香港罕見疾病聯盟
RARE DISEASE HONG KONG

會務報告
ANNUAL REPORT
2021

機構使命：尊重差異、確保權利

核心業務：政策倡導、提昇認知、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 2021 年會務報告

(2022 年 2 月)

Rare Disease Hong Kong Limited Annual Report 2021

(February 2022)

2019 年至今，全球仍未能擺脱新冠病毒的威脅。每當香港市民以為可以本地確診清零和放寬防疫禁令之際，總是出現零星個案或小型社區爆發，今年年頭更迎來第五波疫情，抗疫之路似乎沒完沒了。不過，香港罕見疾病聯盟（下稱「罕盟」）早已適應了疫情下的新常態，以堅定不移的信念和靈活變通的方式去應對各種挑戰，確保各項核心業務維持理想的進展。

Since 2019 till now, COVID-19 has been raging throughout the world. Every time when Hong Kong was about to achieve zero local infection and relax the pandemic control measures, there came some sporadic cases or community-level outbreaks. The outbreak of the fifth wave of COVID-19 infections early this year has made the fight against the pandemic seemingly endless. Nevertheless, Rare Disease Hong Kong (RDHK) has already adapted to the new normal of the post-COVID-19 world. By overcoming challenges with unwavering faith and flexible approaches, RDHK was able to make the desired progress in different aspects of work.

1 政策倡議

1.1. 政府在近幾年的施政報告均有提出回應罕盟和其他病友社群訴求的新措施，例如加大安全網對罕病用藥的支援、優化安全網資產審查機制、建立病人資料庫、展開公眾教育、增加人手、支持科研和臨床研究等。為確保應對罕病的措施能落實執行，罕盟不時主動聯絡食物及衛生局（食衛局）和醫院管理局（醫管局），並於 2021 年 5 月透過視像會議與相關官員會面，了解有關罕病措施的落實進度。

1 Policy advocacy

1.1. In recent years, the Government has proposed new measures in the Policy Address to respond to the demands of RDHK and other patient groups, for example, enhancing the orphan drug coverage and means test mechanism under the Safety Net, developing a patient database, providing public education, increasing manpower and supporting scientific and clinical research, etc. To ensure that those measures dealing with rare diseases are properly implemented, RDHK has taken the initiative to contact the Food and Health Bureau (FHB) and the Hospital Authority (HA) from time to time. In May 2021, the relevant officials were invited to update RDHK on the implementation progress of the measures for rare diseases in an online meeting.

1.2. 有感於政府過往只是零碎地提出一些針對罕病的新措施，欠缺長遠規劃，罕盟於去年 9 月就《2021 年施政報告》公眾諮詢向特首提交意見書，促請政府制定具前瞻性的應對罕病策略藍圖。意見書的具體建議包括：

- 政府牽頭推動官、商、民協作，設立「罕病策略督導委員會」；
- 訂立罕病定義，編製罕病名錄；
- 加快引進新藥程序；
- 廣納「風險共擔」方案，為罕藥納入安全網的審批制度訂下明確指引；
- 加強建立罕病資料庫；
- 擴大初生嬰兒罕見病篩查範圍；及
- 推展一站式覆診，為建立「罕病卓越中心」累積經驗。

1.2. Thinking that the Government in previous years only proposed some measures on rare diseases on a piecemeal basis without long-term planning, in September last year, RDHK responded to the 2021 Policy Address Public Consultation with a submission, where the Government was urged to adopt a macro perspective and foreword-thinking approach to policy development. The following recommendations were made in the submission:

- To set up the "Steering Committee on Strategy for Rare Diseases" with tripartite collaboration among the government, the business sector and the community;
- To define "rare diseases" and compile the rare disease list;
- To speed up the process of introduction of new drugs;
- To adopt "risk sharing" arrangements, and to speed up the inclusion of orphan drugs in the Safety Net by providing clear guidelines on the review mechanism;
- To enrich the rare disease database;
- To expand the scope of newborn screening for rare diseases; and
- To promote and develop one-stop consultation service with a view to gaining experience in establishing the "Centre of Excellence for Rare Diseases".

1.3. 雖然政府在 2018 年 9 月已引入可治療脊髓性肌肉萎縮症（SMA）的藥物（nusinersen），但之後只有十多名年幼患者和一名成年患者獲得治療，其餘數十名成年患者卻只有等待的份兒。經過罕盟兩年來的倡導，官商民各方協作，最終由藥廠透過醫管局於 2021 年 11 月起安排二十多名兒童和成年 I、II 型 SMA 患者免費試用口服新藥 (risdiplam) 至 2022 年年底。

1.3. Although the Government introduced a new drug (nusinersen) for treating spinal muscular dystrophy (SMA) in September 2018, only a dozen or so young patients and one adult patient have since been treated, leaving the remaining dozens of adult patients waiting in despair. After two years of advocacy by RDHK, and in collaboration with HA and the pharmaceutical company, more than 20 pediatric and adult patients with SMA types I & II were offered a new oral drug (risdiplam) for free from November 2021 to the end of 2022.

