



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

Timeline:

20 years journey of IEM service development



1997

HA
Metabolic
clinics
PWH
PMH
QMH
QEH
TMH



2004

HK
Society
IEM



2005

HK MPS
& Rare Genetic
Disease
Mutual Aid
Group



2007

HA
Expert Panel
on ERT for
Rare
Metabolic
Diseases



2009

Joshua
Hellmann
Foundation
for Orphan
Disease



2015

HKSAR
Pilot
Expanded
NBS

HK Society Inborn errors of metabolism

- established in 2004 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



Local IEM registry data

- published HK Medical Diary 2014

- collaborative effort, voluntary reporting from majority HA Paed units
- retrospective data 1/7/1999 - 30/6/2009
- CDARS, CMS, metabolic clinic log, individual hospital units' database
- <19y on 1st consultation
- Diagnosis ICD-9 codes

Hong Kong Metabolic Registry 2012

Disorders of amino acid and peptide metabolism	109
Disorders of carbohydrate metabolism	54
Disorders of fatty acid and ketone body metabolism	26
Disorders of energy metabolism	59
Disorders in the metabolism of purines, pyrimidines and nucleotides	4
Disorders of the metabolism of sterols	5
Disorders of lipids and lipoprotein metabolism	37
Congenital disorders of glycosylation	3
Lysosomal disorders (excluding Pompe disease)	69
Peroxisomal disorders	22
Disorders of neurotransmitter metabolism	17
Disorders in the metabolism of vitamins and cofactors	35
Disorders in the metabolism of trace elements and metals	45
Disorders in the metabolism of xenobiotics	1
	486



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

► 回家為首頁 本會出版的十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



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。髓豆豆和每一粒豆豆一樣，每天努力的吸收養份、曬太陽、做運動，夢想著長大。
可是，髓豆豆一直沒長高...



...「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，認識髓豆豆，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
本會出版的十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。 [詳情按此](#)



- 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 the ultra-expensive ERT in HK
- ▶ To set up treatment guidelines on ERT for specific disease groups
- ▶ To review every new as well as renewal applications



