channels. Last year's subjects included genetic counselling, glioblastoma, interstitial lung disease and pulmonary hypertension. In the hope of drawing public attention to rare disease issues and the rare disease community in an easy-to-understand and interesting way, information was delivered in the form of press releases, op-ed articles, video clips, media group interviews, patient sharing, comics, TV specials and so forth.

2.3. 真人圖書館

「真人圖書館」是藉著罕病患者和照顧者的親身分享，讓年青人更立體地認識各種罕見疾病、患者和照顧者面對的挑戰，以及如何與逆境周旋，啟發他們珍惜健康、尊重生命。去年罕盟總共為 13 間機構舉辦 25 場講座，受眾人數約為 1,520 人，對象包括中、小學生、大學醫護學生及宗教團體。

2.3. Human library

By sharing stories of rare disease patients and caregivers, the Human Library allows young people to have a well-rounded sense of various rare diseases, the challenges faced by patients and caregivers, and how to deal with adversity, inspiring them to cherish health and respect life. Last year, a total of 25 lectures were held for 13 organisations, addressing an audience of about 1,520 people, including primary and secondary school students, undergraduate healthcare students and religious groups.

2.4. 網頁

罕盟於 2021 年優化網頁介面後，2022 年的總點擊率高達 1,419,864 次，是前一年的 4.1 倍。此外，網頁於去年第 4 季增設「會員天地」和「義工園地」，除了讓會員和義工們分享他們參與罕盟活動後的感想外，也讓公眾人士從另一角度了解罕盟的工作。

2.4. Website

After optimising the website layout in 2021, the total click-through rate of RDHK's website in 2022 reached 1,419,864, which was 4.1 times that of the previous year. In addition, the "Members' Corner" and "Volunteer Zone" were added to the website in Q4 last year, allowing members and volunteers to share their feelings after participating in RDHK's activities, as well as allowing the public to understand the work of RDHK from another perspective.

2.5. 臉書

2022 年總共發佈了 95 則臉書貼文，總觸及人數為 1,468,614 人，較去年增加一倍多。最受歡迎的三則貼文是配合「2022 年世界罕病日」推出的短片系列：《給新一屆政府的話》、《藥物資助「關關難關關過！」》和《只要有決心，凡事可成真！》，單是這三則貼文的觸及人數已超過 57 萬人。

2.5. Facebook

A total of 95 Facebook posts were published in 2022, reaching a total of 1,468,614 people, more than double the number last year. The three most popular posts were from a short film series launched as a response to World Rare Disease Day 2022, namely "A Message to the New-term Government", "The Hurdles to Subsidised Drugs" and "Any dream can come true with determination". These three posts alone have reached more than 570,000 people.

2.6. 罕情

《罕情》於 2016 年 1 月創刊，原為半年刊，自 2018 年起改為季刊，每年 1、4、7、10 月出版，內容包括國際和本地的重大罕病政策、罕盟及其他罕病團體的各類活動、最新的罕病和罕藥資訊等，讀者對象包括政府官員、議員、醫護人員、罕病社群、罕盟的合作伙伴和社會大眾。為適應不同讀者的需要，刊物提供電子版和印刷版。去年每期的訂閱人數超過 2,100 人，公眾人士亦可到罕盟網頁下載閱讀。

2.6. *RareCare*

First published in January 2016, *RareCare* was originally a semi-annual publication. Since 2018 it has become a quarterly newsletter published in January, April, July and October every year, covering major international and local policies on rare diseases, various activities of RDHK and other rare disease groups, and the latest information on rare diseases, orphan drugs and so on. Readers include government officials, District or Legislative Council members, healthcare professionals, rare disease communities, RDHK's partners and the general public. To meet the needs of different readers, the newsletter is available in both electronic and print formats. Last year, each newsletter had more than 2,100 subscribers. The public is also welcome to download the newsletters from RDHK's website.