1.4. 鑑於醫管局只會對參與「Risdiplam 恩恤用藥計劃」（以下簡稱「用藥計劃」）的 SMA 患者進行行動能力指標調查，而沒有評估生命質素成效等指標，故罕盟在 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為期一年的追蹤調查，以收集參與或沒有參與「用藥計劃」的 SMA 患者生命質量的數據。這些補充數據有助醫管局日後審議是否將 risdiplam 納入《藥物名冊》和安全網時更全面地判斷藥物對病人產生的臨床和生命質素作用。罕盟亦希望以這項調查為先導，促進透過善用「風險共擔」藥物試用的過程，全面收集臨床和真實世界的數據，為日後基於實證加快審批其他罕病藥物提供參照模式。

1.4. Knowing that HA will only assess the motor function in those SMA patients participating in the "Risdiplam Compassionate Use Programme" (CUP) without measuring their quality of life, RDHK commissioned the Jockey Club School of Public Health and Primary Care, CUHK in November 2021 to conduct a one-year research study to collect data on the quality of life of SMA patients who participated in or did not participate in the CUP. These supplementary data can help HA to make a more comprehensive assessment of both clinical and quality of life outcomes of risdiplam when considering whether to include it in the Drug Formulary and Safety Net. It is hoped that this research study can serve as an example for the authorities concerned, so that more performance-based risk-sharing schemes can be adopted in the future to collect clinical data and real-world evidence, which will reduce the approval lag of orphan drugs.

1.5. 自 2019 年初「撒瑪利亞基金」和「關愛基金醫療援助項目」的經濟審查機制獲優化以來，罕盟一直關注新機制的成效。因此，罕盟於 2021 年 2 至 3 月期間進行「昂貴罕病藥物藥費分擔機制執行情況」調查，訪問了 16 名於 2020 年成功申請上述藥物資助計劃的申請人，蒐集他們對新機制的申請資格、申請流程、資助金額和各項細節安排的意見，然後與食衛局分享調查結果，並提出相關改善建議和要求當局持續優化機制運作。

1.5. Since the means test mechanism of the Samaritan Fund (SF) and Community Care Fund (CCF) was optimised in early 2019, RDHK has been concerned about the effectiveness of the new mechanism, and therefore conducted a survey on the implementation of the drug cost-sharing mechanism for expensive orphan drugs between February and March 2021. 16 successful applicants who applied for SF or CCF in 2020 were interviewed about their opinions on the eligibility requirements, application procedures, amount of subsidy and every detailed arrangement of the new mechanism. The findings were submitted to FHB together with suggestions for improvement and request for continuous optimisation of the operation of the mechanism.

1.6. 為評估醫管局轄下的「智友站」在提升大眾對特定疾病認知的成效，罕盟於 2021 年 5 至 6 月期間對二百多名罕病、癌病或長期病患者和照顧者進行線上問卷調查，並把調查結果和改善建議提交到食衛局和醫管局作參考。

1.6. To assess the effectiveness of "Smart Patient Website" which is under the auspices of HA in raising public awareness of specific diseases, RDHK conducted an online survey of more than 200 patients and caregivers with rare diseases, cancers or chronic diseases between May and June 2021. The findings and suggestions for improvement were submitted to FHB and HA for reference.

1.7. 去年罕盟共舉辦了兩場圓桌會議，並有幸邀得各方專家、學者、罕病患者和不同持份者參與，集思廣益。於 2021 年 2 月 28 日舉行的公開圓桌會議主題為「如何改善香港罕藥可及性」，罕盟參詳各與會者的觀點和意見後，撰寫《改善香港罕病藥物可及性建議書》，於 4

1.7. Honoured by the participation of experts from all sides, scholars, patients with rare diseases and different stakeholders, RDHK organised two roundtables last year. The theme of the open roundtable held on 28 February 2021 was "How to improve orphan drug accessibility in Hong Kong". After reviewing the views and opinions of the participants, RDHK submitted in mid-April a proposal on "Ways to improve access to orphan drugs in Hong Kong" to the Chief Executive and FHB for consideration. Then, a press release with

月中呈交予特首及食衞局考慮，又於 5 月把建議書連同新聞稿一起發放，引起公眾對此議題的關注和討論。另一場於 12 月 11 日舉行的閉門圓桌會議旨在探討把龐貝氏症納入香港初生嬰兒代謝病篩查服務的可行性。經過深入探討，罕盟認為仍需收集更多數據，方可向政府提出能讓病人真正受惠的可行建議。

the proposal attached was sent out in May in order to arouse public awareness and provoke discussion on the topic. The closed-door roundtable held on 11 December aimed to explore the feasibility of including Pompe disease in the scope of the Newborn Screening Programme for Inborn Errors of Metabolism in Hong Kong. After in-depth discussions, RDHK decided that more data would be required before feasible and pragmatic recommendations could be made to the Government.