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

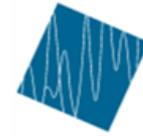
MPS

- 2 MPS I
- 2 MPS VI

Other LSDs

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

Joshua Hellmann Foundation

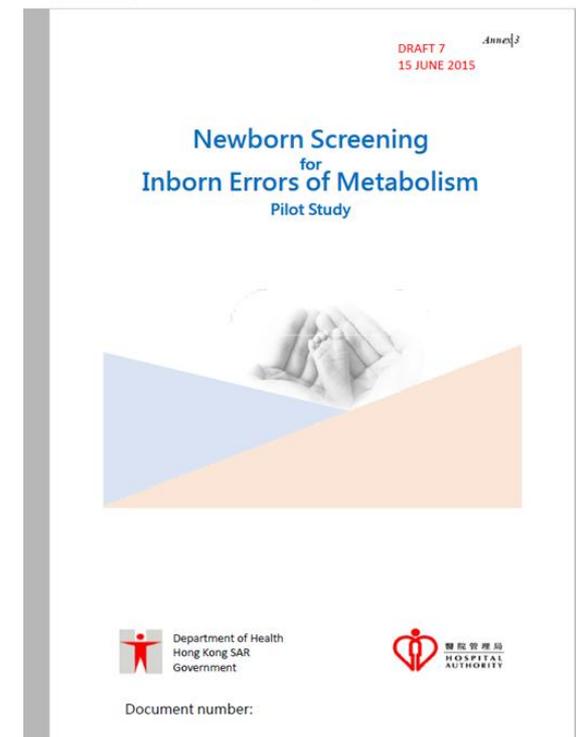


The screenshot shows the website's layout. At the top left is the logo and the text 'JOSHUA HELLMANN FOUNDATION FOR ORPHAN DISEASE'. A quote by Rumi Shorneoy is displayed: 'The smallest of action is always better than the noblest of intentions.' A 'MAKE A Donation' button is in the top right. A search bar is located below the logo. A navigation menu on the left lists: 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. The main content area features a large photo of a young boy with the quote 'Never give up, never surrender!' by Joshua Hellmann. Below this is a news item about a charity dinner on 8th October 2014. To the right, a graphic shows a row of people with one in blue, stating 'ESTIMATED NUMBER OF PEOPLE SUFFERING FROM AN ORPHAN DISEASE 1 IN 2,000'. Below that is a 'VITAL STEP' campaign logo with the text 'Raising Awareness on Inborn Errors of Metabolism' and 'A Vital Step within the First 7 Days Changes Your Baby's Life'. A testimonial from Michael Wong, Ambassador MICHAEL WONG MAN-TAK, is included. At the bottom left, there is a 'FOR ENQUIRIES 2297-2484' contact box and a 'SUBSCRIBE TO OUR e-NEWSLETTER' form. A Facebook-like widget at the bottom right shows 'Joshua Hellmann Fou...' with '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 Expanded Newborn screening for Inborn Errors of Metabolism

- 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



HKSAR Expanded Newborn screening for Inborn Errors of Metabolism



2015

2015/16

2016/17

2017/18

2018/19

Pilot
(Phase 1)

Pilot
(Phase 2)

Extension into territory wide
program in 2 phases

Chief Executive Policy address :

(study the feasibility of trying out in public healthcare system a screening program for newborn babies for IEM)

HA Hospitals	QMH QEH	QMH QEH	PWH KWH TMH	PMH PYNEH UCH
Newborns	Term	All (including preterm & sick term infants)		
No. of IEM	21	24		
% of live births covered (HA)	< 25%	25%	70%	100%
Confirmed IEM	2	2	~15 - 27 new cases/ year	
All serious disorders, often with significant neurological deficit or even fatal if not treated early	<ul style="list-style-type: none"> Carnitine uptake defect PKU 	<ul style="list-style-type: none"> MCAD deficiency Citrin deficiency 	<p>Estimated incidence of IEM 1/2250 - 1/4000</p> <p>Deliveries 60,000/ year</p>	

The Way Forward



1997

Metabolic
Clinics
PWH
PMH
QMH
QEH
TMH



2004

HK
Society
IEM



2005

HK
MPS &
Rare
Genetic
Disease
Mutual
Aid
Group



2007

HA Expert
Panel on
ERT for
Rare
Metabolic
Diseases



2009

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2018

Opening
HKCH

IEM patients & HKCH



IEM patients' needs

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



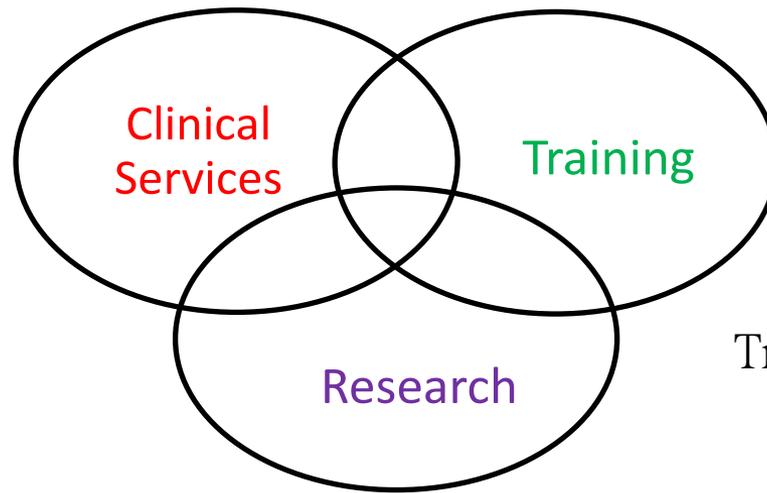
HK Children's Hospital

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



- '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

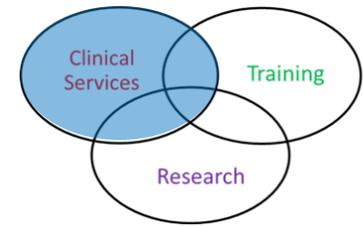
Metabolic Medicine Division Hong Kong Children's Hospital



