



HONG KONG COLLEGE OF PAEDIATRICIANS

Committee for Subspecialty Boards

Forum on the Impact of Hong Kong Children's Hospital on
the future paediatric training and service

Proposal of the service model & training programme of Metabolic Medicine

Date : 24 September 2016 (Saturday)

Time : 2:00 pm – 5:50 pm

Venue : Function Room 1, 2/F , HKAM Jockey Club Building

HK Society Inborn errors of metabolism

- Established in 2004
- Started by few paediatricians, chemical pathologists & geneticists who saw the need of developing IEM services in HK
- a platform for discussion & collaboration among various specialists
- more diagnostic investigative tools are being developed locally
- obsoletes the previous dependence on overseas laboratories
- the diagnostic yield of various metabolic diseases greatly enhanced
- metabolic clinics set up at 4 HA hospitals
- PWH, QMH, QEH & PMH





香港黏多醣症暨罕有遺傳病互助小組

Hong Kong Mucopolysaccharidoses & Rare Genetic Diseases Mutual Aid Group

► 按此為首頁 本會出版的多本書籍已製作成電子書，部份更加添了簡體字版，歡迎大家免費下載。




主頁
關於我們
黏豆
本會出版
會員應有的書籍
媒體
活動
網上教材
「腦有大作為」比賽
支持我們
聯絡我們

我們的故事

這是一顆豆豆的故事，也是一夥人的故事。

夏天來了，園子裏的豆豆落到泥土上，等待發芽。黏豆和每一粒豆一樣，每天努力的吸收養份、曬太陽、做運動，夢想著長大。

可是，黏豆一直沒長高...



...「長不大、活不長」是醫生對他發出的不祥預言。

黏多醣症的故事，大都是這樣開始。黏多醣症和其他罕有遺傳病一樣，產前難預知，治療費用高昂，讓很多病人和病人的父母心力交瘁。是基因牽他們開玩笑？還是命運出題考驗他們的毅力？

他們的身軀很小，夢想卻很大很大，總是努力的向前走。



請按左邊的欄目選擇，認識黏豆，和您可以怎樣幫忙。

本會消息

2015年10月12日
誠邀全港中學派隊參加
「腦有大作為」全港中學生發明大賽！[詳情按此](#)

2015年5月20日
罕有病黏多醣症兄弟求生短片獲
多項國際獎項，病者訴求卻仍然
落空！[詳情按此](#)

2015年5月1日
本會出版的多本書籍已製作成電
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- Established 2005
- mutual support children & their families with rare diseases
- a strong advocate for enzyme replacement therapy for lysosomal storage diseases



Expert panel on Enzyme replacement therapy for rare metabolic diseases



Set up by Hospital Authority 2007

Panel members: HA administrators, Clinicians, Pharmacists

Regular meetings 3-4 times per year

- ▶ To oversee commissioning of ERT in HK
- ▶ To set up treatment guidelines on ERT for specific disease groups
- ▶ To assess relative priorities for funding for ERT
- ▶ To review every new as well as renewal applications
- ▶ To evaluate or facilitate independent evaluations of ERT
- ▶ To appraise new evidence on ERT

LSD patients currently on ERT funded by HA (19 patients/2016)

