罕病研討會 2024
Rare Disease Symposium 2024

合辦機構 Co-organisers

香港罕見疾病聯盟
RARE DISEASE HONG KONG

HKU Med
LKS Faculty of Medicine
The University of Hong Kong
香港大學李嘉誠醫學院

香港大学深圳医院
The University of Hong Kong - Shenzhen Hospital

北京協和醫院
PEKING UNION MEDICAL COLLEGE HOSPITAL

中国罕见病联盟
China Alliance for Rare Diseases

罕病研討會 2024
程序表
Rare Disease Symposium 2024
Programme Rundown

(I) 開幕儀式 Opening Ceremony

地點 Venue：7 樓宴會廳 A Nina Ballroom Hall A, 7/F
語言：普通話及英語（設即時傳譯） Languages: Putonghua & English（with simultaneous interpretation）
主持 Moderator：陳振勝教授（香港大學李嘉誠醫學院資深顧問(政策及管治)）
Prof. Danny CHAN, (Senior Advisor (Policy & Governance), LKS Faculty of Medicine, The University of Hong Kong)

時間 Time	程序 Programme	講者 Speaker
08:00 – 08:30	登記入場 Registration	
08:30 – 08:35	致賀辭 Congratulatory remarks	*中國香港醫務衞生局局長 盧寵茂教授* *Prof. LO Chung-mau, Secretary for Health, Hong Kong, China*
08:35 – 08:40	致歡迎辭 Welcoming remarks	*香港大學副校長（健康）、李嘉誠醫學院院長 劉澤星教授* *Prof. LAU Chak-sing, Vice-President & Pro-Vice-Chancellor (Health), and Dean of Medicine, The University of Hong Kong*
08:40 – 08:45	致歡迎辭 Welcoming remarks	*中國罕見病聯盟執行理事長 李林康先生* *Mr. LI Linkang, Executive Director, China Alliance for Rare Diseases*
08:45 – 08:50	致謝辭 Vote of thanks	*香港罕見疾病聯盟會長 曾建平先生* *Mr. TSANG Kin-ping, Chairman, Rare Disease Hong Kong*
08:50 – 09:00	大合照 Group photos	

(II) 主題演講 Keynotes

地點：7 樓宴會廳 A　　Venue: Nina Ballroom Hall A, 7/F
語言：普通話及英語（設即時傳譯） Languages: Putonghua & English（with simultaneous interpretation）

時間 Time	主題 Topic	講者 Speaker
09:00 – 09:20	中國罕見病診療研究體系建設 Rare Disease Diagnosis and Treatment Research in China	*北京協和醫院院長 張抒揚教授* *Prof. ZHANG Shuyang, President, Peking Union Medical College Hospital*
09:20 – 09:40	香港罕見病服務發展 Rare Disease Service Development in Hong Kong	*中國香港醫院管理局質素及安全總監黃立己醫生* *Dr. Michael WONG, Director (Quality & Safety), Hospital Authority, Hong Kong,China*
09:40 – 10:00	香港大學深圳醫院的罕見病服務發展 Rare Disease Service Development in The University of HK-Shenzhen Hospital	*香港大學深圳醫院代表(待定)* *Representative, The University of Hong Kong-Shenzhen Hospital (TBC)*
10:00 – 10:20	彌合全球承諾和區域現實：區域在塑造世界衛生大會決議對罕病的影響的角色 Bridging Global Commitments and Regional Realities: The Region's Role in Shaping the Impact of the World Health Assembly Resolution on Rare Diseases	*Ms. Alexandra HEUMBER-PERRY, Chief Executive Officer, Rare Diseases International* *(國際罕見疾病聯盟行政總裁)*
10:20 – 10:50	**茶歇 Tea Break**	