Tripartite functions

- 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)



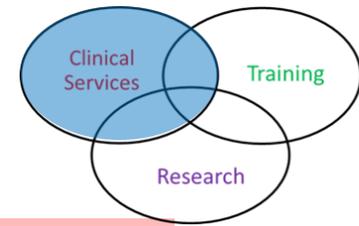
Aims

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

Objectives

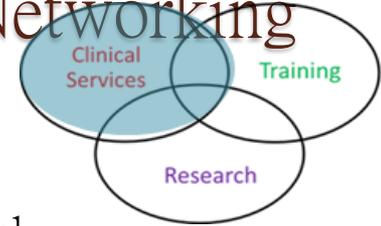
- provide dedicated in & outpatient facilities to IEM patients
- provide high quality dietary &/or drug treatment and care comparable to international standards
- provide consultative services to other subspecialties at HKCH, paediatric services at regional services/ private sectors in the diagnostic 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

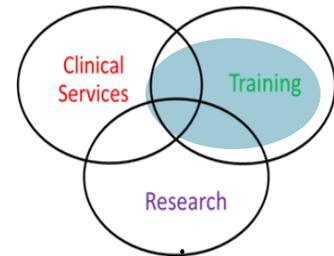
- Anesthesiology
- Cardiac
- Endocrine
- Genetics
- Hematology Oncology
- Hepatobiliary Gastrointestinal
- Intensive care
- Neonatology
- Neurology
- Ophthalmology
- Orthopedics
- Radiology
- Renal
- Respiratory
- Surgery (ENT, Cardiothoracic, Neurosurgical)



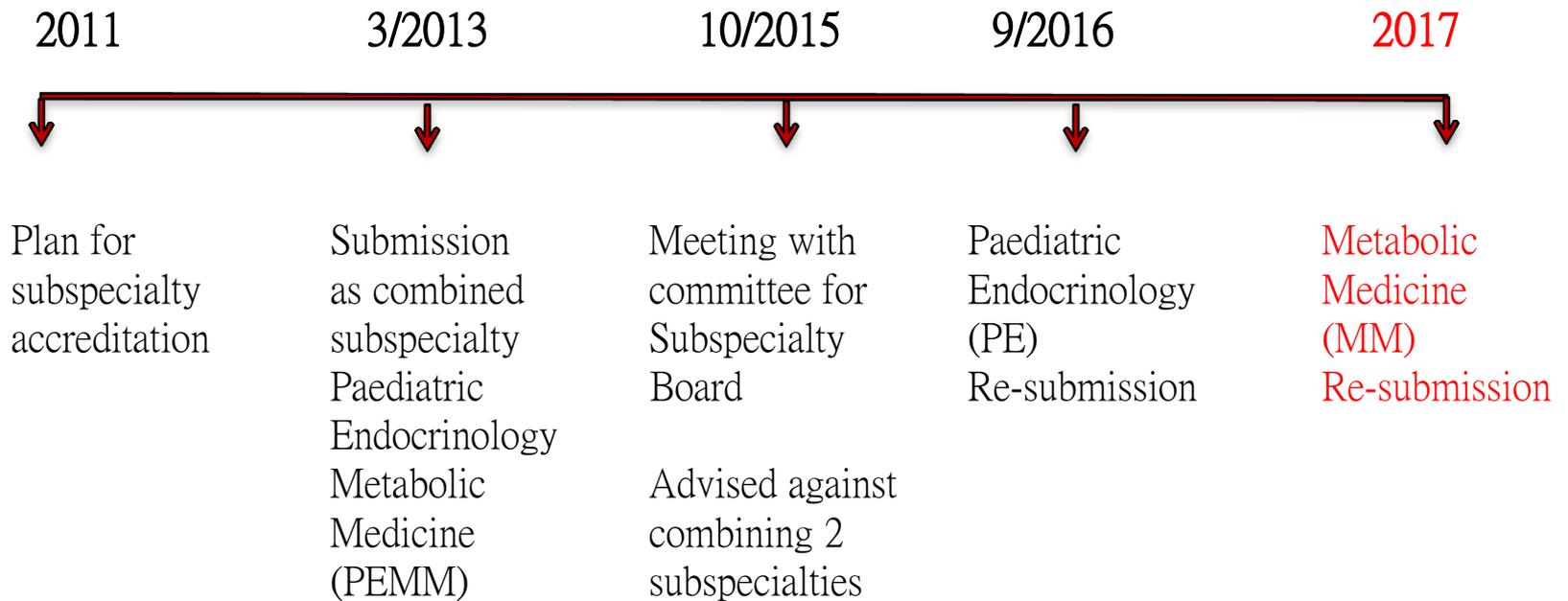
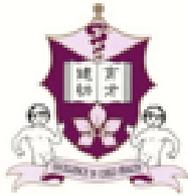
- Other specialties need support for diagnosis of possible IEM & joint care

