香港罕見疾病聯盟
RARE DISEASE HONG KONG

會務報告
ANNUAL REPORT
2020

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 utilization & outcome-based approach

Management principles:
Openness, transparency, integrity & accountability

香港罕見疾病聯盟有限公司 2020 年會務報告

會長：曾建平 (2021 年2 月)

Rare Disease Hong Kong Limited Annual Report 2020

Chairman : TSANG Kin Ping (February 2021)

2020 年是極不平凡的一年，新冠肺炎疫情蔓延全球，給世界各地的方方面面造成前所未有的衝擊。在此特殊和艱難的時刻，香港罕見疾病聯盟（下稱「罕盟」）不忘使命，緊守崗位，順勢而為，沉著應對，各項核心業務依然取得理想的進展。

2020 has been an extraordinary year. The global spread of coronavirus has an unprecedented impact on every aspect of the world. During this special and difficult time, Rare Disease Hong Kong (RDHK), bearing in mind the mission, has remained steadfast and committed to our duties. By overcoming challenges with adaptability and calmness, we were still able to make good progress in different aspects of work.

1 政策倡議

去年罕盟的政策倡議工作以落實「香港罕見疾病行動計劃」為主軸，持續推動官、商、民三方協作，敦促政府履行施政報告的承諾，令患者得到及時適切的治療，病人權利得到尊重和保障。

1 Policy advocacy

Last year, RDHK's policy advocacy work was focused on the implementation of the Hong Kong Rare Disease Action Plan. We have been promoting the tripartite collaboration among the government, the business sector and the community, as well as urging the government to fulfil its promises in the policy address, so that patients can receive timely and appropriate treatment, and patient rights are respected and protected.

1.1. 政府在 2019 年施政報告提出若干針對罕病的新措施，但落實進度極不理想。去年罕盟多次主動與食物及衞生局和醫院管理局（醫管局）聯絡，要求通報有關措施的落實計劃和進度。在 10 月向施政報告提交的意見書，罕盟要求訂立落實新措施的時間表和成效指標，就落實進度定期與病人組織溝通討論。

1.1. The government put forward a number of new measures to combat rare diseases in the 2019 Policy Address, but the implementation progress is far from satisfactory. In 2020, RDHK approached the Food and Health Bureau and Hospital Authority (HA) on many occasions, requesting notification of the implementation plan and progress of the relevant measures. In the submission on 2020 Policy Address submitted in October, RDHK requested the setting of a timeline and outcome indicators for the implementation of the new measures, and regular communication and discussion with patient groups about the implementation progress.

1.2. 意見書還要求為罕見疾病藥物納入安全網的審批制度訂下清晰明確指引，讓臨床專家、病人和藥劑業界有所適從；並再次敦促成立由政府牽頭、各方持份者組成的罕病策略督導委員會，以及訂定本地罕病定義和公佈罕病名單。

1.2. In the submission, RDHK also requested the setting of clear guidelines on the review mechanism of inclusion of orphan drugs into the Safety Net for easy reference of the clinical physicians, patients and the pharmaceutical industry. And once again, we urged the government to establish a steering committee on strategy for rare diseases led by the government and comprising various stakeholders, and to formally define 'rare disease' and publish the rare disease list.

1.3. 為了解患者對 2019 年優化後藥物安全網經濟審查機制執行的意見，罕盟於去年上半年對十多名獲得資助的患者進行問卷調查，就審批時間、工作人員處事方式、取藥及共付藥費安排、資訊透明度等整理成報告提交給政府及關愛基金，作為進一步優化機制的建議。

1.3. The means test mechanism for drug subsidies under the Safety Net was improved in 2019. In order to understand patients' opinions on the implementation of the improved mechanism, RDHK launched a questionnaire survey in the first half of 2020. More than ten subsidised patients were interviewed. A report on the processing time, modus operandi of the staff, arrangements of drug collection and co-payment, information transparency, etc. will be compiled and submitted to the government and the Community Care Fund as suggestions for further optimizing the mechanism.