3 能力建設

3.1. 加強會員與社區的接觸

罕盟經常邀請不同會員擔任各類講座的分享嘉賓，以及鼓勵他們接受新聞媒體的訪問，此舉除有助提升公眾對罕見疾病和罕病社群的認知外，更重要的是讓會員有更多機會與外界接觸，從而增強個人的自信心、表達能力和溝通技巧。去年較活躍參與講座或願意接受訪問的基本會員共有十多名，希望日後我們可發掘更多「新星」，一起向公眾發放正能量。

3.2. 專題講座

去年罕盟主辦或與其他機構合辦了多個線上專題講座，主題包括：為何要儘快接種新冠疫苗、幼兒接種新冠疫苗安全性和成效、軟餐介紹、面肩肱型肌營養不良症及叢狀神經纖維瘤，總參加人數約 200 人。

3 Capacity building

3.1. Enhancing members' community engagement

RDHK often invites different members to be guest speakers of various talks and encourages them to be interviewed by the news media, which not only helps to raise public awareness of rare diseases and the rare disease community, but more importantly, allows members to have more opportunities to interact with the outside world, thereby enhancing self-confidence, presentation skills and communication skills. Last year, there were more than 10 full members who were active in participating in talks or willing to be interviewed. It is hoped that more "new stars" can be discovered in the future so that more positive vibes can be spread to the public together.

3.2. Subject talks

Last year, RDHK hosted or co-organised a number of online seminars on topics such as "COVID-19 Vaccination: Why need to get it soon?", "Safety and efficacy of vaccination for young children", "Introduction to soft meals", "Facioscapulohumeral muscular dystrophy" and "Neurofibromatosis type 1". The total number of participants was about 200.

3.3. 社交及消閒活動

2022 年下半年新冠疫情形勢緩和，罕盟開始舉辦各類消閒活動，以助會員舒展身心和促進人際交流。活動包括：「音樂旅途」~ 歌唱表演及音樂治療講座、和諧粉彩體驗工作坊、織織復織織工作坊、罕友與狗狗互動會、中秋燈迷會、花燈製作班、園藝體驗工作坊和健體及餐桌禮儀示範工作坊。上述活動均十分受歡迎，總參加人數超 250 人。

3.3. Social and leisure activities

In the second half of 2022, as the COVID-19 situation improved, RDHK began to organise various social and leisure activities to help members relax and promote interpersonal communication, including Music Journey~ singing performance and talk on music therapy, Harmony Pastel Workshop, Weaving Workshop, Therapy Dog Mingling Sessions, Mid-Autumn Festival Lantern Riddle Challenge, Lantern Making Class, Horticulture Experience Workshop, Workshops on Exercise Demonstration and Table Manners. All the activities were very well received, with a total of over 250 participants.

3.4. 義工培訓

在過去一年，總共有 34 名來自不同行業的人士登記成為罕盟義工，佔總登記義工人數的一半，可見越來越多人關注罕病社群。此外，罕盟繼續與香港大學護理學院、香港中文大學醫學院和香港大學李嘉誠醫學院合作，為 11 名護士學生和 4 名醫科學生提供培訓和實習

3.4. Volunteer training

A total of 34 people from different sectors registered as RDHK volunteers in the past year, accounting for half of the total number of registered volunteers, indicating that more and more people are paying attention to the rare disease community. In addition, RDHK continued to collaborate with the HKU School of Nursing, CUHK Faculty of Medicine and HKU Li Ka Shing Faculty of Medicine by offering training and internship opportuities to 11 nursing students and 4 medical students. They were required to help design,

機會，讓他們協助設計、策劃和參與各種類型的活動，例如興趣班、工作坊、講座、家訪、罕病資料搜集等。此外，罕盟透過參與「築夢同行義工計劃」，為來自主辦機構（義務工作發展局）和支持機構（匯豐銀行慈善基金）的六十多名員工提供培訓，使他們認識罕病患者和照顧者面對的處境，以同理心與服務對象溝通和相處，並一同體驗圓夢的過程。

plan and participate in various types of activities, such as interest classes, workshops, talks, home visits, collection of rare disease information, etc. Besides, participating in the "Share Your Dreams" Volunteer Project, RDHK provided training to over 60 staff members from the project organiser (Agency for Volunteer Service) and the supporting organisation (The Hongkong Bank Foundation) with a view to enabling them to understand the situation faced by patients and caregivers of rare diseases, communicate and get along with the service target with empathy, and experience the journey to achieving dreams together.

