Hospital Authority Convention 2016

3-4 May 2016

Department of Health (DH) and Hospital Authority (HA)'s Pilot Study of Newborn Screening for Inborn Errors of Metabolism

初生嬰兒代謝病篩查先導計劃

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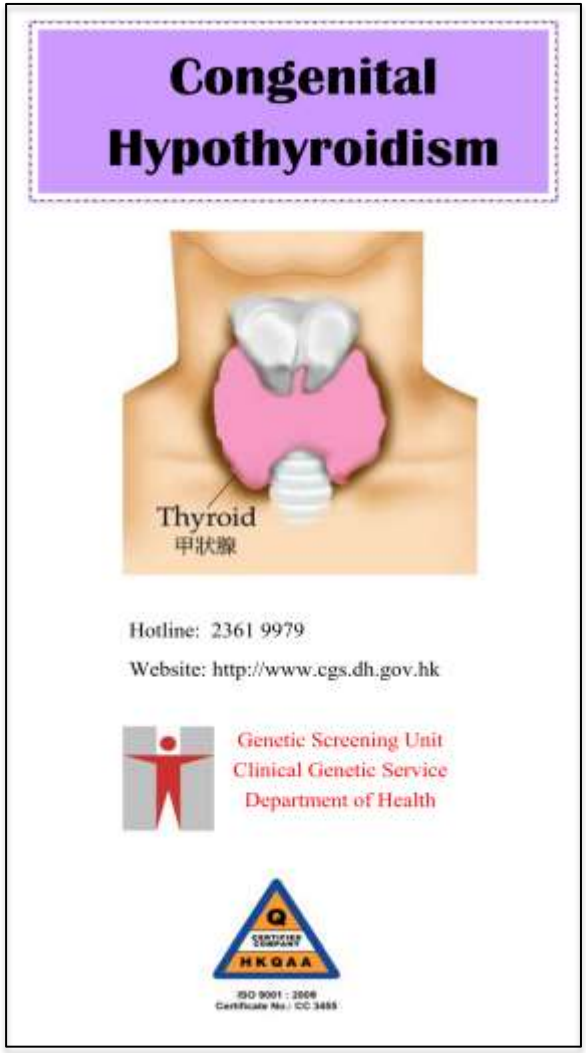
Clinical Genetic Service
Department of Health

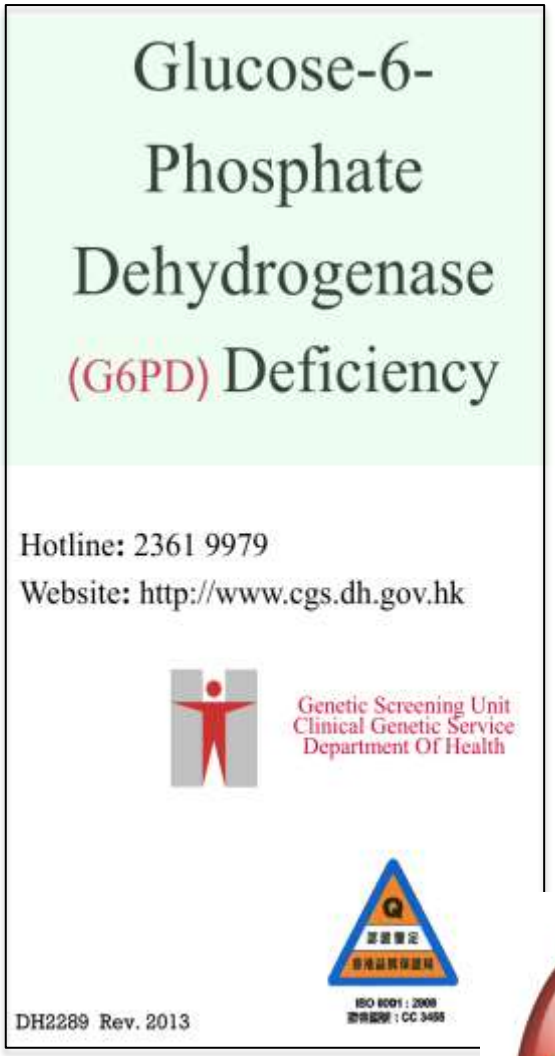
Department of Health
Hong Kong SAR Government

