第一屆罕盟週年會員大會 。1st Annual General Meeting

罕情 創刊號 #RareCare First Issue

香港罕見疾病聯盟會訊 Newsletter of HKARD January 2016 (Issue No. 1)

第一屆罕盟週年會員大會圓滿舉行

HKARD 1st Annual General Meeting Held Successfully

罕盟於 2016 年 1 月 23 日於灣仔温莎公爵社會服務大廈舉行了第一屆週年會員大會，順利選出第一屆的理事。

HKARD's 1st AGM was held on January 23, 2016 at Duke of Windsor Social Service Building, Wan Chai, 1st Council Members were elected.

十二位理事分別為曹綺雯女士、梁七根先生、曾建平先生、方緯谷先生、陳振勝教授、黃婉冰女士、賴家衞先生、熊德鳳女士、周權棣先生、鍾侃言醫生、何立明先生和黃敏兒女士。新一屆理事成員分別由四位患者、四位照顧者及四位專業人士組成。理事代表曾建平先生向各創會臨時理事過去一年多的努力工作致謝，承諾第一屆理事會會秉承罕盟的宗旨，努力不懈聯繫香港不同的罕病病友組織及持份者，爭取公平和合理的社會資源分配。大會尾聲由香港科研製藥聯會執行董事陳素娟女士主講「香港的藥物註冊制度及對病人的影響」。雖然當天氣溫只有攝氏10度，會場卻洋溢着溫暖團結的氣氛。會員在大會中積極提問及提供意見，大會在一片歡愉共融的氣氛中結束。

The new Council consists of members from four rare disease patients, four caregivers and four professionals. Mr K P Tsang, on behalf of the 1st Council Members, expressed the gratitude to members of the Provisional Executive Board. He stated that 1st Council Members would work as per HKARD's mission & objective, commit to put their full efforts to collaborate with various patients groups and stakeholders in Hong Kong, for a fair and rational allocation of social resources in managing rare diseases. The AGM ended by Ms Sabrina Chan, Executive Director of the Hong Kong Association of the Pharmaceutical Industry, who delivered an education session to all the participants with topic "Drug Registration System in Hong Kong and the Impact on Patients". Although the outdoor temperature was only 10 degrees Celsius, the meeting hall was filled with an atmosphere of warm solidarity. Members asked questions actively and expressed invaluable opinions to Council Members. HKARD 1st AGM ended in a happy atmosphere of communion.

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罕盟#新友 HKARD# New Members

罕盟現有會員來自以下 18 種的罕病類別 ：

Current members of HKARD are patients or carers of the below 18 kinds of rare diseases :

1. 肢端肥大症 (巨人症, Acromegaly)
2. 重症肌無力症；(MG, Myasthenia Gravis)
3. 多發性硬化症；(MS, Multiple Sclerosis)
4. 肌營養不良症 (MD, Myotonic Dystrophy)
5. 神經纖維瘤 (Neurofibromatosis)
6. 雷特氏症 (Rett Syndrome)
7. 小腦萎縮症 (SCA, Spinocerebellar Ataxia)
8. 溶血性尿毒綜合症 (HUS, Hemolytic-uremic Syndrome)
9. 天使綜合症 (Angelman Syndrome)
10. 龐貝氏症 (Pompe Disease)
11. 軟骨發育不全症 (Achondroplasia)
12. 魯賓斯坦-泰必氏綜合症 (RTS, Rubinstein Tabyi Syndrome)
13. 結節性硬化症 (TSC, Tuberous Sclerosis Complex)
14. 視網膜色素病變 (RP, Retinitis Pigmentosa)
15. 罕見骨骼疾病 (Skeletal Dysplasia)
16. 脊椎性肌肉萎縮症 (SMA, Spinal Muscular Atrophy)
17. 卡爾曼氏綜合症 (KS, Kallmann Syndrome)
18. GRIN 1 (Glutamate Receptor, Ionotropic, N-Methyl D-Aspartate 1)

期待大家共同協力，教育及提昇公眾對罕病的關注，從而逐步爭取社會在資源和政策上加強對罕見疾病的支援。

We look forward to work together for the objective to educate and raise the public awareness of rare diseases. Thus, gradually strengthen the community's support for rare diseases in terms of resources and policy.

2015 年 11 月 30 日的病友交流活動。
Patient group meeting on 30th November, 2015.