1.8. 罕盟就罕病藥物事宜曾多番致函醫管局，促請當局釐清納入「不常見疾病」及為極度昂貴藥物提供資助的準則，以進一步提升市民對罕見疾病的認識和理解，以及要求儘快通過把「氯苯唑酸」（tafamidis）治療轉甲狀腺素蛋白澱粉樣心肌病變（transthyretin amyloid cardiomyopathy）適應症部分和「艾替班特」（icatibant injection）治療遺傳性血管性水腫（hereditary angioedema）的適應症納入《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。

1.8. RDHK wrote to HA at different times with concerns about orphan drugs. The authority concerned was urged to clarify the definition of "uncommon disorders" and the criteria of granting subsidies for ultra-expensive drugs so as to further enhance the public awareness and understanding of rare diseases; and to approve the listing of another indication of "tafamidis" for the treatment of transthyretin amyloid cardiomyopathy and the indication of "icatibant injection" for the treatment of hereditary angioedema on the Drug Formulary with safety net coverage so that the relevant patients can receive appropriate treatment at an early stage.

1.9. 得悉聯合國大會在 2021 年 12 月通過罕病決議案後，罕盟隨即發新聞稿應和，並促請政府制訂和落實與國際接軌的罕病策略和規劃，承擔及處理香港的罕病問題。

1.9. Learning that the United Nations General Assembly formally adopted a resolution on addressing the challenges of persons living with rare disease and their families in December 2021, RDHK immediately stroke a chord by sending out a press release urging the Government to formulate and implement strategies and plans for rare diseases that are geared to the international standards, and to undertake the responsibility of dealing with rare diseases in Hong Kong.

2 提昇認知

2.1. 汲取了近年在疫情陰霾下舉辦活動的經驗，罕盟於去年 9 月 25 日以線上線下模式同步舉行以「照顧差異 • 實踐公平」為主題的「罕病研討會 2021」，共吸引逾四百名來自不同界別的人士參與活動，人數打破歷年紀錄。一眾來自醫療界和學術界的嘉賓講者除了各自分享他們的見解或研究成果外，亦回應台下觀眾的提問，共同探討罕病患者對公共醫療服務需要的特殊性。

2 Public awareness

2.1. Drawing on the experience of holding activities during the pandemic in recent years, RDHK held the Rare Disease Symposium 2021 with the theme of "Tackling Differences and Implementing Equity" in hybrid mode on September 25 last year. A record number of more than 400 people from different sectors participated in the event. Apart from sharing their insights or research findings, the guest speakers from the healthcare sector and academia also responded to questions from the audience, and to explore the special needs of rare disease patients for public healthcare services.

2.2. 為提升大眾對不同罕見疾病和罕見癌症的認識，罕盟分別於去年第二、三、四季推出關於特發性肺纖維化、間質性肺病、肺動脈高壓、罕見癌症和惡性胸膜間皮瘤的公眾教育計劃，主要透過臉書專頁發佈圖片和影片帖文，並邀請社交媒體意見領袖分享，觸及人數接近 27 萬。同時，罕盟亦推出專題網頁 (https://www.knowingild.org.hk)，以介紹多種與肺纖維化相關的疾病資訊，藉此提醒公眾注意身體發出的警號。此外，罕盟與香港復康會社區復康網絡合辦「你讚我捐 • 關懷肺纖及慢阻肺患者」活動，在臉書平台引起超過 6 萬人對相關疾病和患者的關注。

2.2. To raise public awareness of different rare diseases and rare cancers, RDHK launched public awareness campaigns on idiopathic pulmonary fibrosis (IPF), interstitial lung disease, pulmonary hypertension, rare cancers and malignant pleural mesothelioma in the second, third and fourth quarters of last year. Messages were conveyed to the public through photo and video posts on RDHK Facebook page. Having been shared by some social media KOLs, those Facebook posts reached nearly 270,000 people. At the same time, RDHK launched a dedicated webpage (https://www.knowingild.org.hk) to introduce a variety of diseases related to pulmonary fibrosis, thereby reminding the public to be alert to the warning signs from the body. In addition, RDHK and the Community Rehabilitation Network, Hong Kong Society for Rehabilitation jointly organised a Facebook event called "You Like, I Donate - Care for patients with IPF and chronic obstructive pulmonary disease". Aiming to arouse public awareness of the relevant diseases and support for the patients, the event attracted the attention of more than 60,000 people.