4 其他工作

4.1. 周年大會和理事選舉

罕盟第三屆會員周年大會和第二屆理事選舉於 2022 年 5 月 28 日舉行，當選的理事會成員名單請參閱本會務報告的附錄二。

4.2. 籌集各方資助與捐贈

去年在疫情最嚴峻時，罕盟獲得不同機構的資助與捐贈，包括各類防疫物資、日用品、超市禮券、捐款、免費送遞服務等，受惠會員人次超過 600 人。

4 Miscellaneous work

4.1. Annual general meeting & election of council members

The third Annual General Meeting and Election of the Second Council of RDHK were held on 28 May 2022. Please refer to Appendix II of this Annual Report for the list of elected Council members.

4.2. Soliciting monetary and in-kind donations

During the peak of the epidemic last year, RDHK received monetary and in-kind donations from different organisations, including various anti-epidemic supplies, daily necessities, supermarket vouchers, donations, free delivery services, etc., benefiting more than 600 members.

附錄一、會員及所涵蓋病種

截至 2022 年年底為止，會員總人數為 686，其中：

1. 普通會員（關注及支持罕病患者人士）數目：78
2. 基本會員（罕病患者或家屬）數目：608
   a) 個人會員數目：600
   b) 團體會員數目：8

Appendix I – Members and types of diseases

As at the end of 2022, the total number of members was 686, among which:

1. Number of ordinary members (those who care about and support rare disease patients): 78
2. Number of full members (rare disease patients or families): 608
   a) Number of individual members: 600
   b) Number of organisational members: 8

（按英文名稱排序）
(In alphabetical order)

- 香港平滑肌肉瘤基金會有限公司
  Hong Kong Leiomyosarcoma Foundation Limited
- 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 Association
- PNH 病人權益關注組
  PNH Concern Group
- 勉逆歷協進會有限公司
  Primary Immunodeficiency League Association Limited
- 香港視網膜病變協會
  Retina Hong Kong