1.4. 作為對病人組織訴求的回應，政府在 11 月發表的施政報告，提出進一步完善昂貴藥物安全網的經濟審查機制，並繼續按既定機制增加兩個基金下涵蓋的藥物及放寬現有藥物的臨床準則。

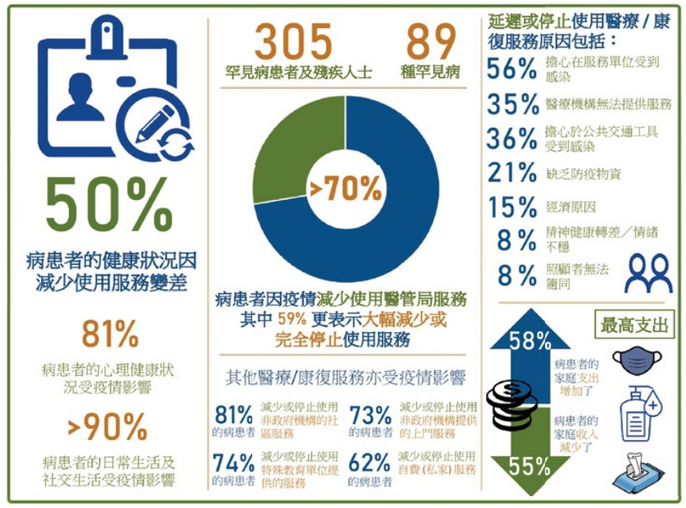
1.4. In response to the patient groups' demand, the government proposed in the Policy Address released in November 2020 that they will further refine the means test mechanism under the Safety Net and will continue to increase the number of drugs covered under the two Funds and relax the clinical criteria of existing drugs in accordance with the established mechanism.

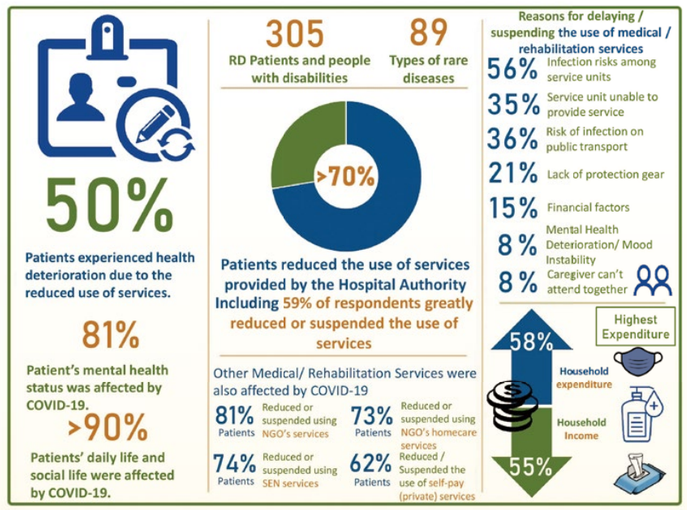
1.5. 脊髓肌肉萎縮症（SMA）用藥於 2018 年在行政長官親自介入後，十多名年幼患者開始接受治療，效果理想。然而為數更多的二型及三型患者卻被醫管局一再借詞拖延，用藥無期。罕盟先後於 2020 年年初和年中舉行新聞發佈會，介紹外國二型及三型患者用藥的成效，並向醫管局提出借鏡英國的經驗，由醫管局、病人組織和藥廠協商，推行藥物試用期計劃，收集實證數據，讓試用後達至臨床準則的患者繼續接受治療。

1.5. Due to the Chief Executive's intervention in 2018, more than a dozen young patients with Spinal Muscular Atrophy (SMA) began to receive medication treatment which works well for them. However, there is a larger group of Type 2 and Type 3 SMA patients who have been denied access to medication by the HA for various excuses. RDHK held press conferences in early and mid-2020 to introduce the treatment effect to Type 2 and Type 3 SMA patients overseas after receiving medication. We have also advised the HA to learn from the experience of the United Kingdom, and to implement a drug trial programme to collect empirical data and allow patients who meet clinical criteria after trial to continue to receive treatment.