2.3. 罕盟自 2018 年起推出「罕見疾病真人圖書館」，主要對象為中、小學和大專院校學生。旨在透過罕病患者的親身分享和即時交流，提升年青人對罕見疾病和罕病群體的認識，並鼓勵他們逆境自強和共建關愛共融的社會。2021 年罕盟為 15 間院校舉行 21 場互動講座，受眾人數約為 3,500 人。

2.3. The Human Library Programme has been launched since 2018 for students from primary, secondary and tertiary schools. It aims to enhance young people's understanding of rare diseases and rare disease community through storytelling and immediate interaction with the rare disease patients. Students are also encouraged to push through adversity during challenging times and build a caring and inclusive society. A total of 21 interactive talks for 15 schools were held in 2021, addressing an audience of about 3,500 people.

2.4. 在勞工及福利局的撥款資助下，罕盟得以優化網頁介面和內容。經過年多的籌備，新網頁終於在去年 4 月面世，可提供更多資訊，包括介紹罕病基本資料的「罕病小百科」、為不同罕病小冊子或書籍而設的「罕病小書架」、以多媒體方式發放的罕盟最新動態等。截至 2021 年年底，新網頁的點擊次數已達 347,416 次。

2.4. With funding from the Labour and Welfare Bureau, RDHK was able to optimise the layout and content of its website. After over a year of preparation, the new website was finally launched in April last year with more information such as the "Rare Disease Wiki" which introduces the basic information of rare diseases, the "Rare Diseases Bookshelf" for different rare disease booklets or books, the latest news about RDHK which are presented in multimedia formats, and so on. As at the end of 2021, the new website received 347,416 hits.

2.5. 去年機構臉書帖文總共觸及 661,734 人，與 2020 年比較，增幅超過 50%。

2.5. Last year, RDHK's Facebook posts reached a total of 661,734 people, an increase of over 50% compared with 2020.

2.6. 《罕情》是與本地罕病群體息息相關的季刊。內容除涉獵國際和本地的重大罕病政策外，也報導香港罕病團體的各類活動，圖文並茂，是政府部門、服務機構、病人團體和社會大眾認識及了解罕病相關資訊的重要媒介。為適應不同讀者的需要，刊物提供電子版和印刷版。

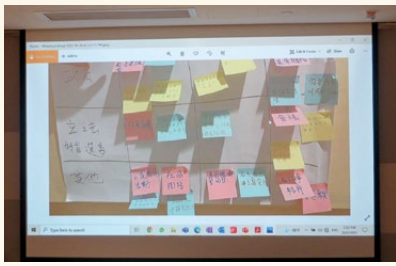
2.6. *RareCare* is a quarterly newsletter closely related to the local rare disease community. It covers not only major international and local rare disease policies, but also various activities of Hong Kong rare disease groups with numerous illustrations. *RareCare* has become an important channel for government departments, service organisations, patient groups and the public to learn about and understand information relating to rare diseases. To cater for the needs of different readers, the publication is available in both electronic and print formats.

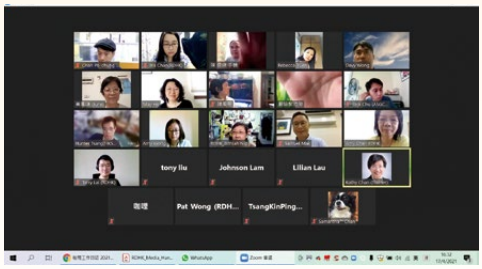
3 能力建設

3.1. 罕盟於 2021 年上半年舉辦了兩場罕病領袖培訓工作坊，主題分別為「應對傳媒訪問」和「游說工作多面體」，旨在提升參加者面對傳媒訪問、進行倡導和游說工作時所需的技巧。

3 Capacity building

3.1. Two training workshops for leaders of rare disease groups were run in the first half of 2021. The themes were "Media training" and "Lobbying skills" respectively. Those workshops aimed to enhance participants' media interview, advocacy and lobbying skills.