醫院管理局
HOSPITAL AUTHORITY

HK Screening Programme for newborn

- Since 1984
- Free-of-charge service
- Specimen - cord blood





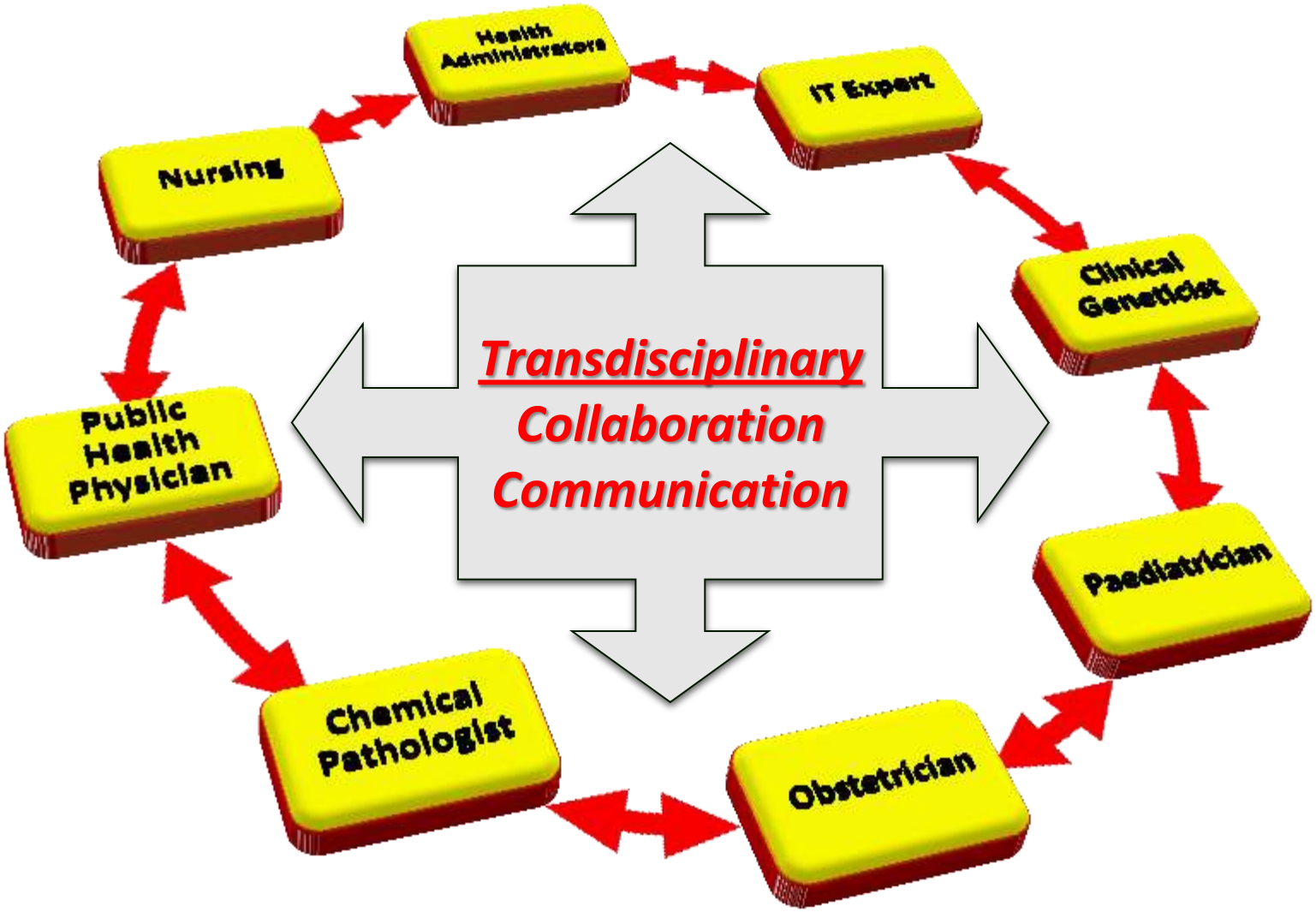
Workgroup

- In 2013
- To review the information and relevant evidence for the expansion of newborn screening programme to cover Inborn Errors Metabolism (IEM)
- Finally decided to recommend to include IEM