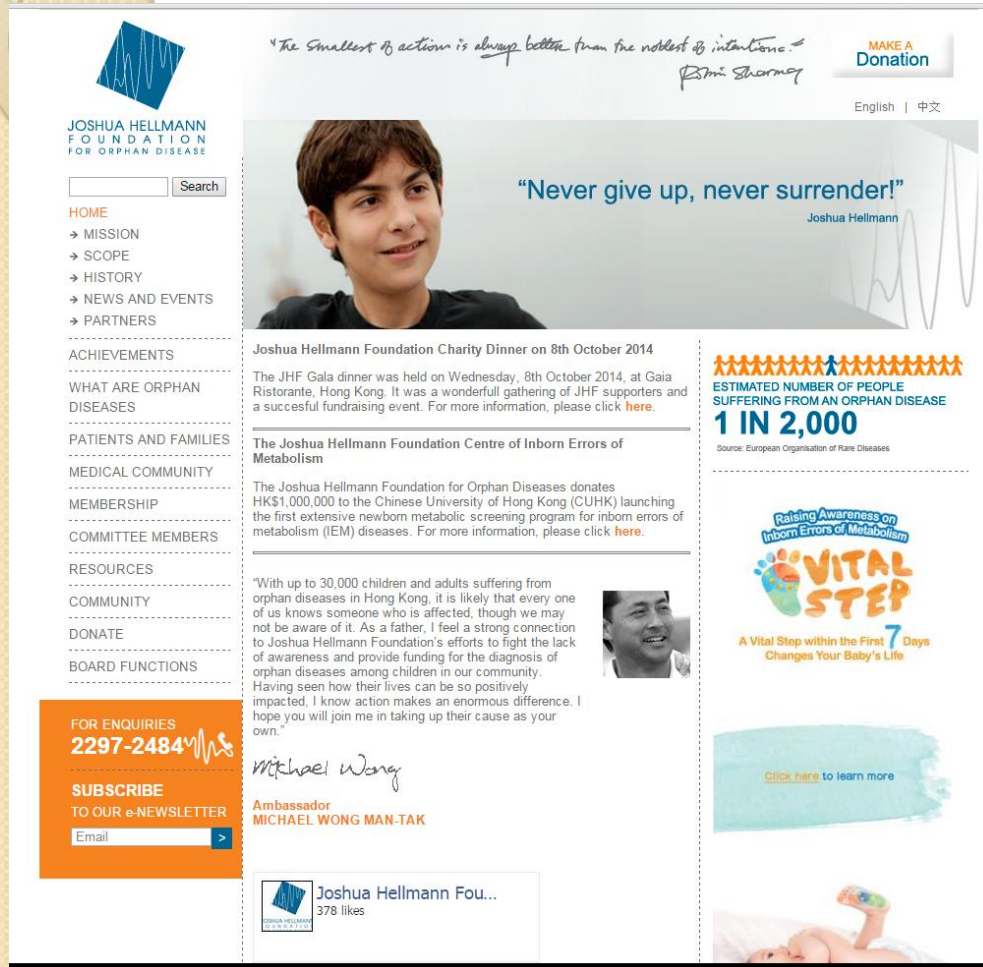
MPS

- 2 MPS I
- 2 MPS VI

Other LSDs

- 8 Pompe (2 infantile, 7 late onset)
- 2 Gaucher
- 5 Fabry

Joshua Hellmann Foundation



The screenshot shows the homepage of the Joshua Hellmann Foundation. At the top, there is a quote by Rumi: "The smallest of action is always better than the noblest of intentions." and a "MAKE A Donation" button. The main header features a photo of Joshua Hellmann with the text "Never give up, never surrender!" and "Joshua Hellmann". Below this, there is a section about the "Joshua Hellmann Foundation Charity Dinner on 8th October 2014". To the right, there is a graphic showing "ESTIMATED NUMBER OF PEOPLE SUFFERING FROM AN ORPHAN DISEASE 1 IN 2,000" with a source from the European Organisation of Rare Diseases. Further down, there is a section titled "Raising Awareness on Inborn Errors of Metabolism" with a "VITAL STEP" logo and a quote from Michael Wong, Ambassador. The left sidebar contains a search bar and a list of navigation links: HOME, MISSION, SCOPE, HISTORY, NEWS AND EVENTS, PARTNERS, ACHIEVEMENTS, WHAT ARE ORPHAN DISEASES, PATIENTS AND FAMILIES, MEDICAL COMMUNITY, MEMBERSHIP, COMMITTEE MEMBERS, RESOURCES, COMMUNITY, DONATE, and BOARD FUNCTIONS. At the bottom left, there is a contact section for enquiries (2297-2484) and a subscribe button for the e-newsletter. The bottom right shows a Facebook-like interface with the text "Joshua Hellmann Fou..." and "378 likes".