1.6. 就新冠疫情對罕病患者的影響，罕盟於 4 月向三百多名患者和照顧者進行問卷調查，並與香港大學協作整理成報告，要求政府正視及回應；香港大學還將資料編寫成論文在國際醫學期刊發表。

1.6. In view of the impact of the coronavirus epidemic on patients with rare diseases, RDHK conducted a questionnaire survey in April with more than 300 patients and caregivers participating. The survey report was compiled in collaboration with the University of Hong Kong and submitted to the government who was requested to face up and respond to our report. Furthermore, the University of Hong Kong compiled the information into a paper that was published in international medical journals.





1.7. 有見部份未符合藥物安全網資助的患者往往因現金週轉難以及時接受藥物治療，罕盟與社區藥房、信用卡公司、藥廠等合作，去年 9 月推出藥費分期付款計劃，首階段涵蓋三種罕病和癌症藥物。罕盟繼續接觸其他持份者如私家醫院等，爭取進一步擴大計劃，令更多患者受惠。

1.7. Knowing that some patients who are not eligible for drug subsidies under the Safety Net often have difficulty in receiving timely medication treatment due to cash flow problem, RDHK collaborated with community pharmacies, credit card companies, pharmaceutical companies, etc., to launch a Medication Payment Instalment Plan in September 2020. Three rare disease and cancer drugs were selected as the starting point for the Plan. RDHK keeps reaching out to other stakeholders like private hospitals, etc. to further expand the Plan to benefit more patients.

1.8. 此外，罕盟還就醫管局藥物名冊雙年檢討，以及衞生署先進療法製品規管提交書面意見；並要求醫管局為及時準確診斷部份罕病改善檢測設施。

1.8. In addition, RDHK sent in submissions on the biennial comprehensive review of the HA drug formulary and Regulation of Advanced Therapy Products under the Department of Health, requesting the HA to improve testing facilities for timely and accurate diagnosis of some rare diseases.

2 提昇認知

因應新冠疫情對實體活動帶來的影響，罕盟去年充份利用網上平台開展傳訊工作。

2.1. 去年罕盟與香港中文大學合作舉辦「世界罕病日」研討會，因疫情一再延期，十月中旬透過網上進行，參加者超過三百名，是歷年最多。研討會配合罕盟的倡議工作，邀請政府官員、醫管局、學者、藥廠代表等擔任講者，透過官、商、民各抒己見，探討香港有效應對罕病挑戰的各項策略、計劃和措施，集結各方智慧，調動持份者的力量，搭建新的網絡，共同為香港罕病患者的福祉謀求對策。

2 Public awareness

In response to the impact of the coronavirus epidemic on physical activities, RDHK moved most of the public awareness events online last year.

2.1. The World Rare Disease Day 2020 Symposium jointly run by RDHK and The Chinese University of Hong Kong, after being postponed twice due to the epidemic, was held online in mid-October. With more than 300 participants, it was the highest attended event over the years. In line with the advocacy work of RDHK, officials or representatives from the government, the HA, academia and the pharmaceutical industry, etc. were invited as speakers of the Symposium. Through opinion exchange among the government, business sector and the community, various strategies, plans and measures for effectively coping with the challenges of rare diseases in Hong Kong were explored. It is hoped that by gathering the wisdom of all parties, mobilizing the power of stakeholders and building a new network, innovative ideas and strategies could be jointly conceived for the well-being of patients with rare diseases in Hong Kong.