Policy Address 2015

Task Force

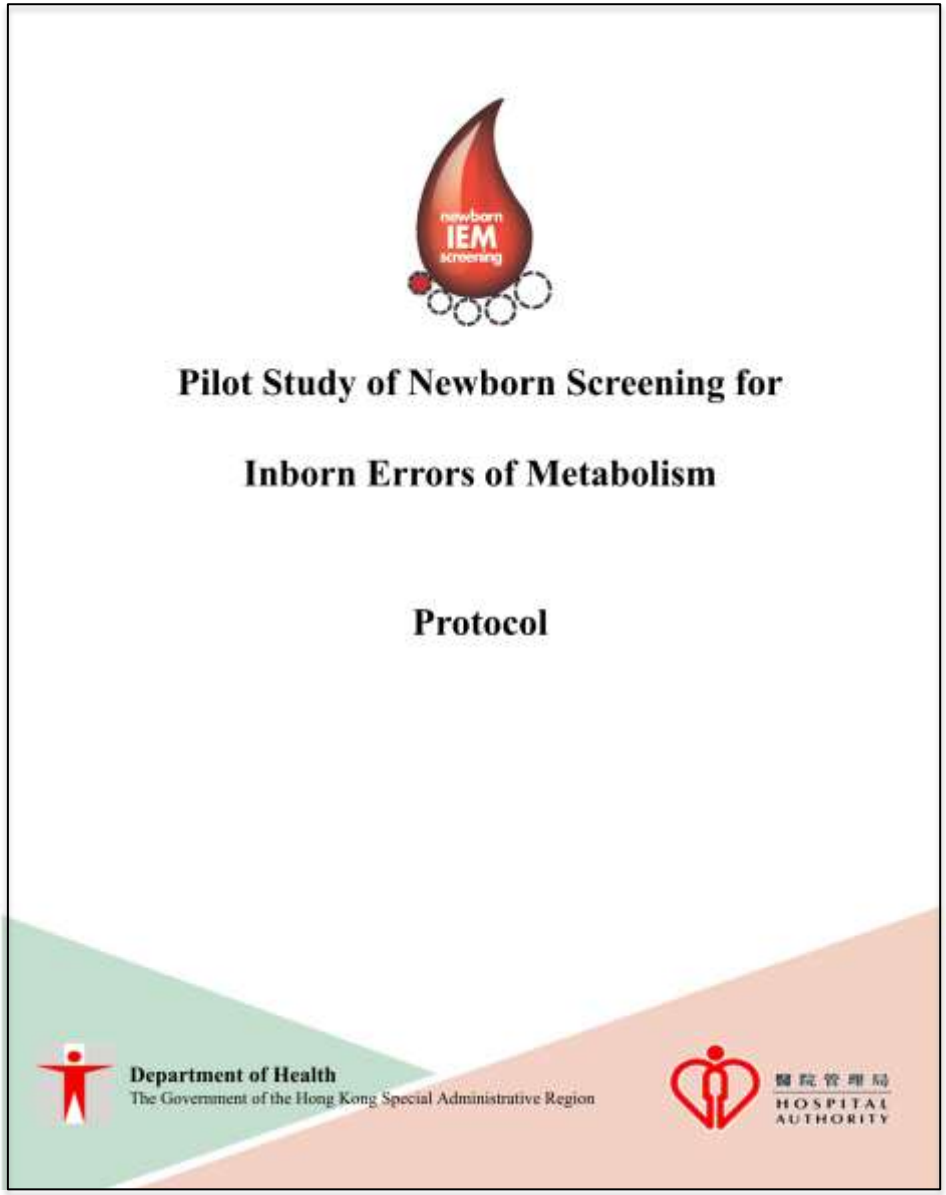
- To plan and prepare for the implementation of a pilot study of newborn screening for IEM



Systematic workflow of NBS

Step	Main task(s)
1. Education to parents	• Information delivery and education about the NBS for IEM
2. Consent	• Obtain written consent (from parents)
3. Specimen collection	• Blood spot collection
4. Dispatch of specimen	• Transportation of dry blood spots to laboratory
5. Dried blood spot preparation and testing	• Specimen preparation and testing in NBS laboratory
6. Reporting	• Results are interpreted and reported by chemical pathologists
7. Recall and repeat testing	• Recall patients with normal /uncertain/ abnrormal results for repeat testing
8. Confirmatory testing	• Diagnostic testing
9. Treatment and monitoring	• Continuous management and monitoring
10. Evaluation	• Assess the outcomes and result of the programme

