影音使團於 6 月 19 日特設「天堂奇癒記」免費電影欣賞專場招待 217 位罕盟會友於父親節共聚了一個歡愉的下午。 A free "Miracles from Heaven" movie was offered by The Media Evangelism Ltd. on Father's Day dedicated for 217 HKARD's members & friends.

罕情 第二期 #RareCare Second Issue

香港罕見疾病聯盟會訊 Newsletter of HKARD July 2016 (Issue No. 2)

罕盟#匯報 HKARD #Activity Report

罕盟會友活動 RD Activities Joined

15/02 : 天使綜合症基金會新春團拜暨電影會 HKASF CNY & Movie Gathering

20/02 : 小而同罕有骨骼疾病基金會新書發佈會 LPHK Book Launching

09/06 : 天使綜合症基金會步行籌款 HKASF Angelman Walk (HK Station)

14/05 : 香港黏多醣症暨罕有遺傳病互助小組 11 週年聚餐 HKMPS 11th Anniversary Luncheon

父親節電影欣賞會剪影 Snapshots of Father's Day movie gathering.

罕盟病友傳媒訪問 Media Featuring

更多內容 More inside! pg. 3

出席父親節電影會的罕病組織/病友 RD groups/patients met & watched movie on Father's Day

- 香港雷特氏症協會 Hong Kong Rett Syndrome Association
- 香港神經纖維瘤協會 Hong Kong Neurofibromatosis Association
- 香港小腦萎縮症協會 Hong Kong Spinocerebellar Ataxia Association
- 香港黏多醣症暨罕有遺傳病互助小組 Hong Kong Mucopolysaccharidosis & Rare Genetic Disease Mutual Aid Group
- 香港結節性硬化症協會 Tuberous Sclerosis Complex Association of HK
- 小而同罕有骨骼疾病基金會 Little People of Hong Kong
- 威廉氏綜合症病童家長 Parents of William's Syndrome Children
- 天使綜合症基金會 Hong Kong Angelman Syndrome Foundation

罕盟#成員
HKARD#Members

罕盟現有會員來自以下 19 種的罕病類別 ：
Current members of HKARD are patients or carers of the below 19 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)
19. 克斯提洛氏彈性蛋白缺陷症 (Costello Syndrome)

克斯提洛氏彈性蛋白缺陷症

CS 最早於 1977 年被發現命名，患者的皮膚黝黑且皺褶較多、鼻子塌陷、毛髮稀少捲曲、厚唇、關節及肌肉鬆軟、心臟病變、生長遲緩等。研究認為應與 HRAS 基因的突變有關。當 HRAS 基因突變後產生的 HRAS 蛋白(負責調控細胞的生長和分裂)會過度活化，使細胞生長的速度失控，不受細胞外生長因子的控制，所以常導致腫瘤細胞的產生。目前全世界約有 250 個患者，為非常罕見的疾病。此症目前並無可根治的方式，僅能以症狀治療，如：患童的飲食狀況不佳，通常需要以鼻胃管或胃造口餵食；手指或手腕的易位需要復健、輔具或以手術校正；物理、職能治療則可提升患者的各項發展。 此外，心臟疾病問題與惡性腫瘤有可能會造成患者生命的危險，因此需要定期追蹤血壓變化、心臟功能、腫瘤是否變大，疾病的各項症狀若能早期發現和治療，患者的生命週期幾乎與常人相當。（節錄自台灣罕見遺傳疾病一點通網站 http://www.genes-at-taiwan.com.tw/genehelp/）

