SPRING SPECIAL ISSUE (2016) 春季號外

News of World Rare Disease Day 2016 Symposium

2016 世界罕病日「愛不罕見」研討會

罕情 #RareCare 號外

香港罕見疾病聯盟會訊 Newsletter of HKARD March 2016 (Issue No.1.1)

愛，真的不罕見！

Thank you for the support of all our guests and partners !

罕盟 2016 世界罕病日「愛不罕見」研討會已於 2 月 28 日在灣仔聖雅各福群會順利舉行。大會很榮幸邀請到立法會衞生服務界議員李國麟教授作主禮嘉賓並致賀詞。當天來賓超過 170 人，出席率達九成。共有十七個罕病病友組織的代表到場支持。到訪傳媒有 10 個，翌日媒體報導有 23 篇。 罕盟希望透過此次研討會教育及提昇公眾及決策者對罕病的關注，逐步爭取社會在資源和政策上加強對罕病患者的支持。

HKARD WRDD 2016 Symposium – Care is not Rare was held successfully on 28 February 2016 (Sunday), at St. James Settlement, Wan Chai, H.K. We are pleased to have Prof Hon Joseph Lee, LegCo member, as our officiating guest to support us. Over 170 guests participated; show up rate was over 90%. RD patients/carers from 17 patient groups joined us. Journalists from 10 local media have attended. Total 23 media exposure on the next day. We hope that it will be a big step forward to raise RD attention in our community within the public and our policy makers for the purpose of a fair and justified allocation of resources to RD patients.

到場支持機構：
Our supporting organizations :

謝謝以下 17 個組織的代表來臨支持！
We were proud to have representatives from 17 organizations to attend the Symposium ! They are :

1. 香港天使綜合症基金會 HK Angelman Syndrome Foundation
2. Fragile X Hong kong
3. 香港醫護聯盟 Hong Kong Health Care Alliance
4. 夏約書孤兒症基金會 Joshua Hellmann Foundation for Orphan Disease
5. 小而同罕有骨骼疾病基金會 Little People of HK
6. 香港紅十字會甘迺迪中心校友會 HK Red Cross John F.Kennedy Centre Alumni Association
7. 香港黏多醣症暨罕有遺傳病互助小組 HK Mucopolysacchairidosis & Rare Genetic Disease Mutual Aid Group
8. 香港肌健協會 HK Neuro-Muscular Disease Asso.
9. 香港病人組織聯盟 HK Alliance of Patients' Organizations Limited
10. 香港病人政策連線 Hong Kong Patients' Voices
11. PNH 病人權益關注組
12. 香港視網膜病變協會 Retina Hong Kong
13. 香港雷特氏症協會 HK Rett Syndrome Association
14. 香港知足協會 HK Slix Society
15. 香港小腦萎縮症協會 HK Spinocerebellar Ataxia Asso
16. 脊椎性肌肉萎縮症慈善基金 Families of SMA Charitable Trust
17. 香港結節性硬化症協會 Tuberous Sclerosis Complex

主講嘉賓 Fabulous speakers !

鍾侃言醫生
Dr Brian Chung, HKU

罕盟科學及醫療諮詢委員會主席／香港大學李嘉誠醫學院兒童及青少年科學系臨床副教授

陳浩然教授
Prof Edwin Chan, CUHK

香港中文大學生命科學系教授

李焯翹先生
Mr Johnson Lee, HK PolyU

香港理工大學社會工作博士候選人

主禮嘉賓 Officiating Guest
立法會衞生服務界議員
李國麟教授
Prof Hon Joseph Lee,
Legislative Council Member, HKSAR

大會精華剪影 Snapshots

當中大陳浩然教授(左)遇上港大陳振勝教授(右)。
When Prof Edwin Chan CUHK (L) met with Prof Danny Chan, HKU (R).

罕盟會長 曾建平先生致歡迎詞
Mr KP Tsang, President, HKARD, delivered welcome remarks.

中文大學教授王紹明醫生和李志光醫生。
We have Prof Raymond Wong and Prof CK Li of CUHK attended the meeting .

香港知足協會創會會長
鍾愛寶小姐。Ms Chung Oi Po, Founding President of HK Patients' Voices. Voices.

香港病人政策連線主席林志釉先生。
Mr Alex Lam, Chairman of HK Patients' Voices.

