

罕病研討會 2024

Rare Disease Symposium 2024

罕建網絡，罕建未來
Living Connected, Brightens Living with the Rare

日期Date : 30 – 11 – 2024 (六Sat)

時間Time : 08:30 – 17:30

地點Venue : 荃灣西如心酒店 (香港新界荃灣楊屋道8號)

Nina Hotel Tsuen Wan West (No. 8 Yeung Uk Rd,
Tsuen Wan, NT, Hong Kong)

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



北京协和医院
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	香港醫務衛生局局長 盧寵茂教授 Prof. LO Chung-mau, Secretary for Health, Hong Kong
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	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
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科主任

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	<i>Prof. DAI Yi, Deputy Director, Department of Neurology, Peking Union Medical College Hospital</i>
14:20 – 14:40	DLC1 isoform 1 (DLC1-i1) as a potential gene therapy for Spinal Muscular Atrophy	<i>Prof. Martin CHEUNG, Associate Professor, Associate Director (Research & Innovation), School of Biomedical Sciences, LKS Faculty of Medicine, The University of Hong Kong</i>
14:40 – 15:00	Overview on Neurocutaneous Syndrome	<i>Dr. LUK Ho-ming, Chief of Service, Department of Clinical Genetics, Hong Kong Children's Hospital</i>
15:00 – 15:20	Treatments of Neurocutaneous Syndromes	<i>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</i>
15:20 – 15:30	Q&A and Panel Discussion	
15:30 – 15:50	Personalized Treatment for Neurological Rare Diseases	<i>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</i>
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	<i>Dr. XIAO Haibing, Associate Consultant, Department of Neurology, The University of HK-Shenzhen Hospital</i>
16:30 – 16:50	Treatment Options in Neurometabolic Diseases	<i>Dr. Sheila WONG, Associate Consultant, Neurology team, Department of Paediatrics and Adolescent Medicine, Hong Kong Children's Hospital</i>
16:50 – 17:10	Lysosomal Storage Diseases in Adults	<i>Dr SHENG Bun, Consultant, Department of Medicine and Geriatrics, Princess Margaret Hospital</i>
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 ○ 填補空白 鄭嫺女士 — 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 ○ 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	同聲相印 ○ 共創未來 嚴青女士 — 同聲肝豆信息服務中心（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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