Costello Syndrome

CS is an extremely rare disorder that affects multiple organ systems of the body. This condition is characterized by loose skin (cutis laxa) on the neck, palms, fingers, and soles. In some cases, certain areas of the skin may become unusually dark (hyperpigmentation). Individuals with CS will have developmental delay and intellectual disability; and a characteristic facial appearance. There is an increased incidence of congenital abnormalities of the heart. Affected individuals have an increased lifetime risk to develop specific malignant tumors. CS is an autosomal dominant genetic condition caused by mutations in the HRAS gene. Approximately 350 affected individuals have been reported worldwide. Infants with CS typically exhibit poor sucking ability, have swallowing difficulties, and fail to grow and gain weight at the expected rate. The treatment of CS is directed toward the specific symptoms that are apparent in each individual. Early intervention is important to ensure that children with CS reach their potential. Services that may be beneficial include special remedial education, speech therapy, special social support, and other medical, social, and/or vocational services. (source from NORD : http://rarediseases.org/rare-diseases/costello-syndrome/)

罕盟#新友 Welcome, Kenny !

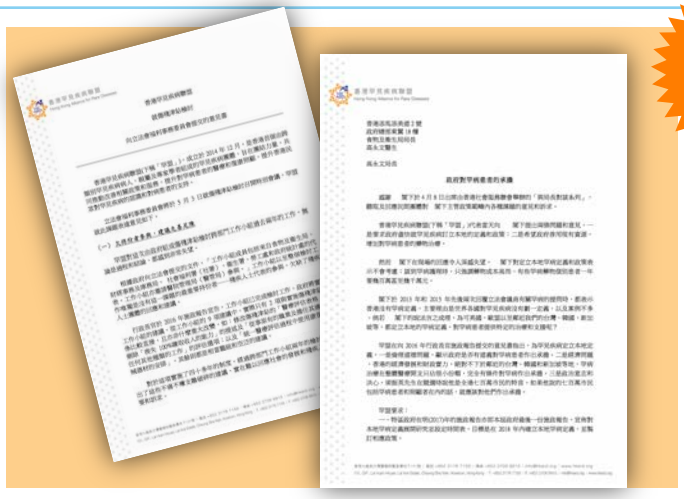
父親的心願 A Father's Wish

在剛過去的父親節， Kenny 與家人的訪問於 NOW 電視播放了。Kenny 是罕盟的新會員。 他是一個四人家庭的爸爸，愛惜家人。 幼子悅悅只有三歲，今年四月確診克斯提洛氏彈性蛋白缺陷症 (Costello Syndrome, CS)。三年的求診路上，與太太既徬徨又擔心，為著一對子女，必須鼓起勇氣向前行。 然而，要帶著兒子進出多間醫院覆診、政府沒有積極的罕病管理策略、緊缺的社會資源支援罕病家庭等；全是他們的障礙，使他們極度疲憊。雖然悅悅已被確診而 CS 仍未能有藥物根治，但是這能代表他沒有獲得社會的支持，發輝潛能的權利嗎？

On the past Father's Day, Kenny's interview was broadcasted on NOW TV. Kenny Leung is HKARD's new member, a father from family of four. His 3-yr old little son, Yuet-yuet, was diagnosed with Costello Syndrome in April. Eventhough their son was diagonosed with this disease, full of hesitation and worries, Kenny & his wife still continues walking forward with hope & courage. Obstacles like mutli-hospitals follow ups, lack of proactive RD management policy in HK, the limited resources available to support RD family in the community, etc, made Kennny extremely exhausted. Although Yuet-yuet has already been diagnosed with CS, which is an RD that curretly has no effective treatment, does that mean he shouldn't have the right to receive support from the community to help him develop his full potential and to contribute the society in the future?

罕盟 # 預告 HKARD #Notice

- 07/2016 ： 請留意！香港結節性硬化症協會將有一個 TSC 公眾教育 LED 展示燈箱設於中環行人扶手電梯傍。TSCAHK will have a TSC disease education LED advertising box located by the side of the pedestrian escalator at Central. Please take a look.
- 09/2016 ： 繼續跟進並爭取新一屆立法會衞生事務委員會立案商討罕病政策事宜。 Legco Health Panel follow up on RD policy discussion.
- 29/10/2016：香港演藝學院及香江粵樂社合辦「香江粵調 -- 香江粵樂社成立音樂會」免費招待罕盟會友，敬請留意電郵報名。Hong Kong Cantonese Music Society will provide free tickets to HKARD members & friends to go for Cantonese Music Charity Concert in HKAPA. Please pay attention to our upcoming e-mail.
- 25/02/2017 ： 世界罕見疾病日 2017 研討會， 敬請留意詳情。WRDD 2017 Seminar & Patient Workshop tentatively will be held, please pay attention to further announcement.



cont.