會員所涵蓋的病種 *：192
Types of diseases among members *: 192

1. 10q26 Microdeletion Syndrome (Genetic Disorder)
2. 14q Deletion Syndrome (Genetic Disorder)
3. 16q11.2 & q22.2 Deletion Syndrome (Genetic Disorder)
4. 18q21.2x3 Deletion Syndrome (Genetic Disorder)
5. 1p36 Deletion Syndrome (Genetic Disorder)
6. 1q44 Deletion Syndrome (Genetic Disorder)
7. 3q23 & 3q25 Deletion Syndrome (Genetic Disorder)
8. 8p Inverted Duplication/Deletion Syndrome (Invdupdel(8p)) (Genetic Disorder)
9. Achondroplasia (軟骨發育不全症)
10. Acromegaly (肢端肥大症)
11. Albinism (白化病)
12. Alexander Disease (亞歷山大症)
13. Allan-Herndon-Dudley Syndrome (AHDS) / MCT8 Deficiency (MCT8 缺乏症)
14. Alstrome Syndrome (AS) (阿爾斯特倫綜合症)
15. Alveolar Soft Part Sarcoma (ASPS) (肺泡狀軟組織肉瘤)
16. Amyloidosis (AL) (類澱粉沉積症)
17. Amyotrophic Lateral Sclerosis (ALS) (肌萎縮性脊髓側索硬化症)
18. Angelman Syndrome (AS) (天使綜合症)
19. Anomalous origin of Left Coronary Artery from Pulmonary Artery (ALCAPA) (肺動脈左冠狀動脈異常症)
20. Anti-MOG Associated Encephalomyelitis (抗 MOG 相關腦脊髓炎)
21. Anti-NMDA Encephalitis (自體免疫性腦炎)
22. Apert Syndrome (亞伯氏症)
23. Aplastic Anemia (再生不良性貧血症)
24. Aromatic L-amino Acid Decarboxylase Deficiency (AADC) (芳香族 L- 胺基酸類脱羧基酶缺乏症)
25. Arthrogryposis Multiplex Congenita (AMC) (先天性多發性關節攣縮症)
26. ATAD3A Syndrome (Genetic Disorder)
27. ATR-X Thalassemia Syndrome (ATRX) (α地中海貧血 X- 連鎖智力障碍症)
28. Atypical Hemolytic Uremic Syndrome (aHUS) (非典型溶血性尿毒症候群)
29. Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (ARSACS) (Charlevoix-Saguenay 型隱性痙攣性共濟失調症)
30. Behcet's Disease (BD) (貝賽特氏症)
31. Bilateral Perisylvian Polymicrogyria (BPP) (雙側西耳維厄斯周圍多小腦回畸形症)
32. Burning Mouth Syndrome (口腔灼熱症侯群)
33. Cardiofaciocutaneous Syndrome (CFC) (CFC 症侯群)
34. Castleman Disease (CD) (卡斯爾曼氏症)
35. Central Core Disease (CCD) (肌中央軸空症)
36. Charcot-Marie-Tooth Disease (CMT) (腓骨肌萎縮症)
37. Chronic Hemolytic Anemia (慢性溶血性貧血症)
38. Chronic Inflammatory Demyelinating Polyneuropathy (CIPD) (慢性脱髓鞘性神經炎)
39. Cockayne Syndrome (柯凱因氏症侯群)
40. Coenzyme Q10 Deficiency-7 (Genetic Disorder)
41. Collagen XII Myopathy (膠原蛋白 XII 缺失肌肉萎縮症)
42. Cone-rod Dystrophy (視幹細胞營養不良症)
43. Congenital Glaucoma (先天性青光眼)
44. Congenital Insensitivity to Pain with Anhidrosis (CIPA) (先天性痛不敏感症合併無汗症)
45. Congenital Muscular Dystrophy (CMD) (先天性肌營養不良症)
46 Cornelia De Lange Syndrome (CdLS) (狄蘭氏症侯群)
47 Costello Syndrome (克斯提洛氏彈性蛋白質缺陷症)
48 Cri-Du-Chat Syndrome (貓哭症)
49 Crohn's Disease (克隆氏症)
50 CTNNB1 Syndrome (Genetic Disorder)
51 Cushing's Syndrome (CS) (庫欣氏症候群)
52 Dandy-Walker Syndrome (Dandy-Walker 氏症候群)
53 Ddx3x Syndrome (Genetic Disorder)
54 DiGeorge Syndrome (DGS) (迪喬治症侯群)
55 DNM1-L Syndrome (Genetic Disorder)
56 Dravet Syndrome (DS) (卓飛症候群)
57 Duchenne Muscular Dystrophy (DMD) (杜興氏肌肉營養不良症)
58 DYRK1A Syndrome (Genetic Disorder)
59 Ectodermal Dysplasia (外胚層發育不良症)
60 Ehlers-Danlos Syndrome (埃勒斯 - 當洛二氏症)
61 Eosinophilic Granulomatosis with Polyangiitis (EGPA) (過敏性肉芽腫血管炎)
62 Epidermolysis Bullosa (EB) (表皮溶解水皰症)
63 Epithelioid Hemangioendothelioma (EHE) (上皮樣血管內皮瘤)
64 Fabry Disease (法布瑞氏症)