2.2. 去年第四季罕盟先後舉辦特發肺纖維化（IPF）和多發性硬化症（MS）兩場網上專題講座，邀請專科醫生介紹疾病管理知識，病友分享應對疾病的心得，觀眾即時與講者進行互動。罕盟還推出網上疾病教育計劃，由一連六則有關 IPF 及間質性肺病（PF-ILD）的專題帖文及短片組成，並邀請社交媒體意見領袖（social media KOL）分享，共接觸 315,400 人次和獲得 3,132 個點讚及 145 個留言。

2.2. In the fourth quarter of 2020, two online talks on Idiopathic Pulmonary Fibrosis (IPF) and Multiple Sclerosis (MS) were held successively. In those talks, specialists were invited to introduce knowledge about disease management, and patients were invited to share their experience in dealing with the disease. Meanwhile, the audience could interact with the speakers in real time. In addition, RDHK launched an online disease awareness programme consisting of a series of six thematic posts and short videos on IPF and Progressive-Fibrosing Interstitial Lung Disease (PF-ILD). Having been shared by some social media KOLs, the posts and videos reached 315,400 people, gained 3,132 likes and 145 comments.

2.3. 與社區和持份者直接交流是提昇認知的有效途徑。罕盟的「真人圖書館」計劃去年受疫情影響，部份被迫取消，部份改以網上進行。2020年先後為香港中文大學醫學院、明愛醫院護士學院校和幾間中、小學舉行 8 場講座交流，受眾人數約為1,562 人。

2.3. Face to face communication with the community and stakeholders is an effective way to raise awareness. Being affected by the epidemic, some of the sharing sessions under our Human Library Programme were either cancelled or moved online. In 2020, a total of 8 sharing sessions were conducted for the Faculty of Medicine, The Chinese University of Hong Kong, School of Nursing, Caritas Medical Centre, and a few primary and secondary schools, addressing an audience of about 1,562 people.

2.4. 去年罕盟網頁錄得 71,247 訪問人次；機構臉書帖文觸及438,486人，穩步上昇。

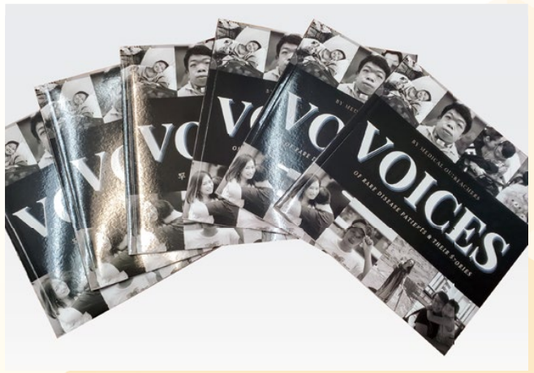
2.4. Last year, there were 71,247 visits to the website of RDHK, whereas our Facebook posts reached 438,486 people, showing a steadily increasing post reach.

2.5. 《罕情》是綜覽本地罕病群體的刊物。為適應不同讀者的需要，它以有限的篇幅，既涉獵國際和本地的重大罕病政策，亦報導香港罕病團體的各類活動，圖文並茂，提供電子版和印刷版，成為政府部門、服務機構、病人團體以及社會大眾了解和認識罕病相關資訊的重要媒介。去年出刊四期，共印發五千份。

2.5. *RareCare* is a publication that provides an overview of the local rare disease community. In order to meet the needs of different readers, we try to cover not only international and local major rare disease policies, but also various activities of Hong Kong rare disease groups with constrained length, illustrated with pictures. Both electronic and printed versions are available. *RareCare* has become an important channel for government departments, service organisations, patient groups and the general public to learn about and understand information relating to rare diseases. Four issues were published last year, and a total of 5,000 copies were printed and distributed.

2.6. 此外，罕盟去年支持和贊助醫科學生義工組織「醫心」編撰印發罕病故事書，以十多名罕病患者的真實經歷為內容，圖文並茂，共印發一千五百冊。

2.6. In addition, last year, RDHK supported the medical student volunteer organisation 'Medical Outreachers' by sponsoring them for the compilation and publication of a true storybook based on real life of over a dozen rare disease patients, illustrated with pictures. A total of 1,500 copies were published.