罕盟建議醫管局優化「藥費封頂計劃」造福病人

罕盟於 11 月 23 日致信醫院管理局聯網服務總監張偉麟醫生就已推出惠及罕見癌症皮膚癌黑色素瘤病人和罕見 ALK 肺癌的「藥費封頂計劃」提出意見。罕盟期望醫管局對現行計劃予以優化，盡快推動「安全網藥費封頂先導計劃」，透過關愛基金或撒瑪利亞基金的機制，資助通過經濟審查的病人支付「藥費封頂計劃」的藥費自付部份。罕盟於 12 月 28 日收到張醫生書面回覆，但未有就罕盟的訴求作具體承諾。

罕盟與醫管局兒童醫院籌劃工作組會面

罕盟臨時理事會理事於 11 月 30 日在醫管局總部與兒童醫院籌劃工作組會面，反映本會對將於 2018 年投入服務的兒童醫院的期盼及建議。罕盟代表罕見疾病患者及其家屬向醫管局強調新生兒檢查、早期確診、及早治療、病例名冊、臨床研究、培訓臨床基因遺傳專家、病人和家屬心理支援及社區資源的配合等....，對有效治療或管理罕見疾病尤其重要。罕盟促請兒童醫院在規劃針對罕病的服務時，全面考慮這些因素。罕盟得到籌劃工作組正面的回應，工作組承諾會繼續在兒童醫院的各發展階段徵詢罕盟的意見。

HKARD's Provisional Council Members met with the Task Force Members of HK Children Hospital in HA headquarter

On behalf rare diseases patients and their carers, HKARD members met with HKCH Planning and Commissiong Committee representatives to share the Alliance's opinions and expectations on the service deliverable of HK's future Children's Hospital. The Alliance stated that new born babies screening, early diagnostic support, effective & fast treatment and evidence base registry & research were crucial in rare diseases management. Dr Libby Lee replied that all these were well aligned with the current plan of HKCH's service implementation. The Committee committed to maintain communication with HKARD at every HKCH's development and opinion gathering phase.

HKARD wrote to the Hospital Authority suggesting ways to optimize the newly launched "Medication Capping Program"

HKARD wrote to Dr Cheung Wai-lun, Director (Cluster Services), Hong Kong Hospital Authority, on 23rd November, 2015 to express our concern on and suggested ways to optimize the newly launched "Medication Capping Program" for rare cancers, melanoma and ALK+ lung tumor. The Alliance suggested that the government should consider subsidizing patients who fall into the safety net by paying the self-paid portion of the Capping Program under the Community Care Fund or the Samaritan Fund. This is an initiative of "Pilot Medication Capping Program for Safety Net Patients". The Alliance received a reply letter from Dr Cheung on 28th December,2015. No specific commitment was stated.

罕盟第一屆理事 **HKARD 1st Council Members**

理事會正式成立！We are on board !

罕盟第一屆理事（圖左至右）：方緯谷先生、賴家衛先生、周權棣先生、曹綺雯女士、梁七根先生、曾建平先生、黃婉冰女士、熊德鳳女士、陳振勝教授。鍾侃言醫生（缺席）、何立明先生（缺席）和黃敏兒女士（缺席）。

項目主任（對外事務）：林文珊女士。 導盲犬 ：Deanna

1st Council Members of HKARD (from left to right) : Mr Dennis Fong, Mr Terry Lai, Mr Ronald Chow, Ms Tso Yee Man, Mr Leung Chat Kan, Mr K P Tsang, Ms Maria Wong, Ms Anchor Hung, Prof Danny Chan. (Apologise : Dr Brian Chung, Mr Ho Lap Ming and Ms Amy Wong)

Project Officer (External Affairs) : Patti Lam

KP's guide dog : Deanna

罕盟#點點情

「罕病於我，並不罕見。」

…毋須質疑的接受與承擔。
- 亨廷頓舞蹈症（Huntington Disease, HD)

「媽媽於 12、13 年前， 約 50 多歲時開始發病。這個病影響她身體的平衡與協調性， 肌肉會不自主顫動、常跌。看着她日漸消瘦，繼而進食和消化有困難；語言表達、活動及認知能力逐步退化 ……。現在很珍惜媽媽的每個發出來的字語。她已變成我的媽媽 BB 了！」

「其實，HD 於我， 並不罕見 ！」

現在任職企業傳訊工作，30 多歲的 Ken， 杜勤創，大學修讀語言及翻譯。回想自己在小學階段，已接觸到 HD 症狀了。

「媽媽現在的症狀，就跟外婆當時的情況一樣。街坊鄰里也這樣說。」

輕描淡寫的一句話，正正帶出了 HD 或其它罕病與遺傳基因不可分割的關係。Ken 說 ：「他正看着 50%的發病機率在家族中出現。」理性的數據從無奈的語調中說出來。

Ken 的媽媽是印尼華僑，有十兄弟姐妹，排行第二的母親是當中首位 HD 患者，隨後已有三位阿姨和舅夫有 HD 的症狀甚至已過世了。Ken 是老大，未婚。另有兩位已婚妹妹。Ken 說：「自己現階段不打算接受基因測試，因覺得測試結果如何，也都會是壞消息。」 他說 ：「活在當下！」