罕盟副會長 方緯谷先生作大會總結。
Mr Dennis Fong, Vice-president, HKARD, concluded the Symposium

World Through My Eyes

嘉賓與講者於'World Through our Eyes'展板前留影。展板以亞洲區不同的罕病個案為主題，由'Rainbow Across Borders Asia'提供。(www.rabasia.org)

Guests and Speakers in front of the 'World Through our Eyes' display panel , a project of Rainbow Across Borders Asia. It is an exhibit with the theme of : Rare Disease - A Photographic Journal featured RD patients stories in Asia. (www.rabasia.org)

新聞稿
Press Release

2016 世界罕病日「愛不罕見」研討會暨新聞發佈會

HKARD 2016 World Rare Disease Day Symposium cum Press Conference - Care is not Rare

(香港，2016 年 2 月 28 日) 按世界衛生組織資料顯示，每 1 萬人當中有 6.5 至 10 個個案的疾病便定為罕病。有不少罕病是由於基因突變或遺傳基因缺陷導致的。人體約有 25,000 至 30,000 個基因，婚孕的男女如有同一隱性致病的基因或是某一方有家族遺傳病史，又或是基因偶發地產生突變，他們的下一代有可能出現基因異常的罕見疾病。

香港中文大學陳浩然教授稱：「以小腦萎縮症為例，遺傳機制是染色體顯性遺傳，即一對基因中只要其中一個存在致病的突變便會引起病徵。致病的基因突變可以由男或女性患者傳到下一代。每一個第二代都有五成機會遺傳到致病的基因突變，男女遺傳機會相同。」

罕病　是否罕見？

據 2013 年優先藥物用於歐洲和世界報告[1]指出，全球約有 7,000 種罕病，大約每 15 人有 1 人患上此病，受影響的兒童高達七成五，而有三成兒童更在五歲生日前確診患有罕病。

由此推算，香港最少有 400,000 人受罕病影響，當中絕大多數是家族性基因遺傳。鑑於目前本港的醫療體制對罕病還未定義，亦未成立疾病及病人名冊，故在醫療及社福上的支援相當有限；然而，罕病對患者造成身心長期的創傷，以及照顧者背負的沉重負擔卻是無限。

食物及衛生局局長高永文先生在 2013 年立法會上稱：「目前，國際間對於罕見疾病並沒有一致的定義…由於罕見疾病的個案數目不容易掌握，關於其病因的可靠資料或數據亦不足，而部分疾病之治療方法獲發現的時間尚短，醫管局現時並沒有就罕見疾病作出定義。」

早期診斷　控制病情

香港有國際認可的公共衛生政策及高質素的醫療系統，但對罕病的認知、防治及保障卻遠遠落後於歐美及亞洲鄰近等地區。

香港大學李嘉誠醫學院兒童及青少年科學系臨床副教授鍾侃言醫生稱：「以前本地的基因診斷還未成熟，樣本一般會送往外國化驗，再加上醫生對罕病的認知不足，患者出現症狀時只能找相關的專科醫生確診；如醫生對罕病有足夠認知，便可及早將病人轉介接受治療。」一直以來，政府以病人多寡作為分配資源準則，因此一般常見疾病會得到較多資源；相比病友人數少的罕病，所得到的支援卻較少。鍾侃言醫生稱：「我們期望罕病有更多資源，特別是兒童醫院將於 2018 年興建，若在此設立遺傳基因診所，便可讓病童及他的家人一併接受治療。」

罕見疾病兒童家庭的社會需要－香港首項研究初步結果

由香港理工大學社會工作博士候選人李焯翹先生進行。研究目的：一）探討香港罕病兒童家長所面對的壓力與困境；二）檢視家長在照顧上的需要和如何面對困境；三）讓醫護專業人士和政府瞭解病者和家人的需要，以訂定醫療、復康、教育、福利及社會參與的政策。研究以個案型式邀請 12 個家庭作深入訪談（家中有 18 歲或以下的罕病兒童）。研究顯示他們均面對醫療（如確診、資訊及知識）、家長的心理情緒壓力、家庭成員間的關係、家庭經濟、照顧者的壓力、社區支援、兒童成長發展（包括就學及社會接納／歧視）等困擾。

(Hong Kong, 28 February 2016) According to The World Health Organization's findings, Rare Disease (" RD") is defined as the prevalence of 6.5 to 10 disease cases for every 10,000 of population. RD cases are commonly due to genetic mutation or resulted from genetic defects. The anatomy has about 25,000 to 30,000 genes. If a pregnant wife and husband carry the same recessive pathogenic genes or either partner has a history of hereditary disease, or perhaps due to sporadic occurrences of gene mutation, their next generation is likely to exhibit genetically abnormal RD.

Professor Edwin Ho-Yin Chan, CUHK says: "Take spinocerebellar ataxia, SCA, for instance, it is autosomal dominant inheritance in terms of genetic mechanisms, meaning just one gene in a pair with pathological mutation is sufficient for triggering symptoms. Pathological gene mutation can be passed down to the next generation either from the father or mother. Each offspring has a 50% chance of receiving pathological gene mutation, and this genetic probability is the same for both males and females."