3 能力建設

能力建設是去年最受疫情影響的核心業務。

3.1. 罕盟於去年 6 月舉辦了一場罕病領袖交流會，十多個罕病組織的負責人分享組建和開展活動的經驗；於年初支援結節性硬化症（TSC）和家族性澱粉樣物多發性神經病變（FAP）病友舉辦活動，加強溝通交流；以及協助面肩肱型肌肉營養不良症（FSHD）病友建立聯絡網絡。

3 Capacity building

The work of capacity building was most affected by the epidemic last year.

3.1. A sharing session for rare disease group leaders was held in June 2020, in which over a dozen leaders of rare disease organisations shared their experience in organising and carrying out activities. In early 2020, RDHK assisted patients with Tuberous Sclerosis Complex (TSC) and Familial Amyloid Polyneuropathy (FAP) in organising activities to enhance communication, and helped Facioscapulohumeral Muscular Dystrophy (FSHD) patients to establish their network of contacts.

3.2. 罕盟去年動員和資助數十名病友參加內地罕病組織於 8 月和 9 月舉行的網上研討會，了解內地和國際在診斷和治療罕病的最新資訊。

3.2. Dozens of patients were motivated and subsidised by RDHK to participate in the online seminars hosted by the rare disease organisations in the Mainland in August and September 2020, in which the latest information on diagnosis and treatment of rare diseases in the Mainland and overseas was shared.

4 其他工作

4.1. 罕盟於去年完成更改為有限公司註冊手續，於 5 月 30 日舉行更改註冊後的第一屆會員大會及選出首屆理事會（名單請見附錄二）；6 月 1 日起正式更名為「香港罕見疾病聯盟有限公司」（Rare Disease Hong Kong Limited）。

4 Miscellaneous work

4.1. RDHK completed the registration procedures for changing to a limited company last year. The first General Meeting after being registered as a limited company was held on 30 May 2020, in which the first Council (please refer to Appendix II for the list of Council members) was elected. RDHK was officially renamed as 'Rare Disease Hong Kong Limited' effective from 1 June 2020.

香港罕見疾病聯盟
RARE DISEASE HONG KONG

4.2. 為支援會員抗疫，罕盟於去年多次向有需要的會員和罕病組織發送防疫物資，包括口罩八萬多個和清潔用品等；並為部份會員提供到戶物理治療服務。

4.2. To support members in fighting the epidemic, RDHK sent anti-epidemic supplies including over 80,000 face masks, cleaning supplies and so on, to members and rare disease organisations in need several times. Besides, home physiotherapy services were provided to some of the members.

4.3. 會長曾建平先生去年獲選連任國際罕病聯盟（Rare Disease International）理事，並擔任亞太罕病聯盟（Asia Pacific Alliance of Rare Disease Organisations）司庫。

4.3. Chairman Mr Tsang Kin Ping was re-elected to the Council of Rare Disease International and appointed Treasurer of Asia Pacific Alliance of Rare Disease Organisations in 2020.

附錄一、會員及所涵蓋病種

截至 2020 年年底為止，會員總人數為 527，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：474

   a) 個人會員數目：466

   b) 團體會員數目：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

Appendix I – Members and types of diseases

As at the end of 2020, the total number of members was 527, among which:

1. Number of ordinary members (those who care about and support rare disease patients): 53

2. Number of full members (rare diseases patients or family): 474

   a) Number of individual members: 466

   b) Number of organisational members: 8

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

1	10q26 Microdeletion Syndrome
2	14q
3	16q11.2-q22.2
4	18q21.2 x 3
5	1p36 Deletion Sydrome
6	1q44 deletion
7	22q13.33 Microdeletion Syndrome
8	3q23-3q25 Deletion
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	Amyotrophic lateral sclerosis（肌萎縮性脊髓側索硬化症）
16	Angelman Syndrome（天使綜合症）
17	Anhidrotic Ectodermal Dysplasia（家族性外胚層發育不良症）
18	Anti-NMDA Encephalitis（自體免疫性腦炎）
19	Apert Syndrome（亞伯氏症）
20	Aplastic Anemia（再生不良性貧血）
21	Aromatic L-amino Acid Decarboxylase Deficiency（芳香族 L- 胺基酸類脫羧基酶缺乏症）
22	Arthrogryposis Multiplex Congenita（先天性多發性關節攣縮症）
23	ATRX Thalassemia Syndrome（α 型海洋性貧血 / 智能發展遲緩）
24	Atypical Hemolytic Uremic Syndrome（非典型性尿毒溶血症候群）
25	Autosomal recessive spastic ataxia of Charlevoix-Saguenay（ARSACS）
26	Behcet's Disease（貝賽特氏症）
27	Bilateral Perisylvian Polymicrogyria（雙側西耳維厄斯周圍多小腦回畸形）
28	Burning Mouth Syndrome（口腔灼熱症候群）
29	Cardiofaciocutaneous Syndrome（CFC 綜合症）
30	Charcot Marie Tooth Disease（進行性神經性腓骨萎縮症）
31	Chronic Hemolytic Anemia（慢性溶血性貧血）
32	Cockayne Syndrome（柯凱因氏症候群）
33	Collagen XII Myopathy（骨膠原蛋白第 12 基因引致肌肉病變）
34	Cone-rod Dystrophy （視幹細胞營養不良）
35	Congenital Glaucoma（先天性青光眼）
36	Congenital Insensitivity to Pain with Anhidrosis, CIPS（先天性痛不敏感症合併無汗症）
37	Costello Syndrome（克斯提洛氏彈性蛋白質缺陷症）
38	Cri Du Chat Syndrome（貓哭症）/ Chromosome 5p Deletion Syndrome
39	Crohn's Disease（克隆氏症）

40 CTNNB1 Syndrome (CTNNB1 綜合症)
41 Cushing's Syndrome (庫欣氏症候群)
42 Dandy Walker Syndrome (Dandy-Walker 綜合症)
43 DDx3x (DDx3x 基因突變)
44 De lange syndrome (狄蘭吉氏症候群)
45 DiGeorge Syndrome (迪喬治症候群)
46 DNM1-L
47 Dystonia (肌張力障礙)
48 Ehlers-Danlos Syndrome (埃勒斯 - 當洛二氏症候群)
49 Eosinophilic Granulomatosis with Polyangiitis (過敏性肉芽腫血管炎)
50 Epidermolysis Bullosa (表皮溶解水皰症)
51 Fabry Disease (法布瑞氏症)
52 Familial Amyloid Polyneuropathy (家族性澱粉樣物多發性神經病變)
53 Fibrodysplasia Ossificans Progressiva (進行性肌肉骨化症)
54 Fragile X Syndrome (X 染色體脆弱症)
55 Glutamate Receptor, Ionotropic, N-Methyl D-Aspartate 1 (GRIN 1)
56 Glycogen Storage Disease (肝醣儲積症)
56.1 Glycogen Storage Disease - Type 1，G6PC variants (肝醣儲積症第 1 型，G6PC 病變異)
56.2 Glycogen Storage Disease - Type 2 (肝醣儲積症第 2 型) / Pompe Disease (龐貝氏症)
56.3 Glycogen Storage Disease - Type 6 (肝醣儲積症第 6 型)
57 Guillain-Barré Syndrome (格林 - 巴利綜合征)
58 Hemolytic-uremic Syndrome (溶血性尿毒綜合症)
59 Hereditary Angiodema (遺傳性血管性水腫)
60 Hereditary Spastic Paraplegia (遺傳性痙攣性下身麻痺)
61 Huntington's Disease (亨丁頓舞蹈症)
62 Ichthyosis (斑色魚鱗癬)
63 Idiopathic Intracranial Hypertension (特發性顱內壓增高)
64 Idiopathic Pulmonary Fibrosis (特發性肺纖維化)
65 IgG4-related Disease (IgG4 相關性疾病)
66 Insulin Antibody (胰島素抗體)
67 Invdupdel (8p)
68 Jacobsen Syndrome (11q 缺失症候群)
69 Kallmann Syndrome (卡爾曼氏綜合症)
70 KCNQ2
71 Kennedy Disease (甘迺迪氏症)
72 Klippel–Trénaunay Syndrome (靜脈畸形骨肥大綜合症)
73 Langerhans Cell Histiocytosis (蘭格罕細胞組織球增生症)
74 Larsen Syndrome (顎裂 - 先天性脫位症候群)
75 Leopard Syndrome (Leopard 症侯群)
76 Lipid Storage Myopathy (脂質沉積性肌病)
77 Maple Syrup Urine Disease (楓糖尿症)
78 McCune Albright Syndrome (纖維性骨失養症)
79 Medulloblastoma (髓母細胞瘤)