(III) 科學主題講座：從實驗室到臨床
Science Keynotes: From Laboratory to Clinic

地點：7 樓宴會廳 A　　Venue: Nina Ballroom Hall A, 7/F

語言：普通話及英語（設即時傳譯）Languages: Putonghua & English（with simultaneous interpretation）

時間 Time	主題 Topic	講者 Speaker
10:50 – 11:10	基於基因組學的罕見病健康服務 Diagnostics and Treatment Development in Hong Kong for Rare Diseases	*香港基因組中心首席醫務及科學總監 鍾侃言醫生* *Dr. Brian CHUNG, Chief Medical and Scientific Officer, Hong Kong Genome Institute*
11:10 – 11:30	軟骨發育不全的基礎科學及轉化研究 Basic Science and Translational Study on FGFR3-related Osteochondrodysplasia	*Prof. Laurence LEGEAI-MALLET, Director of Research, French National Institute for Health and Medical Research, Imagine Institute, Université de Paris Cité* *(法國國家健康與醫學研究所研究主任)*
11:30 – 11:50	北京協和醫院罕見病醫學科建科 Rare Disease Service Development at Peking Union Medical College Hospital	*北京協和醫院罕見病醫學科 常務副主任(主持工作) 沈敏教授* *Prof. SHEN Min, Deputy Executive Director, Department of Rare Diseases, Peking Union Medical College Hospital*
11:50 – 12:10	針對臨床基因組定序後仍未確診的罕見病個案臨床整合多組學計畫 RDNow 的成效 Outcomes of a Clinically Integrated Multi-omic Rare Disease Programme, RDNow, for Individuals who Remain Undiagnosed after Clinical Genomic Sequencing	*Prof. Tiong Yang TAN, Clinical Geneticist, Victorian Clinical Genetics Services, Murdoch Children's Research Institute, Australia* *(澳洲默多克兒童研究所維多利亞臨床遺傳學服務中心臨床遺傳學家)*
12:10 – 12:50	**圓桌討論 Round-table discussion** 主題 Topic: 從實驗室到臨床 From Laboratory to Clinic 主持 Moderator: *陳振勝教授 Prof. Danny CHAN*[1] 討論嘉賓 Panelists: *Prof. Nick BISHOP*[2] *, Prof. Tae-Joon CHO*[3] *, 馮卓穎醫生 Dr. FUNG Cheuk-wing*[4] *, Prof. Laurence LEGEAI-MALLET*[5] *, Prof. Tiong Yang TAN*[6] *, 杜啟峻教授 Prof. Michael TO*[7]	
12:50 – 14:00	**午膳時間 Lunchtime**	

[1] *Senior Advisor (Policy and Governance), LKS Faculty of Medicine, The University of Hong Kong 香港大學李嘉誠醫學院資深顧問(政策及管治)*

[2] *Chair in Paediatric Bone Disease, Clinical Medicine, School of Medicine and Population Health, University of Sheffield, UK 英國謝菲爾德大學醫學與人口健康學院臨床醫學小兒骨病主任*

[3] *Professor, Department of Orthopaedic Surgery, Seoul National University College of Medicine, Korea 韓國首爾國立大學醫學院骨科教授*

[4] *Consultant (Metabolic Medicine), Department of Paediatrics and Adolescent Medicine, Hong Kong Children's Hospital 香港兒童醫院兒童及青少年科顧問醫生(先天性新陳代謝科)*

[5] *Director of Research, French National Institute for Health and Medical Research, Imagine Institute, Université de Paris Cité 法國國家健康與醫學研究所研究主任*

[6] *Clinical Geneticist, Victorian Clinical Genetics Services, Murdoch Children's Research Institute, Australia 澳洲默多克兒童研究所維多利亞臨床遺傳學服務中心臨床遺傳學家*

[7] *Head, the Orthopedic Centre, The University of Hong Kong-Shenzhen Hospital 香港大學深圳醫院骨科醫學中心主任*

（IV） 分論壇 Parallel Sessions

科學分論壇 (一) Science breakout 1：
罕見的神經系統疾病 Rare Neurological Disorders

地點 Venue：11 樓 2 號室 Rm 2, 11/F　　語言 Language：全英語 English only

Moderator： Dr. FUNG Cheuk-wing（Consultant (Metabolic Medicine), Department of Paediatrics and Adolescent Medicine, Hong Kong Children's Hospital）