65. Facioscapulohumeral Muscular Dystrophy (FSHD) - Type 1 (面肩肱型肌肉營養不良症第 1 型)
66. Familial Amyloid Polyneuropathy (FAP) (家族性澱粉樣物多發性神經病變)
67. Fibrodysplasia Ossificans Progressiva (FOP) (進行性肌肉骨化症)
68. Floating-Harbor Syndrome (FHS) (Floating-Harbor 綜合症)
69. Fragile X Syndrome (FXS) (X 染色體脆弱症)
70. Glioblastoma Multiforme (GBM) (膠質母細胞瘤)
71. Glutamate Receptor, Ionotropic, N-Methyl D-Aspartate 1 (GRIN 1)
72. Glycogen Storage Disease (GSD) - Type 1，G6PC Variants (肝醣儲積症第 1 型，G6PC 病變異)
73. Glycogen Storage Disease (GSD) - Type 2 (肝醣儲積症第 2 型) / Pompe Disease (龐貝氏症)
74. Glycogen Storage Disease (GSD) - Type 6 (肝醣儲積症第 6 型)
75. Hereditary Angiodema (HAE) (遺傳性血管性水腫症)
76. Hereditary Metabolic Disease (HMD) (遺傳性代謝病)
77. Hereditary Spastic Paraplegia (HSP) (遺傳性痙攣性下身麻痺症)
78. Hirschsprung's Disease (HD) (先天性巨結腸症)
79. Huntington's Disease (HD) (亨丁頓舞蹈症)
80. Ichthyosis (斑色魚鱗癬症)
81. Idiopathic Intracranial Hypertension (IIH) (特發性顱內壓增高症)
82. Idiopathic Pulmonary Fibrosis (IPF) (特發性肺纖維化)
83. IgG4-related Chronic Sclerosing Sialadenitis
84. Interstitial Pneumonia (間質性肺炎)
85. Jacobsen Syndrome (JS) (雅各森症候群)
86. Kallmann Syndrome (KS) (卡爾曼氏症候群)
87. KCNB1 Syndrome (Genetic Disorder)
88. KCNQ2 Syndrome (Genetic Disorder)
89. Kennedy Disease (甘迺迪氏症) / Spinal and Bulbar Muscular Atrophy (SBMA) (脊髓延髓性肌肉萎縮症)
90. KIF5C Syndrome (Genetic Disorder)
91. Klippel-Trénaunay Syndrome (KTS) (靜脈畸形骨肥大綜合症)
92. Langerhans Cell Histiocytosis (LCH) (蘭格罕細胞組織球增生症)
93 Larsen Syndrome (顎裂 - 先天性脫位症候群)
94 Leigh Syndrome (Leigh 氏症候群)
95 Leopard Syndrome (豹皮症候群)
96. Linear Scleroderma (線性硬皮症)
97. Lipid Storage Myopathy (LSM) (脂質儲積性肌病)
98. Lissencephaly (平腦症)
99. Maple Syrup Urine Disease (MSUD) (楓糖尿症)
100. McCune-Albright Syndrome (MAS) (纖維性骨失養症)
101. Medulloblastoma (髓母細胞瘤)
102. Melanomae / Eye Ciliary Body Tumor (眼睫狀體黑色素瘤)
103. Mesothelioma (間皮瘤)
104. Methylmalonic Acidemia (MMA) (甲基丙二酸血症)
105. Mitochondrial Cardiomyopathy (線粒體心肌病變)
106. Mitochondrial Disease (線粒體病)
107. Monoclonal Gammopathies (單克隆免疫球蛋白沉積病)
108. Motor Neuron Disease (MND) (運動神經元病)
109. Mowat-Wilson Syndrome (MWS)
110. Mucopolysaccharidoses (MPS) - Type 2 (黏多醣症第 2 型)
111. Mucopolysaccharidoses (MPS) - Type 3 (黏多醣症第 3 型)
112. Mucopolysaccharidoses (MPS) - Type 4 (黏多醣症第 4 型)
113. Mucopolysaccharidoses (MPS) - Type 4A (黏多醣症第 4A 型)
114. Mucopolysaccharidoses (MPS) - Type 6 (黏多醣症第 6 型)
115. Multiple Epiphyseal Dysplasia (MED) (多發性骨骺發育不全症)
116. Multiple Pituitary Hormom Deficiency (MPHD) / Anterior Pituitary Hypoplasia (腦下垂體發育不全症)
117. Multiple Sclerosis (MS) (多發性硬化症)
118. Multiple System Atrophy (MSA) -Pakinsonian Subtype (多重系統退化症 - 非典型巴金森症)
119. Myasthenia Gravis (MG) (重症肌無力症)
120. Nemaline Myopathy (桿狀體肌肉病變)
121. Neurofibromatosis (NF) - Type 1 (神經纖維瘤第 1 型)
122. Neurofibromatosis (NF) - Type 2 (神經纖維瘤第 2 型)
123. Neuromyelitis Optica (NMO) (視神經脊髓炎)
124. Nicolaides-Baraitser Syndrome (NCBRS)
125. Non-Infectious Non-Anterior (NINA) Uveitis (非感染性非前段葡萄膜炎)
126. Noonan Syndrome (努南氏症候群)
127. Olfactory Neuroblastoma (ONB) (嗅神經母細胞瘤)
128. Optic Neuropathy (ON) (視神經病變)
129. Osteogenesis Imperfecta (OI) (成骨不全症)
130. Paroxysmal Nocturnal Hemoglobinuria (PNH) (陣發性夜間血紅素尿症)
131. Pemphigus Vulgaris (天皰瘡)
132. Persistent Hyperplasia of Primary Vitreous (PHPV) (持續增生性原始玻璃體症)