罕盟#匯報
HKARD #Activity Report

罕盟病友傳媒訪問 Media Featuring

4-6 月 Apr to Jun

罕病教育宣傳 RD Education

2 月 - 友邦保險 Feb, at AIA

5 月 - 艾伯維生物製藥公司 May, at AbbVie

罕盟於上半年應邀參加了兩場機構罕病教育講座。希望有關行業能對罕病及罕盟的工作加深認識。再次感謝友邦保險郭卓華先生團隊和艾伯維生物製藥公司的邀請和支票捐助。 HKARD was invited to deliver two sessions of education talks on RD and to introduce the role of HKARD in the first half of 2016. We hope to increase the support of RD patients in relevant industry sectors in HK. Thank you for the invitation and cheque donations of AIA (Mr Stephen Kwok's Team) and AbbVie to HKARD.

罕盟面書 4 月面世了！ HKARD Facebook Fan Page was launched in April

罕盟會努力恆常於面書發放本地及海外罕病資訊，請多給我們「讚」啊！ HKARD will try our very best to keep frequent update of local and overseas RD information via this platform. Please do remember to give us a "Like"！

香港罕見疾病聯盟 - Hong Kong Alliance for Rare Diseases

請進入我們的面書，網上登記成為罕盟一份子吧！Act now to submit your HKARD membership application online via our fan page !

罕盟#政策倡議
HKARD # Policy Stance

1. **04/04/2016** 罕盟代表出席「會見食物及衞生局局長高永文醫生」活動。罕盟要求 (一)高局長在現任政府任期內、展開訂定罕病定義和政策的工作。(二) 要求醫院管理局善用每年 4,500 萬元為非常見疾病提供藥物補助的政府恒常撥款，把受惠病人從現時六種需要替代酵素治療的患者擴及至更多罕病類別。高永文局長在回應時，再次拒絕考慮訂立罕病定義；在談到罕病治療時，他只強調藥價高昂，沒有作出具體承諾，令人失望。**Representatives of HKARD met Dr Ko Wing Man, SFH, in the "Meeting the Secretary" public forum. HKARD urge SFH to (1) kick off the work on establishment of RD definition and its related policy by the end of his serving term; and (2) ensure the full utilization of the $4.5 billion yearly budget for uncommon dieseases by extending its eligibility to more treatment for rare diseases. No positive replied from Dr Ko was received.**

2. **19/04/2016** 立法會福利事務委員會於 5 月 3 日就傷津檢討召開特別會議，罕盟起草並提交意見書。罕盟就此課題表達意見如下：(一)欠持份者參與，建議乏善足陳；(二) 有必要全面審視殘疾定義及發放傷津目的；(三) 應由跨專業評審申請個案；(四) 罕病患者申領傷津時出現的阻滯。**Legco's Panel on Welfare Services hosted a special meeting on evaluation of current disable allowance policy on 3 May. HKARD submitted opinions as follows : (1) lack of stakeholder's presence in the task force, the recommendation is uninspiring; (2) to re-evaluate the definition of "disable" and the aim of the issue of allowance thoroughly is critical; (3) case application should be reviewed by cross-sector professionals; (4) pay attention to the factors that hinder the application of disable allowance for RD patients.**