Time	Topic	Speaker
14:00 – 14:20	Experience Sharing on Diagnosis and Management of Duchenne Muscular Dystrophy in China	*Prof. DAI Yi, Deputy Director, Department of Neurology, Peking Union Medical College Hospital*
14:20 – 14:40	DLC1 isoform 1 (DLC1-i1) as a potential gene therapy for Spinal Muscular Atrophy	*Prof. Martin CHEUNG, Associate Professor, Associate Director (Research & Innovation), School of Biomedical Sciences, LKS Faculty of Medicine, The University of Hong Kong*
14:40 – 15:00	Overview on Neurocutaneous Syndrome	*Dr. LUK Ho-ming, Chief of Service, Department of Clinical Genetics, Hong Kong Children's Hospital*
15:00 – 15:20	Treatments of Neurocutaneous Syndromes	*Prof. Godfrey CHAN, Honorary Clinical Professor, Department of paediatrics and Adolescent Medicine, School of Clinical Medicine, LKS Faculty of Medicine, The University of Hong Kong*
15:20 – 15:30	Q&A and Panel Discussion	
15:30 – 15:50	Personalized Treatment for Neurological Rare Diseases	*Prof .Sophelia CHAN, Clinical Associate Professor, Department of Paediatrics and Adolescent Medicine, School of Clinical Medicine, LKS Faculty of Medicine, The University of Hong Kong*
15:50 – 16:10	Spinal Muscular Atrophy and Duchenne Muscular Dystrophy: Development of New Treatments	
16:10 – 16:30	Management Update of Neuromyelitis Optica Spectrum Disorders	*Dr. XIAO Haibing, Associate Consultant, Department of Neurology, The University of HK-Shenzhen Hospital*
16:30 – 16:50	Treatment Options in Neurometabolic Diseases	*Dr. Sheila WONG, Associate Consultant, Neurology team, Department of Paediatrics and Adolescent Medicine, Hong Kong Children's Hospital*
16:50 – 17:10	Lysosomal Storage Diseases in Adults	*Dr SHENG Bun, Consultant, Department of Medicine and Geriatrics, Princess Margaret Hospital*
17:10 – 17:20	Q&A and Panel Discussion	

(IV) 分論壇 Parallel Sessions

科學分論壇 (二) Science breakout 2：

罕見的骨骼發育異常與骨科疾病 Rare Skeletal Dysplasia & Orthopaedic Disorders

地點 Venue：11 樓 3 號室 Rm 3, 11/F　　語言 Language：全英語 English only

Moderator：Prof. Janus WONG（Clinical Assistant Professor, Department of Orthopaedics & Traumatology, School of Clinical Medicine, The University of Hong Kong）

Time	Topic	Speaker
14:00 – 14:20	Pseudoachondroplasia / Multiple Epiphyseal Dysplasia	*Prof. Tae-Joon CHO, Department of Orthopaedic Surgery, Seoul National University College of Medicine, Korea*
14:20 – 14:40	Orthopaedic Management in Skeletal Dysplasia	
14:40 – 15:00	Diagnosis and Treatment of Prader Willi syndrome Scoliosis	*Prof. ZHANG Jianguo, Director, Department of Orthopedics, Peking Union Medical College Hospital*
15:00 – 15:20	Novel Treatments for Osteogenesis Imperfecta	*Prof. Nick Bishop, Chair in Paediatric Bone Disease, Clinical Medicine, School of Medicine and Population Health, University of Sheffield, UK*
15:20 – 15:30	Q&A and Panel Discussion	
15:30 – 15:50	Clinical Approach to Skeletal Dysplasia	*Prof. Michael TO, Head, the Orthopedic Centre, The University of HK-Shenzhen Hospital*
15:50 – 16:10	Updates on the Treatment of Bone Mineralisation disorders	*Dr. Joanna Yuet-ling TUNG, Consultant (Endocrinology), Department of Paediatrics and Adolescent Medicine, Hong Kong Children's Hospital*
16:10 – 16:30	Updates of Treatment for Achondroplasia	*Prof. Laurence LEGEAI-MALLET, Director of Research, French National Institute for Health and Medical Research, Imagine Institute, Université de Paris Cité*
16:30 – 16:50	Limb Lengthening for Patients with Skeletal Dysplasia ~ To Do or Not to Do	*Prof. Janus WONG, Clinical Assistant Professor, Department of Orthopaedics & Traumatology, School of Clinical Medicine, The University of Hong Kong*
16:50 – 17:00	Q&A and Panel Discussion	