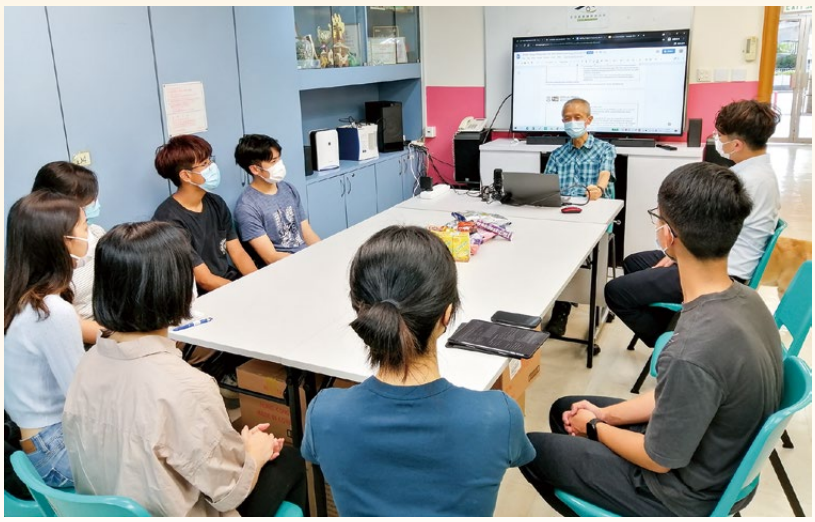


3.2. 因應會員和大眾的不同需要，罕盟以線上、實體或線上加實體混合形式舉辦多元化的活動，例如罕病專題講座（主題包括：顏肩肱型肌肉萎縮症、神經纖維瘤症、神經肌肉疾病病人名冊及疫情應對、TTR 類澱粉沉積症所引起的心肌病變、個人化醫療和基因檢測）、硬地滾球體驗日、會員迎新日、電影欣賞、大笑瑜伽、沙畫班、繪畫班等，參加人數合共約 300 人。

3.2. In response to the different needs of members and the public, RDHK organised a variety of activities in online, offline or hybrid modes, for instance, subject talks on rare diseases (topics included: facioscapulohumeral muscular dystrophy, neurofibromatosis, neuromuscular disease patient registry and coping during a pandemic, transthyretin cardiomyopathy, precise medicine and genetic testing), boccia experience day, member orientation day, movie appreciation, laughter yoga, sand painting classes, painting classes, and so on. The total number of participants was about 300.

3.3. 為增強大眾對罕病群體的了解和支持，罕盟不時與不同機構合辦義工培訓活動。罕盟去年支持香港大學護理學院推出「本地服務學習夥伴計劃 2020-21 - 豐富生命學習」，於 6 至 8 月期間，讓 7 名護士學生在罕盟護理顧問的帶領下，參與機構不同活動和有機會親身接觸罕病患者，護士學生們表示計劃有助提高其溝通技巧和加深了解護士與病人的相互關係，並感謝有機會實習為患者做身體評估和詢問病歷，以及認識罕病患者在日常生活中可能面臨的困難，為日後的工作做好準備。此外，罕盟與義務工作發展局協辦為期 18 個月的「築夢同行」義工計劃，旨在讓一眾香港匯豐銀行職員以義工身份陪同患有罕病或殘障兒童達成心願及改善其生活環境，相關義工培訓於去年 12 月展開。

3.3. To enhance public understanding of and support for the rare disease community, RDHK co-organises volunteer training programmes with various organisations from time to time. Last year, RDHK became a supporting organisation of the "Local Service-learning Partnership Programme 2020-21 – Life Enrichment Learning" rolled out by the University of Hong Kong School of Nursing. Mentored by RDHK's Nursing Advisor, 7 student nurses took part in different activities and met the rare disease patients in person from June to August. Upon completion of the Programme, they reflected that they had acquired better communication skills and understanding of the nurse-patient relationship, and were grateful for having the opportunity to practise physical assessment and history taking, and realise the difficulties that rare disease patients may face in daily life, which made them better equipped for the future work. Besides, RDHK co-organised the 18-month "Share Your Dreams" Volunteer Project with the Agency for Volunteer Service in the hope of involving a group of HSBC staff serving as volunteers in the process of fulfilling the dreams of children with rare diseases or disabilities and improving their living environment. The relevant volunteer training began in December last year.



4 其他工作

4.1. 罕盟第二屆會員周年大會於 2021 年 6 月 12 日舉行，除了恆常的議程外，會後還安排了一個護理訊息分享會，由香港大學護理學院護士學生主講。

4.2. 鑑於罕盟經費的來源有一定比例來自商界的贊助和撥款，故罕盟理事會通過《與商界協作的實務守則》，以加強誠信管理和內部監控，確保所有來自商界的金錢、實物和服務提供均遵循法律及符合操守，並讓所有罕盟的工作人員和與罕盟協作的商業機構有所依從。

4.3. 去年罕盟得到多間機構的慷慨支持，除了成為不同籌款活動的受惠機構外，更獲贈口罩、福袋和無酒精消毒顯紙巾等。罕盟已把相關物資發送給逾 200 個有需要的會員。

4 Miscellaneous work

4.1. The second Annual General Meeting of RDHK was held on 12 June 2021. In addition to the usual agenda, a sharing session on nursing tips conducted by nurse students from HKU School of Nursing was arranged after the meeting.

4.2. As a certain proportion of RDHK's funding comes from sponsorships and grants from the business sector, the RDHK Council adopted *the Code of Practice for Collaboration with the Business Sector* with a view to reinforcing integrity management and internal controls; ensuring that all the money, goods and services from the business sector are lawful and ethical; and providing practical guidance to all RDHK staff and commercial organisations collaborating with RDHK.