3. **26/04/2016** 罕盟去信食物及衞生局局長高永文醫生，促請政府局負對罕病患者的承擔。（一）對本地罕病定義展開研究並設定時間表，目標是在 2018 年內確立本地罕病定義，並製訂相應政策。（二）善用現時政府對醫院管理局每年 4,500 萬元非常見疾病藥物的恒常撥款， 於今年內再增加六種罕見疾病藥物，擴大受惠患者範圍和人數。**HKARD wrote letter to Dr WM Ko, SFH, to urge the government to take up her utmost responsibility to (1) have a time table to start up the investigation work in establishing a rare disease definition for HK. The objective is to have RD definition be stated and the subsequent policy can be developed in 2018. (2) to fully utilize the HK$4.5 billion annually granted budget for uncommon disease treatment by extending the coverage to six more rare disease drug items within this year.**

會長曾建平出席國際罕見疾病聯盟年會及歐洲罕見疾病及孤兒藥品會議簡報

Highlights from Mr KP Tsang after attending Rare Disease International (RDI) 2nd Annual Meeting and The European Conference on Rare Diseases and Orphan Products (ECRD) in May

國際罕見疾病聯盟年會 RDI 2nd Annual Meeting

第二屆國際罕見疾病聯盟年會於 5 月 25 日在蘇格蘭愛丁堡舉行。在是次年會上，國際罕盟提出以下重點工作：

1. 擴大會員，國際罕盟現有會員機構 36 個。
2. 提升認知，在全球範圍內提升社會各界對罕見疾病的認知和關注。
3. 與持份者協作，國際罕盟已積極與聯合國經濟及社會理事會(UN ECOSOC)溝通及討論，在聯合國的平台設立一個關注全球罕病的委員會，結合聯合國制定可持續發展目標(Sustainable Development Goal)的進程，讓各地罕病組織反映意見和訴求，在政府層面推動對罕病的關注。國際罕盟並與其他持份者包括病人組織、科研機構、監管部門等協作，共同達致改善罕病患者的醫療和生活質素。
4. 倡導罕病成為公共衛生優先議題，國際罕盟將在世界衛生組織展開倡導和游說工作，爭取把罕見疾病列為全球公共衛生的優先議題
5. 為會員提供能力建設，國際罕盟將積極為會員機構提供培訓和能力建設，包括提供資訊及網上講座(Webina)等，以提升會員機構在全球各地的倡導工作。

RDI 2nd Annual Meeting was held on 25 May in Edinburgh of Scotland. Key focus of RDI's upcoming mission is as follows :

1. Further engage of new members. There are 36 member societies currently.
2. Increase the worldwild awareness of rare disease.
3. Pay active collaboration with RD stakeholders. Both in the United Nation's Sustainable Development Goal platform as well as local government levels. Objective is to improve the situation of RD patients both in clinical and quality of life aspects.
4. Drive RD issues as a public health agenda to be considered in higher priority in World Health Organization.
5. To raise the leading role of RDI members in their home place by actively support relevant advance technology and infrastructure, eg. Webina, etc.

有關國際罕盟及是次年會的詳細資料，可到以下連結閱覽。 Please refer to the below link for further information.
http://www.rarediseasesinternational.org/membership

歐洲罕見疾病會議

第八屆歐洲罕見疾病及孤兒藥品會議(European Conference on Rare Disorders and Orphan Products)於 5 月 26 至 28 日在蘇格蘭愛丁堡舉行。會議由歐洲罕見疾病聯盟(EURORDIS)主辦。 這是由罕病患者組織舉辦的大型重要會議，每兩年一次，近年已從地區性發展成為全球性的罕病會議，聚集罕病患者、照顧者、科研人員、臨床專家、政府官員、藥劑行業等，共同探討如何改善和加強對罕病的治療和支援。本屆會議出席者近 800 人，其中 500 多名是來自全球各地 48 個國家的病人組織代表。會議從診斷、治療、研究、藥物開發、社會支援等各方面，探討如何改善和加強對罕病患者的支援。除全體大會外，我主要參與了關於病人組織的工作坊，獲益不淺。現將下列點滴與大家分享。