133. Peutz-Jeghers Syndrome (PJS) (黑斑息肉症候群)
134. Phakomatosis Pigmentovascularis (PPV) (色素血管性母班症)
135. Phenylketonuria (PKU) (苯丙酮尿症)
136. Phosphatase and Tensin Homolog (PTEN) (磷酸酯酶與張力蛋白同源物)
137. Pierre Robin Syndrome (PRS) (皮爾羅賓氏症候群)
138. Prader-Willi Syndrome (PWS) (普瑞德威利症候群)
139. Primary Ciliary Dyskinesia (PCD) (先天性纖毛運動障礙症)
140. Primary Immunodeficiency (PID) (原發性免疫缺陷病)
141. Progressive Pseudorheumatoid Dysplasia (PPD) (進行性假性類風濕性骨發育不良症)
142. Propionic Acidemia (丙酸血症)
143. PTEN Hamartoma Tumor Syndrome (PHTS) (PTEN 過誤腫症)
144. Pulmonary Alveolar Proteinosis (PAP) (肺泡蛋白質沉著症)
145. Pulmonary Hypertension (肺動脈高壓)
146. Pyruvate Dehydrogenase Deficiency (丙酮酸鹽脱氫酵素缺乏症)
147. Relapsing Polychondritis (RP) (復發性多軟骨炎)
148. Retinitis Pigmentosa (RP) (視網膜色素病變)
149. Rett Syndrome (RTT) (雷特氏症)
150. Rhabdomyosarcoma (RMS) (橫紋肌肉瘤)
151. Rubinstein-Tabyi Syndrome (RTS) (Rubinstein-Taybi 症候群)
152. Russell-Silver Syndrome (RSS) (羅素 - 西弗氏症)
153. Sacral Chordoma (骶骨脊索瘤)
154 Schaaf-Yang Syndrome (SYS) (Schaaf-Yang 症候群)
155 Schinzel Giedion Syndrome (SGS) (Schinzel Giedion 綜合症)
156. SCN1A Syndrome (Genetic Disorder)
157. SCN2A Syndrome (Genetic Disorder)
158. SCN8A Syndrome (Genetic Disorder)
159. Smith-Magenis Syndrome (SMS) (史密斯 - 馬吉利氏症)
160. Spina Bifida (SB) (先天性脊椎裂症)
161. Spinal Muscular Atrophy (SMA) - Type 1 (脊髓肌肉萎縮症第 1 型)
162. Spinal Muscular Atrophy (SMA) - Type 2 (脊髓肌肉萎縮症第 2 型)
163. Spinal Muscular Atrophy (SMA) - Type 3 (脊髓肌肉萎縮症第 3 型)
164. Spinalcerebellar Ataxia (SCA) - Type 2 (小腦萎縮症第 2 型)
165. Spinalcerebellar Ataxia (SCA) - Type 3 (小腦萎縮症第 3 型)
166. Spondyloepiphyseal Dysplasia Congenita (SEDC) (先天性脊椎骨骺發育不全症)
167. Stiff Person Syndrome (SPS) (僵硬人症)
168. Succinic Semialdehyde Dehydrogenase Deficiency (SSADH) (琥珀酸半醛脱氫酶缺乏症)
169. Supernumerary Chromosome 8 Syndrome (Genetic Disorder)
170. Takayasu Arteritis (TA) (高安氏症)
171. Tetratricopeptide Repeat, Ankyrin Repeat and Coiled-coil containing 2 (TANC2)
172. TRIO and F-actin Binding Protein (TRIOBP) Syndrome (Genetic Disorder)
173. Tuberous Sclerosis Complex (TSC) (結節性硬化症)