4.3. Big thanks to the generous support from a number of organisations last year, RDHK became a beneficiary of different fundraising activities, and also received donations of face masks, lucky bags, alcohol-free disinfectant wipes and so on. All the supplies were distributed to more than 200 members in need.

附錄一、會員及所涵蓋病種

截至 2021 年年底為止，會員總人數為 533，其中：

1. 普通會員（關注及支持罕病患者人士）數目： 53
2. 基本會員（罕病患者或家屬）數目： 480
   a) 個人會員數目：472
   b) 團體會員數目：8

Appendix I – Members and types of diseases

As at the end of 2021, the total number of members was 533, among which:

1. Number of ordinary members (those who care about and support rare disease patients): 53
2. Number of full members (rare disease patients or families): 480
   a) Number of individual members: 472
   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 Patient Group
- 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 * : 171
Types of diseases among members * : 171

1. 10q26 Microdeletion Syndrome
2. 14q (genetic disorder)
3. 16q11.2-q22.2 (genetic disorder)
4. 18q21.2 x 3 (genetic disorder)
5. 1p36 Deletion Syndrome
6. 1q44 deletion (genetic disorder)
7. 22q13.33 Microdeletion Syndrome
8. 3q23-3q25 Deletion (genetic disorder)
9. Achondroplasia (軟骨發育不全症)
10. Acromegaly (肢端肥大症 (巨人症))
11. Albinism (白化病)
12. Alexander Disease (亞歷山大病)
13. Alstrom Syndrome (Alstrom 氏症候群)
14. Alveolar Soft Part Sarcoma (肺泡狀軟組織肉瘤)
15. Amyloidosis (類澱粉沉積症)
16. Amyotrophic lateral sclerosis (肌萎縮性脊髓側索硬化症)
17. Angelman Syndrome (天使綜合症)
18. Anti-MOG associated encephalomyelitis
19. Anti-NMDA Encephalitis (自體免疫性腦炎)
20. Apert Syndrome (亞伯氏症)
21. Aplastic Anemia (再生不良性貧血)
22. Aromatic L- amino Acid Decarboxylase Deficiency (芳香族 L - 胺基酸類脱羧基酶缺乏症)
23. Arthrogryposis Multiplex Congenita (先天性多發性關節攣縮症)
24. ATRX Thalassemia Syndrome (α 型海洋性貧血 / 智能發展遲緩)
25. Atypical Hemolytic Uremic Syndrome (非典型性尿毒溶血症候群)
26. Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS)
27. Behcet's Disease (貝賽特氏症)
28. Bilateral Perisylvian Polymicrogyria (雙側西耳維厄斯周圍多小腦回畸形)
29. Burning Mouth Syndrome (口腔灼熱症候群)
30. Cardiofaciocutaneous Syndrome (CFC 綜合症)
31. Central Core Disease (CCD) (肌中央軸空病)
32. Charcot Marie Tooth Disease (進行性神經性腓骨萎縮症)
33. Chronic Hemolytic Anemia (慢性溶血性貧血)
34. Cockayne Syndrome (柯凱因氏症候群)
35. Collagen XII Myopathy (骨膠原蛋白第 12 基因引致肌肉病變)
36. Cone-rod Dystrophy (視幹細胞營養不良)
37. Congenital Glaucoma (先天性青光眼)
38. Congenital Insensitivity to Pain with Anhidrosis, CIPS (先天性痛不敏感症合併無汗症)
39. Congenital Muscular Dystrophy (先天性肌失養症)
40. COQ10D7 (genetic disorder)
41. Costello Syndrome (克斯提洛氏彈性蛋白質缺陷症)
42. Cri Du Chat Syndrome (貓哭症) / Chromosome 5p Deletion Syndrome
43. Crohn's Disease (克隆氏症)
44. CTNNB1 Syndrome (CTNNB1 綜合症)
45. Cushing's Syndrome (庫欣氏症候群)
46. Dandy Walker Syndrome (Dandy-Walker 綜合征)
47. DDx3x (genetic disorder) (DDx3x 基因突變)
48. De Lange Syndrome (狄蘭吉氏症候群)
49. DiGeorge Syndrome (迪喬治症候群)
50. DNM1-L (genetic disorder)
51. Duchenne Muscular Dystrophy (杜興氏肌肉營養不良症)
52. Ectodermal Dysplasia (外胚層發育不良症)
53. Ehlers-Danlos Syndrome (埃勒斯 - 當洛二氏症候群)
54. Eosinophilic Granulomatosis with Polyangiitis (過敏性肉芽腫血管炎)
55. Epidermolysis Bullosa (表皮溶解水皰症)
56. Fabry Disease (法布瑞氏症)