80 Methylmalonic Acidemia (甲基丙二酸血症)
81 Mitochondrial Cardiomyopath (線粒體心肌病變)
82 Motor Neuron Disease (運動神經元病)
82.1 Amyotrophic Lateral Sclerosis (肌萎縮性脊髓側索硬化症)
83 Mowat-Wilson Syndrome
84 84.1 Mucopolysaccharidoses Type 2 (黏多醣症第 2 型)
84.2 Mucopolysaccharidoses Type 3 (黏多醣症第 3 型)
84.3 Mucopolysaccharidoses Type 4A (黏多醣症第 4A 型)
84.4 Mucopolysaccharidoses Type 6 (黏多醣症第 6 型)
85 Multiple Epiphyseal Dysplasia (多發性骨骺發育不全)
86 Multiple Pituitary Hormome Deficiency Anterior Pituitary Hypoplasia (腦下垂體發育不全)
87 Multiple Sclerosis (多發性硬化症)
88 Multiple System Strophy-Pakinsonian Subtype (多重系統退化症 - 非典型巴金森症)
89 Muscular Dystrophy (肌肉營養不良症)
89.1 Congenital Muscular Dystrophy (先天性肌失養症)
89.2 Duchenne Muscular Dystrophy (杜興氏肌肉營養不良症)
89.3 Facioscapulohumeral Muscular Dystrophy (面肩肱型肌肉營養不良症)
90 Myasthenia Gravis (重症肌無力症)
91 Nemaline Myopathy (桿狀體肌肉病變)
92 Neurofibromatosis (神經纖維瘤)
92.1 Neurofibromatosis type I (神經纖維瘤第 1 型)
93 Neuromyelitis Optica (視神經脊髓炎)
94 Nicolaides-Baraitser Syndrome
95 NINA Uveitis (葡萄膜炎)
96 Osteogenesis Imperfecta (成骨不全症)
97 Paroxysmal Nocturnal Hemoglobinuria (陣發性夜間血紅素尿症)
98 Pathogenic Variant GSD 9A
99 Peutz-Jeghers Syndrome (黑斑息肉症候群)
100 Phakomatosis Pigmentovacularis (色素血管性母班症)
101 Phenylketonuria (苯丙酮尿症)
102 Prader Willi Syndrome (小胖威利症)
103 Primary Ciliary Dyskinesia (先天性纖毛運動障礙)
104 Primary Immunodeficiency (原發性免疫缺陷病)
105 Primary Pulmonary Arterial Hypertension (肺動脈高壓)
106 Progressive Pseudorheumatoid Dysplasia (進行性假性類風濕性骨發育不良)
107 Propionic Acidemia (丙酸血症)
108 Pulmonary Alveolar Proteinosis (肺泡蛋白質沉著症)
109 Pyruvate Dehydrogenase Deficiency (丙酮酸鹽脫氫酵素缺乏症)
110 Relapsing Polychondritis (復發性多軟骨炎)
111 Remphigus Vulgaris (天皰瘡)
112 Retinitis Pigmentosa (視網膜色素病變)
113 Rett Syndrome (雷特氏症)
114 Rhabdomyosarcoma (橫紋肌肉瘤)