Rare Disease　Does it rare?

With reference to the World Health Organization 2013[1] "Priority Medicines for Europe and Around the World" reported that, **there are approximately 7,000 rare diseases worldwide. About 1 in every 15 persons is a rare disease patient.** Among these patients, affected children constitute up to 75%, and 30% of child patients receive positive diagnoses before their 5th birthday.

Therefore, **it is inferred there are at least 400,000 people affected by rare diseases in Hong Kong**, and among them most cases are due to familial genetics. In view of the absence of any rare disease definition in Hong Kong's health care system, while rare disease patient registry and rare disease classification have yet to be established, hence medical and social welfare supports are severely limited. However, rare diseases bring about long-term physical and psychological trauma for sufferers, as well as endless heavy burden on carers.

In 2013, Dr Ko Wing Man, Secretary for Food and Health, once cited in Legislative Council:

"Presently, there is no unanimous definition in the international community regarding rare diseases..., due to difficulties in ascertaining rare disease case statistics, while reliability of information and data concerning underlying causes is questionable, and also treatment options for some of these diseases have just been recently devised, the Hospital Authority currently has not determined any definition regarding rare diseases."

Early Diagnosis　Better Control

Hong Kong possesses internationally recognized public health policies and exceptional medical systems. However, prevention and safeguard measures against RD are lagging far behind than those implemented in Europe, the USA and neighboring Asia regions.

Dr Brian Hon-Yin Chung, Clinical Associate Professor, Department of Paediatrics and Adolescent Medicine, LKS Faculty of Medicine, HKU says: "Previously, when local genetic diagnosis was not well developed, samples would usually be sent overseas for testing. In addition, doctors did not have sufficient knowledge about rare diseases, patients exhibiting symptoms could only search for related specialists for diagnosis. If doctors have satisfactory knowledge about rare diseases, patients can be expediently referred for due medical treatment." The government has been following the criterion of patient numbers in resource allocation, therefore general common diseases receive more resources. On the contrary, available support for rare disease patients is relatively inadequate due to their fewer numbers.

Dr Chung says, we wish to strive for more resources for rare diseases, and particularly the operation of Hong Kong Children's Hospital to be commenced in 2018, with the possibility of setting up a genetics clinic within the location, thereby facilitating simultaneous treatment of RD child and their family members.

The social needs of families with RD child patients - preliminary first research findings in Hong Kong

By Mr Johnson Lee. Doctor of Social Work candidate, The Hong Kong Polytechnic University Research Objectives: (1) To examine the stress and difficulties encountered by the patients in taking care of children with rare diseases in Hong Kong; (2) To determine the caring needs and coping strategies adopted by the parents in taking care of the children; (3) To inform health care professionals and policy makers of the supports and needs of children with rare diseases and their families in policy advocacy, health care, rehabilitation, education, welfare and social participation.

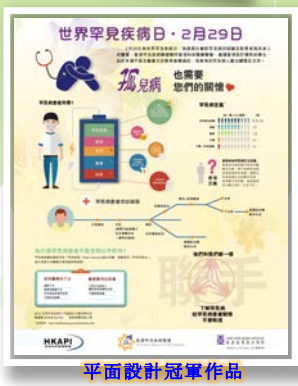
It is a qualitative cases study. Data collected by face-to-face semi-structured interviews with 12 families which have rare disease child under age of 18 years old. The research findings revealed that all interviewed families were suffered stress and difficulties on:

- Medical aspects, eg. diagnosis, information and knowledge;
- Psychological and emotional stress;
- Spouse and family relationships;
- Care management;
- Resources, eg. Social support and financial stress.
- Child development, schooling and social acceptance / stigmatization

2016 世界罕見疾病日 平面及動畫設計比賽
Infographic Design Contest for World Rare Disease Day 2016

罕見疾病的發生率非常低但對生命卻造成威脅。目前大眾對罕病的認知甚少。透過是次比賽，希望以香港知專設計學院(HKDI)學生的創意思維及創新表達方式，增加大眾對罕病的認知。比賽由香港罕見疾病聯盟(HKARD)和香港科研製藥聯會 (HKAPI)協辦，並得到有線電視支持成為官方媒體的合作夥伴。

Rare disease is a life-threatening disease with a very low incidence rate. Currently, general public is unaware or have very little knowledge about rare diseases. With an aim to raise public awareness on the situation of rare disease in Hong Kong, this contest serves to explore new way of presentation to educate public on rare diseases through the creative minds of the diploma students of Hong Kong Design Institute (HKDI). This contest is co-organized by the Hong Kong Alliance for Rare Diseases (HKARD) and the Hong Kong Association of the Pharmaceutical Industry (HKAPI). This meaningful program also won support by Cable TV as an official media partner.