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



Application for subspecialty accreditation with Hong Kong College of Paediatricians



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

Manpower requirement benchmarking with established overseas IEM centres

	London	Netherlands	The Children' s Hospital at Westmead NSW Australia	HKCH
No. of IEM consultants/ specialists	12	20 6 centres	5	5 - 10
Population (million)	8.63	17	7	7.3
No. of children' s hospital treating IEM	2 GOSH Evelina Children' s Hospital	7 including Amsterdam & Nijmegen	1	1
No. of IEM specialists per million population	1.4	1.4	0.7	0.7 - 1.4

Infrastructure requirements for Metabolic service (Proposed core team at HKCH)



- 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



Latest Stocktake of Manpower for HK IEM service

SSIEM conference
Rome 2016

WE ARE READY



	Metabolic/Endocrine Clinic	Neuro metabolic Clinic	Consultant	SMO / AC	RS	University Staff
PWH	Yes	No	0	1		1
PMH	Yes	Yes	0	1 [^]	0.5	
QMH/DKCH	Yes	Yes	0	2 + 1 [^]	0.5	0.5
QEH	Yes	No	0.5	1		
TMH	Yes	No	0	0.5	0.5	

[^] Neurologist providing IEM and neurometabolic service

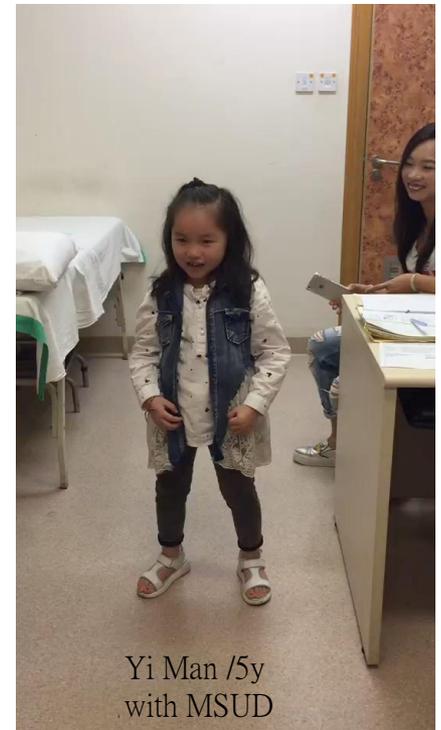
Conclusion

- Metabolic medicine service in Hong Kong is ready to take on a new start at the HKCH
- HKCH is the ideal setting for taking care of IEM patients & their families with hand in hand supporting stepdown care offered by regional hospitals
- At full implementation of expanded newborn screening 2018/19, the coordinating centre will be based at the HKCH (Hub) working closely with the regional obstetric & paediatric units (Spokes)

Our common Goal :
a brighter & more promising future
for all IEM patients & their families



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.



Yi Man /5y
with MSUD