正因如此，眼前輕鬆自若的 Ken，每天工作，也同時積極、全心地照顧着媽媽， 亦樂意多分享、多貢獻，漸漸找出了實質的方法令自己情緒舒緩。「沒有人希望以承受苦難來顯示自己的能力，但既然這些罕有的歷練必須要面對，總有我可以貢獻的契機和要傳遞的信息吧！」Ken 笑說。

故此，我們會看見 Ken 與中國 HD 病友及學者的活躍聯繫、在香港黏多醣症暨罕有遺傳病互助小組中無私的支持會友和樂意地與病友分享自己的經歷，互相鼓勵。 2015 年聖誕更與在印尼的 4、5 位舅父、阿姨和姨丈一起籌劃了一個家族大旅行。一行 60 多人從耶加達出發去不同城市暢遊。行程以一個感動的家族 HD 分享會作總結，把各家族成員緊緊地聯繫在一起，互相扶持！Ken 深深體會到：「就算活在疾病或其陰影之中，人生仍然能夠充滿意義。親人的互相關顧與支持對罕病者家庭尤其重要。」

Ken 現在的工作也繁忙，也會遇到挫折與困難，但他覺得比起他與媽媽跟 HD 無終點、漫長的鬥爭，工作的辛苦反而是他精神壓力的釋放區。在照顧媽媽的經歷中，最磨人的就是社會人士甚至醫護介對罕病認知的不足，媽媽日間的護理服務配套缺乏，政府提供給照顧 HD 患者生活所需要的資源與支緩，幾近於零。Ken 說：「期待罕病病友及組織能把經驗整合，提醒及建議政府擴大並深化罕病的治療和護理工作！」

鳴謝 ： 香港黏多醣症暨罕有遺傳病互助小組
Acknowledgement : Hong Kong Muscopolysaccharidoses and Rare Genetic Diseases Mutual Aid Group (HKMPS)

“Rare disease to me , is not uncommon.” ……. unconditional acceptance and commitment

- Huntington Disease, HD

“The onset of HD of my mom was 12 or 13 years ago when she was around the age of 50. This disease affects her body balance and coordination, involuntary trembling muscles and fall down very often. She keeps losing her body weight, follows by eating and digestive difficulties ; her language ability, mobilization and cognitive ability deteriorated progressively…….. At this moment, I cherish every single words which my mom speaks out. She is my babe now !”

“In fact, rare disease to me, is not uncommon !”

Ken To, around 35 years old, serving the corporate communication field, obtained his first degree in Lingustic and Translation. He recalled that his first experience on HD signs should be at the time he was in primary school.

“The current condition of my mom just like the situation of Grandma in the old days. Our neighbourhoods even say so.” Ken said, “I am experiencing HD’s 50% inheritability amongst members of my whole family.” Ken’s mom is an Indonesian overseas Chinese, with 10 silbings. His mom is the eldest sister. There were three members of her generation showed the signs & symtoms of HD and an uncle even passed away. Ken is the eldest brother in his family. He is single with two married sisters. Ken said, “I don’t think receiving genetic test for myself can bring any good news to me…..Let’s live in the moment!”

“I think no one wants to prove his/her ability by experiencing suffers intentionally, however, if it is the fact that I need to face such a rare experience, I think it is a hint to me that I have the ability to contribute and commit for something.” Ken smiled. Therefore, Ken is being active in collaborating with China’s HD stakeholders, providing volunterring support to HKMPS and sharing his experience constantly amongst RD patients. He recently joined a family trip in Indonesia with over 60 family members organised by several uncles and aunts in last Christmas. The gathering concluded with a dinner nite with a touching HD sharing session among all their family members. Members of the family, who travelled from different places, were closely bonded and united. Ken stated, “The mutual care and support from family members is of ulmost importance in RD family. “ “With the active collaboration of different RD patients or mutual aid groups, I hope all the different experiences from various parties can be consolidated effectively and can help to establish a quality Rare Disease Policy in the society with breadth and depth.” Ken raised out the dreams of every RD conern individuals.