1. 罕病不罕見。蘇格蘭衛生部長在大會開幕致詞時表示，蘇格蘭政府按照英國政府衛生部於 2013 年發表的《英國國家罕病策略》，制定了蘇格蘭罕病工作計劃，名為 《罕病不罕見》(It's Not Rare To Have Rare Disease)，從診斷、治療、預防、社會支援等各方面勾劃出政府的政策和具體行動。詳情可到蘇格蘭政府網頁瀏覽。
2. 全球罕病患者溝通網絡(rareconnect.org)。這是由歐洲罕病患者和照顧者發起的病人溝通網絡，對象是全球各地的患者和照顧者，協助建立各類罕病群體和合作伙伴。
3. 尊重患者的人權和保障他們生活質素，在全體大會上，一位專門研究聯合國《殘疾人權利公約》的英國教授指出《公約》不是昂貴的治療藥物，不需要再研究開發，但如得以落實，卻是尊重罕病患者固有人權和保障他們的生活質素的綱領和內容。
4. 設立個案經理以評估、設計及協調跨專業的治療護理服務。 加拿大政府推行了一項試驗計劃，設立個案經理， 負責統籌所有與罕病患者治療、護理和復康的部門和單位，對個別罕病患者進行需要評估，然後制定護理計劃，隨而統籌協調跨界別跨部門的協作，為患者提供一條龍的服務。計劃成效顯著。
5. 病人參與罕病藥物的研究開發過程。 會議鼓勵和呼籲罕病患者投身參與藥物開發的全過程，與科學家和藥劑行業協作，共同開發適用於病人的藥物。
6. 政府資助病人購買專業護理服務。 荷蘭政府對罕病家庭提供資助，讓他們自行購買罕病子女所需的護理服務。
7. 牛津研究團隊希望與香港合作。 該團隊正進行一種新的開放研究模式，希望與全球各地的科研和臨床專家合作，共享科學資源，共同研究開發罕病藥物。

ECRD was held in Edinburgh, Scotland, on 26-28 May. It is a every two-year meeting organized by EURORDIS. Stakeholders of RD all over the world got together to discuss on RD issues for the purpose of enhancing the support and treatment of RD. There were around 800 participants and 500 of them were patient group representatives from 48 countries. There were symposia and workshops for participants to attend. Key discussion and learning is listed as follows:

1. "It's Not Rare To Have Rare Disease" – specific policy and working plan were launched in Scotland base on UK Strategy for Rare Diseases. Please access Scotland Government Website for details.
2. Rareconnect.org – a connection between patients & caregivers in a worldwide base.
3. Respect the rights of RD patients and preserve their quality of life – a British Professor who is expert in "Convention on the Rights of Persons with Disabilities" stated that the Convention is not a costly medication, no prolong research and trial is required, once it can be practiced, it is the content of patient rights and their living quality.
4. Case Manager – a pilot run of Case Manager of RD launched in Canada. Excellent outcomes were achieved as resources are effectively allocated cross-functionally in a one-stop-shop approach. The care is specific and fit well into the actual needs of the particular patient.
5. Patient's participation in R&D of drug – patients are encourage to partner with scientists or pharmaceutical industry on R&D projects of new drug development.
6. Government's support on purchase of professional healthcare services – Holland Government implements schemes to support RD families to purchase healthcare services for maintaining quality family life.
7. Oxford research team's co-operation with HK's experts – the team stated that they were opened to work in partner with experts all over the world for the sake of effective resources utilization and information sharing on RD research projects.

會議發表了大量具質素的論文和簡報，在歐洲罕病聯盟網頁可供瀏覽下載。Abundant high quality essays and bulletins were released in the conference, please visit EURORDIS website for review & download : http://www.rare-diseases.eu/abstracts/。

罕盟 2016 活動贊助機構 2016 HKARD Activity Sponsors:

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