115 Rubinstein-Tabyi Syndrome (Rubinstein-Tabyi 氏症候群)
116 Russell-Silver Syndrome (羅素 - 西弗氏症)
117 Schaaf-Yang Syndrome (Schaaf-Yang 綜合症)
118 Scleroderma (硬皮症)
119 SCN2A
120 SCN8A
121 Skeletal Dysplasia (骨骼發育異常)
122 Smith-Magenis Syndrome (史密斯 - 馬吉利氏綜合症)
123 Spastic Paraplegia (遺傳性痙攣性下肢麻痺)
124 Spinal Cord AVM (C6-T4)
125 Spinal Muscular Atrophy (脊髓肌肉萎縮症)
126 Spinalcerebellar Ataxia (小腦萎縮症)
126.1 Spinalcerebellar Ataxia Type 2 (小腦萎縮症第 2 型)
126.2 Spinalcerebellar Ataxia Type 3 (小腦萎縮症第 3 型)
127 Spondyloepiphyseal Dysplasia Congenita (先天性脊椎骨骺發育不全)
128 Stiff Person Syndrome (僵硬人症候群)
129 Succinic Semialdehyde Dehydrogenase Deficiency (SSADH 缺乏症)
130 Supernumerary Chromosome 8
131 Takayasu Arteritis (高安氏症)
132 TRIOBP (波形蛋白)
133 Tuberous Sclerosis Complex (結節性硬化症)
134 Usher Syndrome (尤塞氏綜合症)
135 Visceral Myopathy & Visceral Neuropathy
136 Waardenburg Syndrome (瓦登伯革氏症候群)
137 Walker-Warburg Syndrome
138 WDR45
139 Wells Syndrome
140 West Syndrome-Infantile Spasms (韋氏症候群)
141 Williams Syndrome (威廉氏症候群)
142 Wilson's Disease (威爾森氏症)
143 Wolfram Syndrome (Wolfram 氏症候群)
144 Xeroderma Pigmentosum (著色性乾皮症)
145 X-linked Agammagluolinmia (X- 連鎖無丙種球蛋白血癥)
146 X-linked Hypophosphatemic Rickets (性聯遺傳型低磷酸佝僂症)
147 X-linked Retinoschisis
148 X-linked Thrombocytopenia
149 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

(按英文姓氏排序)
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)

附錄三、秘書處成員

賴家衛先生 (倡議主任)
陳蔚斯女士 (項目協調主任)
何翠薇女士 (傳訊主任)
陳淑雲女士 (護理顧問)
黃桂婉女士 (活動助理)

附錄四、顧問

(按英文姓氏排序)
陳浩然教授
陳麗雲教授 , JP
車錫英教授
許鍾妮醫生
盛斌醫生

鳴謝：

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秘書處成員及義工
余兆麒殘疾人士基金

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:

(In alphabetic order by surname)
Professor Danny CHAN (Scientist)
Mr Ronald CHOW (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 Amy WONG (Registered social worker)

Appendix III – Staff of the Secretariat

Mr Terry LAI (Advocacy Officer)
Ms Iris CHAN (Project Coordination Officer)
Ms May HO (Communication Officer)
Ms Amy CHAN (Nursing Advisor)
Ms Pat WONG (Programme Assistant)

Appendix IV – Advisors

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

Acknowledgement

(In alphabetical order)
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The S.K. Yee Fund for the Disabled

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