（IV） 分論壇 Parallel Sessions

患者組織分論壇 Patient Organisation Sub-forum **地點 Venue：11 樓 4 號室 Rm 4, 11/F** **語言 Languages：普通話及廣東話（設即時傳譯）Putonghua & Cantonese（with simultaneous interpretation）** **主持：李林康先生（中國罕見病聯盟執行理事長）**	
14:00-14:10	歡迎辭
主題一：開展科學和社會研究	
14:10-14:20	開場簡介
14:20-14:30	**Dravet 綜合症共患病研究實踐與思考** 伍思嘉女士 — 卓蔚寶貝支持小組（北京）（Dravet 綜合症病友組織）
14:30-14:40	**科患之橋 O 填補空白** 鄭嫚女士 — 成都紫貝殼公益服務中心（四川成都）（硬皮病 / 系統性硬化症病友組織）
14:40-14:50	**患者組織參與藥物研發** 王芳女士 — 泡泡家園神經纖維瘤病關愛中心（深圳市）
14:50-15:00	**「高林綜合征 — 因何而得名」~ 進入第二批罕見病目錄的十年** 仲冬蕾女士 — 並蒂蓮高林綜合征聯盟（河北唐山）
15:00-15:10	問答與交流
主題二：多元創新的宣傳教育	
15:10-15:15	開場簡介
15:15-15:25	**大海撈針** 陳寶婷女士 — 香港視神經脊髓炎協會
15:25-15:35	**罕有不罕見** 阮佩玲女士 — 香港結節性硬化症協會
15:35-15:45	**堅持做小胖威利罕見病傳播的力量在哪裡？** 林曉靜先生 — 小胖威利罕見病關愛中心（浙江杭州）
15:45-15:55	**不可能的「單手」任務 — 打破常規** 賴佩欣女士 — 牽手同行協會（香港）（先天性手部缺失病友組織）
15:55-16:05	**以行為藝術（秀）將銅娃娃進行到底** 晨冰先生 — 銅娃娃罕見病關愛中心（肝豆狀核變性罕見病關愛協會）（河南鄭州）
16:05-16:15	問答與交流

（IV） 分論壇 Parallel Sessions

患者組織分論壇 Patient Organisation Sub-forum 地點 Venue：11 樓 2 號室 Rm 2, 11/F 語言 Languages：普通話及廣東話（設即時傳譯）Putonghua & Cantonese（with simultaneous interpretation） 主持：李林康先生（中國罕見病聯盟執行理事長）	
主題三：回應患者的需要和要求	
16:15-16:20	開場簡介
16:20-16:30	**血管水腫護照** 黃耀文先生 — 香港遺傳性血管水腫協會
16:30-16:40	**回歸初心 O 同心同行** 呂文林先生 — 香港肌健協會（神經肌肉疾病病友組織）
16:40-16:50	**同馨相印 O 共創未來** 嚴青女士 — 同馨肝豆信息服務中心（湖北武漢）（肝豆狀核變性 / 威爾遜氏症病友組織）
16:50-17:00	**不讓隱形罕病侵害患者** 陳娜珍女士 — 紅米粒高膽固醇血症研究與服務中心（陝西西安）
17:00-17:10	**基於患者需求 – 戈謝病親子互動遊戲項目介紹** 王軍先生 — 山西壹心公益發展中心（山西太原）（戈謝病 / 高雪氏症病友組織）
17:10-17:20	問答與交流

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