Joshua Hellmann Foundation for Orphan Disease

HOME

- MISSION
- SCOPE
- HISTORY
- NEWS AND EVENTS
- PARTNERS

ACHIEVEMENTS

WHAT ARE ORPHAN DISEASES

PATIENTS AND FAMILIES

MEDICAL COMMUNITY

MEMBERSHIP

COMMITTEE MEMBERS

RESOURCES

COMMUNITY

DONATE

BOARD FUNCTIONS

FOR ENQUIRIES
2297-2484

SUBSCRIBE
TO OUR e-NEWSLETTER

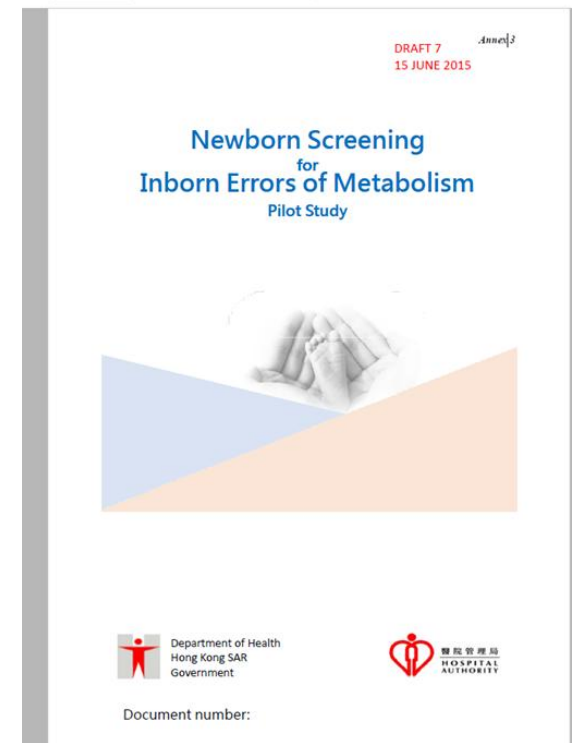
Email

Joshua Hellmann Fou...
378 likes

- Established 2009
- mission of advancing the awareness, diagnosis, treatment and research of orphan diseases
- aims at improving the welfare of children with orphan diseases in Hong Kong
- donation for the establishment of the CUHK pilot NBS program

HKSAR Government Newborn screening for IEM

- Announced in Chief Executive's 2015 Policy address
- Task force set up in 2015
- Members from both Department of Health & Hospital Authority
- Obstetricians, Paediatricians, Chemical Pathologists, Clinical Geneticists, Maternity Child Health clinics
- Pilot study
- rolled out 1st Oct 2015 at 2 birthing units (QMH & QEH)
- extension into territory wide programme for all newborn babies in HK by 2 phases
- 2017-8 (PWH, TMH , KWH)
- 2018-9 (PMH, UCH, PYNEH)



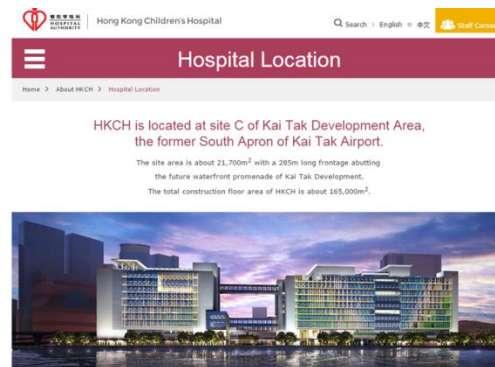
IEM patients: Complex, Serious, Rare

IEM patients

- the rarity and complex nature of IEM requires an integrated specialised clinical & laboratory service to provide satisfactory diagnosis & management

HK Children's Hospital

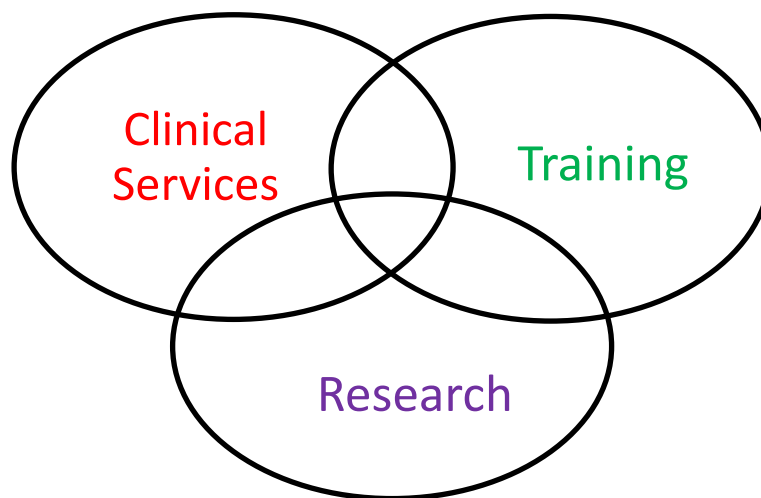
- a highly specialised tertiary care children's Hospital provides the services that these patients need



Current paediatric service provision has therefore to be reviewed to determine how the services can be reconfigured and redistributed to facilitate the development of the overall paediatric service network.