罕盟提交 2016 年特首施政報告和 2016/17 財政預算案的意見書

對於未來一年政府的施政和預算，罕盟於 12 月 14 日向政府提出以下意見和訴求，望有關部門予以考慮及回應 。(一)與各方持份者協商，從速制定香港的罕見疾病定義和政策。(二)加強診斷支援。(三)有效運用資源擴大罕病治療。 (四)有效協調各項護理服務。 (五) 建立患者登記冊及加強臨床研究。(六)在香港兒童醫院設立罕見疾病中心。(七)支援病人組織。(八) 提供一站式的社會支援。 (九) 全面推行初生嬰兒篩查。

HKARD submitted letter of opinion to HKSAR government for the preparation of 2016 Chief Executive Policy Address and 2016/17 Budget Proposal on 14th December 2015

In view of the current health policy on HK's rare diseases management, HKARD's recommendations are summarized as follows: (1) To urge the Government to collaborate with the stakeholders of rare diseases to establish a proper definition on rare diseases in Hong Kong and set out of relevant policy; (2) To enhance the support of early diagnosis of rare diseases; (3) To make better use of resources to provide treatment for more patients with rare diseases under transparent guidelines; (4) To have better coordination of different aspects of medical and social services providers to facilitate all-rounded comprehensive rare diseases care to patients and their care givers; (5) To establish HK rare diseases patients registry and to reinforce the conduction of clinical research which is engaged with patients and patient groups; (6) To set up Center for Rare Diseases at Hong Kong Children's Hospital, which is going to commence its service in 2018, to play an active and leading role in coordination and management of rare diseases in Hong Kong; (7)To provide proactive support to patient groups of rare diseases; (8) To establish policy in Social Welfare Department for a one-stop case management infrastructure; (9) To implement screening to all newborn babies.

罕情#寰宇 HKARD#RD OneWorld

台灣 - 助罕見神經退化病友 全台首座照護中心成立

台灣衛福部於 2015 年 12 月 15 日召開記者會，宣布為提供罕見疾病個別化與整合性醫療服務，衛福部台中醫院已經在今年 11 月正式成立全台第一個「罕見神經退化疾病照護中心」，除漸凍人外，再擴大照護脊髓小腦萎縮症、多發性硬化症等神經退化性疾病患者，希望讓該中心成為台灣罕見疾病家庭照護的資源與後盾 .....

罕見遺傳疾病一點通(台灣)

台灣 - 衛生福利部公告新增三項罕見疾病，共計 210 種公告罕病

台灣衛生福利部於 2016 年 1 月 28 日公告，新增三項罕見疾病。截至目前政府公告之罕見疾病共計 210 種，只要是公告罕病即可享有全民健保重大傷病、罕見疾病防治及藥物法及身心障礙者權益保障法等政府相關福利的保障。新增三項病類資料如下：

（1）先天性肌失養症 (Congenital Muscular Dystrophy)
（2）嬰兒性溶酶體酸性脂肪酶缺乏症 (Infantile form Lysosomal Acid Lipase Deficiency / Wolman Disease)
（3）永久性新生兒糖尿病 (Permanent Neonatal Diabetes Mellitus)

財團法人罕見疾病基金會(台灣)

中國 - CFDA 最新公告，罕见病用药首次列入加快临床药品审批名单

CFDA 於 2015 年 11 月 13 日公告，加快临床急需等药品的审批，符合下列条件之一的，实行单独排队，加快审评审批。（一）防治艾滋病、恶性肿瘤、重大传染病和罕见病等疾病的创新药注册申请；.....

中國罕見病發展中心

中國 - 李克强主持召开国务院常务会议 提到加快罕见病药物产业化

国务院总理李克强 2 月 14 日主持召开国务院常务会议。 ..... 会议确定，一是瞄准群众急需，加强原研药、首仿药、中药、新型制剂、高端医疗器械等研发创新，加快肿瘤、糖尿病、心脑血管疾病等多发病和罕见病重大药物产业化……

中國罕見病發展中心

中國 - 56 种疾病纳入《上海市主要罕见病名录（2016 年版）》上海市卫生和计划生育委员会网站 2016-02-17 发布

海市卫生和计划生育委员会经专家论证，确定将 56 种疾病纳入《上海市主要罕见病名录（2016 年版）》，并印发了《上海市主要罕见病名录（2016 年版）》的通知，以供上海市开展罕见病宣传、筛查、诊断、治疗、康复和进一步制定罕见病防治的相关政策参考。……

加拿大 - Canadian Organization for Rare Disorders (CORD) established Canada's Rare Disease Strategy, May 2015

About 1 in 12 Canadians, two-thirds of them children, are affected by a rare disorder. But because each disease affects only a small number of individuals, understanding and expertise may be limited and fragmented across the country. Canada's Rare Disease Strategy proposes a five-point action plan that will address unnecessary delays in testing, wrong diagnoses and missed opportunities to treat.

1. Improving early detection and prevention,
2. Providing timely, equitable and evidence-informed care,
3. Enhancing community support,
4. Providing sustainable access to promising therapies and
5. Promoting innovative research……

Canadian Organization for Rare Disorders