平面設計冠軍作品
Work of Infographics Winner

頒獎嘉賓與得獎者(圖左至右):
知專設計學院副院長暨署理學術總監 (設計) 林偉強先生、香港科研製藥聯會執行董事 陳素娟女士、平面設計得獎者：**季軍** 羅穎思、**亞軍** 楊玉梅 、**冠軍** 陳嘉寶、動畫設計**得獎組別** ：**呂芷彤、甘俊君、罕盟會長** 曾建平先生、罕盟科學及醫療諮詢委員會主席 鍾侃言醫生。

Award Presenters & Awardees (from left to right) :
Mr Wallace Lam, Vice-principal of HKDI and Acting Academic Director (Design), Ms Sabrina Chan,Executive Director, HKAPI, Infographics Award : Lo Wing Sze **(2nd Runner-up)**, Yeung Yuk Mui **(1st Runner-up)** and Chan Ka Po **(Winner)**; **Best Motion Work** : Lui Tsz Tung & Gan Junjun; Mr KP Tsang, President HKARD, Dr Braing Chung, Chairman, Scientific & Medical Advisory Committee, HKARD.

得獎及參賽作品可瀏覽 Please click the below link for student's works :
https://sites.google.com/site/del14102a/home/classwork
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https://drive.google.com/folderview?id=0B0qmETls150_RmV5UkhTWUdUc00&usp=sharing

罕盟組織架構 The Organizational Structure of HKARD

罕盟第一屆理事與研討會講者(圖左至右):鍾侃言醫生、黃敏兒女士、李焯翹先生(講者)、陳浩然教授(講者)、梁七根先生、陳振勝教授、黃婉冰女士、賴家衞先生、曾建平先生、方緯谷先生及林文珊女士 (項目主任)。 導盲犬 :Deanna。

1st Council Members of HKARD and Symposium Speakers (from left to right) :
Dr Brian Chung, Ms Amy Wong, Mr Johnson Lee (speaker), Prof Edwin Chan (speaker), Mr Leung Chat Kan, Prof Danny Chan, Ms Maria Wong, Mr Terry Lai, Mr KP Tsang, Mr Dennis Fong & Patti Lam (Project Officer). KP's guide dog : Deanna

第一屆罕盟理事已於 1 月 23 日的會員大會選出。 理事會並於 2 月的會議中確定了組織架構。The 1st Council Members where elected in the AGM on 23rd Jan 2016. 1st Council Meeting was held on 2nd Feb 2016 and the organizational structure of HKARD Council is as follows :

會長 President	:	曾建平先生 Mr KP Tsang
副會長 Vice President	:	方緯谷先生 Mr Dennis Fong
義務秘書 Hon. Secretary	:	曹綺雯女士 Ms Tso Yee Man
義務司庫 Hon. Treasurer	:	梁七根先生 Mr Leung Chat Kan

內務委員會 House Committee :
主席 Chairman : 方緯谷先生 Mr Dennis Fong
成員 Members :
曹綺雯女士、梁七根先生、賴家衞先生、曾建平先生和何立明先生
Ms Tso Yee Man, Mr Leung Chat Kan, Mr Terry Lai, Mr KP Tsang & Mr Ho Lap Ming

外務委員會 External Affairs Committee :
主席 Chairman : 曾建平先生 Mr KP Tsang
成員 Members :
方緯谷先生、周權棣先生、賴家衞先生、黃敏兒女士、黃婉冰女士、陳振勝教授、曹綺雯女士、何立明先生和熊德鳳女士
Mr Dennis Fong, Mr Roanld Chow, Mr Terry Lai, Ms Amy Wong, Ms Maria Wong, Prof Danny Chan, Ms Tso Yee Man, Mr Ho Lap Ming & Ms Anchor Hung

科學及醫療諮詢委員會 Scientific & Medical Advisory Committee :
主席 Chairman : 鍾侃言醫生 Dr Brian Chung
成員 Members :
陳振勝教授、曾建平先生、方緯谷先生、周權棣先生和黃婉冰女士
Prof Danny Chan, Mr KP Tsang, Mr Dennis Fong, Mr Ronald Chow & Ms Maria Wong

顧問 Advisors :
車錫英教授、陳麗雲教授、陳浩然教授、許鍾妮教授、王紹明教授。
Prof SY Chair, Prof Cecilia Chan, Prof Edwin Chan, Dr Joannie Hui & Dr Raymond Wong.

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