- 'Golden' opportunity to centralize & upgrade the standard of care for patients & families affected with IEM
- The Metabolic Medicine division will become the territory's tertiary referral as well as coordinating centre for the diagnosis & management of all IEM conditions

Metabolic Medicine Division Hong Kong Children's Hospital



- Build on the ground work taken place in the last two decades
- Better streamlined, seamless, quality, equitable & sustainable service
- Transition care : more IEM patients are reaching adulthood, important to involve and work with adult metabolic physicians to facilitate smooth transition between the paediatric and adult services & complement each other in the overall care of IEM patients

Aims & objectives : Clinical services (I)

Aims

- to identify & diagnose patients suspected of having an IEM
- to improve life expectancy & quality of life for children & their families affected by IEM

Objectives

- provide dedicated in & outpatient facilities to IEM patients
- provide high quality dietary &/or drug treatment and care comparable to international standards
- provide consultative services to other subspecialties at HKCH, paediatric services at regional services/ private sectors in the diagnostic workup & management of suspected IEM patients
- outreach clinics/ shared stepdown care
- ensure smooth transition from paediatric to adult care

Aims & objectives : Clinical services (II)

- ensure smooth implementation of territory wide expanded IEM newborn screening
- to evaluate & appraise the clinical & cost-effectiveness of tandem mass spectrometry based newborn IEM screening & feedback information to all the stakeholders
- compile local IEM registry : ensure accurate coding & classification of IEM disorders with constant maintenance & update
- registry data essential for determining population incidence/prevalence, correct management, providing information on outcome and directing research

Interdependencies with other services: Networking

Laboratories

- Specialized IEM laboratory services & other diagnostic tests crucial
- Newborn screening laboratory

IEM patients need multidisciplinary care

- Anesthesiology
- Cardiac
- Endocrine
- Genetics
- Hematology Oncology
- Hepatobiliary Gastrointestinal
- Intensive care
- Neonatology
- Neurology
- Ophthalmology
- Orthopedics
- Radiology
- Renal
- Respiratory
- Surgery (ENT, Cardiothoracic, Neurosurgical)
- Other specialties need support for diagnosis of possible IEM & joint care

Aims & objectives : Training & Education

- THE local training centre for future IEM specialists
- support the training for all healthcare staff (medical, laboratory, nursing, dietetic, allied health professionals, pharmacist) involved in the delivery of IEM services
- provide expert advice to non-medical professionals including local authorities, non governmental organisations NGOs and the voluntary sectors, to facilitate holistic care for IEM patients and support to their families/carers
- provide age-appropriate written and/or electronic material, including provision of information in the patient/family' s first language, relating to the IEM condition to patients and their families/carers

Subspecialty accreditation with HK College of Paediatricians

- initial submission as a combined subspecialty Paediatric Endocrinology & Metabolic Medicine (PEMM) 3/2013
- College advised against combining 2 subspecialties
- re-submission with Paediatric Endocrinology (PE) as individual subspecialty 9/2016 with IEM training included as an optional (highly recommended) module for PE trainees
- ongoing planning for Metabolic Medicine to re-submit to College as a separate individual subspecialty
- training centre – HK Children's Hospital
- train IEM specialists and other subspecialists involved in the care of IEM patients

Aims & objectives : Research

- research activities target at the clinical translation in order to improve understanding and patient care
- to generate and publish, aiming at increasing public and professional awareness on local IEM cases, experiences & evidence of effective treatments
- to generate and publish experiences with implementation, statistical data at evaluating & appraising the clinical and cost-effectiveness of tandem mass spectrometry based newborn IEM screening
- to participate in and contribute to international research programmes

Infrastructure requirements (Proposed core team)

- 5 senior medical staff (2 Consultants + 3 Associate consultants)
(to be filled by existing HA & university staff with training & experience in IEM)
- 2 fellows (subspecialist training in IEM or other related subspecialties)
- 1 dietician with training and experience in IEM
- 2 specialty metabolic nurses/ genetic counsellors
- 1 secretary for administrative & clerical support
- 1 research assistant & registry manager

Conclusion

- Metabolic medicine service is ready to take on a new start at HKCH
- HKCH is the ideal setting for taking care of IEM patients & their families with supporting stepdown service by regional hospitals

Our common Goal :

a brighter & more promising future for
all IEM patients & their families