57. Facioscapulohumeral Muscular Dystrophy (面肩肱型肌肉營養不良症)
58. Familial Amyloid Polyneuropathy (家族性澱粉樣物多發性神經病變)
59. Fibrodysplasia Ossificans Progressiva (進行性肌肉骨化症)
60. Floating-Harbor Syndrome
61. Fragile X Syndrome (X 染色體脆弱症)
62. Glutamate Receptor, Ionotropic, N-Methyl D-Aspartate 1 (GRIN 1)
63. Glycogen Storage Disease - Type 1 ，G6PC variants (肝醣儲積症第 1 型，G6PC 病變異)
64. Glycogen Storage Disease - Type 2 (肝醣儲積症第 2 型) / Pompe Disease (龐貝氏症)
65. Glycogen Storage Disease - Type 6 (肝醣儲積症第 6 型)
66. Hereditary Angiodema (遺傳性血管性水腫)
67. Hereditary Spastic Paraplegia (遺傳性痙攣性下身麻痺)
68. Huntington's Disease (亨丁頓舞蹈症)
69. Ichthyosis (斑色魚鱗癬)
70. Idiopathic Intracranial Hypertension (特發性顱內壓增高)
71. Idiopathic Pulmonary Fibrosis (特發性肺纖維化)
72. Idiopathic thrombocytopenic purpura (ITP) (自發性血小板缺乏紫斑症)
73. IgG4-Related Chronic Sclerosing Sialadenitis
74. Invdupdel (8p) (genetic disorder)
75. Jacobsen Syndrome (11q 缺失症候群)
76. Kallmann Syndrome (卡爾曼氏綜合症)
77. KCNB1 (genetic disorder)
78. KCNQ2 (genetic disorder)
79. Kennedy Disease (甘迺迪氏症)
80. KIF5C (genetic disorder)
81. Klippel-Trénaunay Syndrome (靜脈畸形骨肥大綜合症)
82. Langerhans Cell Histiocytosis (蘭格罕細胞組織球增生症)
83. Larsen Syndrome (顎裂 - 先天性脫位症候群)
84. Leigh Syndrome (萊氏症)
85. Leopard Syndrome (Leopard 症侯群)
86. Linear Scleroderma (線性硬皮症)
87. Lipid Storage Myopathy (脂質沉積性肌病)
88. Lissencephaly (平腦症)
89. Maple Syrup Urine Disease (楓糖尿症)
90. McCune Albright Syndrome (纖維性骨失養症)
91. Medulloblastoma (髓母細胞瘤)
92. Methylmalonic Acidemia (甲基丙二酸血症)
93. Mitochondrial Cardiomyopathy (線粒體心肌病變)
94. Mitochondrial Disease (線粒體病)
95. Monoclonal Gammopathies (單克隆免疫球蛋白沉積病)
96. Motor Neuron Disease (運動神經元病)
97. Mowat-Wilson Syndrome
98. Mucopolysaccharidoses Type 2 (黏多醣症第 2 型)
99. Mucopolysaccharidoses Type 3 (黏多醣症第 3 型)
100. Mucopolysaccharidoses Type 4A (黏多醣症第 4A 型)
101. Mucopolysaccharidoses Type 6 (黏多醣症第 6 型)
102. Multiple Epiphyseal Dysplasia (多發性骨骺發育不全)
103. Multiple Pituitary Hormone Deficiency Anterior Pituitary Hypoplasia (腦下垂體發育不全)
104. Multiple Sclerosis (多發性硬化症)
105. Multiple System Atrophy-Parkinsonian Subtype (多重系統退化症 - 非典型巴金森症)
106. Myasthenia Gravis (重症肌無力症)
107. Nemaline Myopathy (桿狀體肌肉病變)
108. Neurofibromatosis Type 1 (神經纖維瘤第 1 型)
109. Neurofibromatosis Type 2 (神經纖維瘤第 2 型)
110. Neuromyelitis Optica (視神經脊髓炎)
111. Nicolaides-Baraitser Syndrome
112. Non-infectious non-anterior (NINA) uveitis (非感染性非前段葡萄膜炎)
113. Olfactory Neuroblastoma (嗅神經母細胞瘤)
114. Osteogenesis Imperfecta (成骨不全症)
115. Paroxysmal Nocturnal Hemoglobinuria (陣發性夜間血紅素尿症)
116. Pemphigus Vulgaris (天皰瘡)