174. Usher Syndrome (尤塞氏綜合症)
175. Vitamin D -Resistant Rickets (抗維生素 D 性佝僂病)
176. Waardenburg Syndrome (WS) (瓦登伯革氏症侯群)
177. Walker-Warburg Syndrome (WWS) (Walker-Warburg 綜合症)
178. WDR45 Syndrome (Genetic Disorder)
179. Wells Syndrome (韋爾斯綜合症)
180. West Syndrome-Infantile Spasms (韋氏症 - 嬰兒痙攣症)
181. Williams Syndrome (WS) (威廉氏症候群)
182. Wilson's Disease (WD) (威爾森氏症)
183. Wolf-Hirschhorn Syndrome (WHS) (沃夫 - 賀許宏氏症候群)
184. Wolfram Syndrome (Wolfram 症候群)
185. Worster-Drought Syndrome (WDS) (藍道 - 克萊富勒症候群)
186. WWOX Syndrome (Genetic Disorder)
187. Xeroderma Pigmentosum (XP) (著色性乾皮症)
188. X-linked Agammaglobulinemia (XLA) (X - 連鎖無丙種球蛋白血症)
189. X-linked Hypophosphatemic Rickets (XLH) (性聯遺傳型低磷酸佝僂症)
190. X-linked Myotubular Myopathy (XLMTM) (肌小管病變)
191. X-linked Retinoschisis (XLRS) (Genetic Disorder)
192. 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）
朱嘉豪先生（患者家屬，電訊企業總裁）
黃敏兒女士（註冊社工）

按英文姓氏排序

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 CHOW Kuen Tai Ronald (patient's family, nurse practitioner)
Mr Jack CHU (patient's family, telecommunications director)
Ms Amy WONG (registered social worker)

** In alphabetic order by surname*

附錄三、秘書處成員

何翠薇女士（營運總監）
賴家衛先生（業務發展主任）
陳淑雲女士（服務統籌經理及護理顧問）
黃桂婉女士（活動助理）

Appendix III – Staff of the Secretariat

Ms May HO (Director of Operations)
Mr Terry LAI (Business Development Officer)
Ms Amy CHAN (Service Coordination Manager & Nursing Advisor)
Ms Pat WONG (Programme Assistant)

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

Appendix IV – Advisors

(In alphabetical order by surname)
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

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

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 alphabetic order by surname*

鳴謝

（按英文名稱排序）
所有支持者、捐贈者和贊助商
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
香港賽馬會慈善信託基金

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
The Hong Kong Jockey Club Charities Trust

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

HKCSS 社聯 機構會員 AGENCY MEMBER