117. Peutz-Jeghers Syndrome (黑斑息肉症候群)
118. Phakomatosis Pigmentovacularis (色素血管性母班症)
119. Phenylketonuria (苯丙酮尿症)
120. Phosphatase and tensin homolog (Pten) (genetic disorder) (磷酸酯酶與張力蛋白同源物)(基因病變)
121. Pierre Robin Syndrome (皮爾羅賓氏症)
122. Prader Willi Syndrome (小胖威利症)
123. Primary amyloidosis (原發型類澱粉沉積症)
124. Primary Ciliary Dyskinesia (先天性纖毛運動障礙)
125. Primary Immunodeficiency (原發性免疫缺陷病)
126. Primary Pulmonary Arterial Hypertension (肺動脈高壓)
127. Progressive Pseudorheumatoid Dysplasia (進行性假性類風濕性骨發育不良)
128. Propionic Acidemia (丙酸血症)
129. Pulmonary Alveolar Proteinosis (肺泡蛋白質沉著症)
130. Pyruvate Dehydrogenase Deficiency (丙酮酸鹽脱氫酵素缺乏症)
131. Relapsing Polychondritis (復發性多軟骨炎)
132. Retinitis Pigmentosa (視網膜色素病變)
133. Rett Syndrome (雷特氏症)
134. Rhabdomyosarcoma (橫紋肌肉瘤)
135. Rubinstein-Tabyi Syndrome (Rubinstein-Tabyi 氏症候群)
136. Russell-Silver Syndrome (羅素 - 西弗氏症)
137. Schaaf-Yang Syndrome (Schaaf-Yang 綜合症)
138. SCN1A (genetic disorder)
139. SCN2A (genetic disorder)
140. SCN8A (genetic disorder)
141. Smith-Magenis Syndrome (史密斯 - 馬吉利氏綜合症)
142. Spina Bifida (先天性脊椎裂)
143. Spinal Muscular Atrophy Type 1 (脊髓肌肉萎縮症第 1 型)
144. Spinal Muscular Atrophy Type 2 (脊髓肌肉萎縮症第 2 型)
145. Spinal Muscular Atrophy Type 3 (脊髓肌肉萎縮症第 3 型)
146. Spinalcerebellar Ataxia Type 2 (小腦萎縮症第 2 型)
147. Spinalcerebellar Ataxia Type 3 (小腦萎縮症第 3 型)
148. Spondyloepiphyseal Dysplasia Congenita (先天性脊椎骨骺發育不全)
149. Stiff Person Syndrome (僵硬人症候群)
150. Succinic Semialdehyde Dehydrogenase Deficiency (SSADH 缺乏症)
151. Supernumerary Chromosome 8
152. Takayasu Arteritis (高安氏症)
153. TRIOBP (genetic disorder) (波形蛋白) (基因病變)
154. Tuberous Sclerosis Complex (結節性硬化症)
155. Usher Syndrome (尤塞氏綜合症)
156. Vitamin D Resistant Rickets (抗維生素 D 性佝僂病)
157. Waardenburg Syndrome (瓦登伯革氏症候群)
158. Walker-Warburg Syndrome
159. WDR45 (genetic disorder)
160. Wells Syndrome
161. West Syndrome-Infantile Spasms (韋氏症候群)
162. Williams Syndrome (威廉氏症候群)
163. Wilson's Disease (威爾森氏症)
164. Wolfram Syndrome (Wolfram 氏症候群)
165. WWOX gene deletion (genetic disorder)
166. Xeroderma Pigmentosum (著色性乾皮症)
167. X-linked Agammagluolinmia (X- 連鎖無丙種球蛋白血癥)
168. X-linked Hypophosphatemic Rickets (性聯遺傳型低磷酸佝僂症)
169. X-linked Retinoschisis (genetic disorder)
170. X-linked Thrombocytopenia (genetic disorder)
171. XLMTM Myotubular Myopathy (肌小管病變)

* 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：杜勤創先生（患者家屬，傳訊經理）
義務秘書：黃婉冰女士（患者，退休校長）
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）
馮　斌先生（患者，前電腦工程師）
邵得志醫生（患者，執業醫生）
徐江琼女士（患者家屬，理財顧問）
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 Ken TO (Patient, Communication Manager)
Honorary Secretary: Ms Maria WONG (Patient, retired school principal)
Honorary Treasurer: Mr Jackie LEUNG (Patient, freelancer)
Council Members*: Professor Danny CHAN (Scientist)
Mr CHOW Kuen Tai (Patient's family, nurse practitioner)
Mr FUNG Pun (Patient, former computer engineer)
Doctor Byron SHIU (Patient, medical practitioner)
Ms Helen TSUI (Patient's family, Financial Advisor)
Ms WONG Man Yee (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（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 (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
許鍾妮醫生
陸志聰醫生，太平紳士
盛斌醫生

Appendix IV – Advisors

(In alphabetical order by surname)
Professor CHAIR Sek Ying
Professor Cecilia CHAN, JP
Doctor Joannie HUI
Doctor Che-chung LUK, JP
Doctor Bun SHENG

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 E&D
社會福利署 - 殘疾人士 / 病人自助組織資助計劃
罕盟顧問
理事會成員
罕盟秘書處成員及義工
香港視網膜病變協會

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 staff and volunteers
Retina Hong Kong

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RARE DISEASE HONG KONG

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社聯 HKCSS
機構會員
AGENCY MEMBER