Protocol - *Standardisation*



Criteria for Inclusion of IEM

- Based on Wilson and Jungner screening criteria, four criteria (from clinical perspective) are proposed to consider the inclusion of IEM conditions

	Criteria	***Elaboration***
1	Screening capability	• availability of accurate and reliable screening and diagnostic testing; and of laboratory capability
2	Clinical significance	• Seriousness and number of cases encountered in our locality
3	Availability of treatment	• efficacy and/or effectiveness of the treatment
4	Favourable outcome after early treatment	• adequacy of the understanding of the natural history of the condition and its long-term outcome with early treatment

Inclusion of IEM

第一階段: 2015年10月1日至2016年3月31日(共21項病症)

Disorders of Organic Acids (7 conditions)	有機酸障礙 (七項)
Multiple carboxylase deficiency	多發性羧化酶缺乏症
Glutaric acidaemia type I	戊二酸血症 I 型
Methylmalonic acidaemia	甲基丙二酸血症
Propionic acidaemia	丙酸血症
Isovaleric acidaemia	異戊酸血症
3-hydroxy-3-methylglutaryl-CoA lyase deficiency	白胺酸代謝異常症
Beta-ketothiolase deficiency	貝塔酮硫解酶缺乏症

Disorders of Amino Acids (8 conditions)	氨基酸障礙 (八項)
Phenylketonuria	苯丙酮尿症
6-pyruvoyl-tetrahydropterin synthase deficiency	六- 丙酮酰 - 四氫蝶呤合成酶缺乏症
Argininosuccinic acidaemia	精氨酸血症
Maple syrup urine disease	槭糖尿病
Citrullinaemia type I	瓜氨酸血症 I 型
Citrullinaemia type II	瓜氨酸血症 II 型
Tyrosinaemia Type I	酪氨酸血症 I 型
Homocystinuria	高胱氨酸尿症

Disorders of Fatty Acid Oxidation (6 conditions)	脂肪酸氧化障礙 (六項)
Carnitine uptake deficiency	卡尼丁吸收障礙
Carnitine-acylcarnitine translocase deficiency	卡尼丁穿透障礙
Carnitine palmitoyltransferase II deficiency	卡尼丁結合酵素 II 缺乏症
Medium-chain acyl-CoA dehydrogenase deficiency	中鏈醯輔酶A去氫酶缺乏症
Very long-chain acyl-CoA dehydrogenase deficiency	極長鏈醯輔酶A去氫酶缺乏症
Glutaric acidaemia type II	戊二酸血症的 II 型

第二階段: 2016年4月1日至2017年3月31日加入以下三項病症(合共24項)

Congenital adrenal hyperplasia	先天性腎上腺增生症
Biotinidase deficiency	生物素缺乏
Classic galactosaemia	半乳糖血症

Target Population (Screening Policy)

- Babies born in QEH or QMH
  - free of charge
  - voluntary
  - consent signed by a parent

Phase I: 1 October 2015 - 31 March 2016

All babies born at the two aforesaid hospitals are eligible **unless**:

1. They are born before 34 weeks of gestation,
2. Their birth weight is less than 2000 grams,
3. They are admitted to Neonatal Intensive Care Unit (NICU)

Phase II: 1 April 2016 - 31 March 2017

All babies born at the two aforesaid hospital are eligible.

Notes:
Starting from 1 April 2016, three separate blood specimens will be collected under following conditions:

1. *premature (less than 34 weeks of gestation),*
2. *low birth weight (less than 2000g), and babies*
3. *admitted into NICU.*

The first specimen is to be collected on admission to NICU, the second specimen during 48 – 72 hours of life, and the third specimen upon discharge or on day 28 of life, whichever earlier.

Flow

1 Antenatal visit

Antenatal health education related to IEM screening is provided by healthcare professionals.

2 Consent

With parent's written consent, majority of babies born after 24-72 hours and preferably milk feeding for 24 hours will receive heel pricking procedure for blood specimen collection.

3

Collection of blood specimen on a filter paper card.

4 Laboratory

Laboratory analysis

5 Screening results

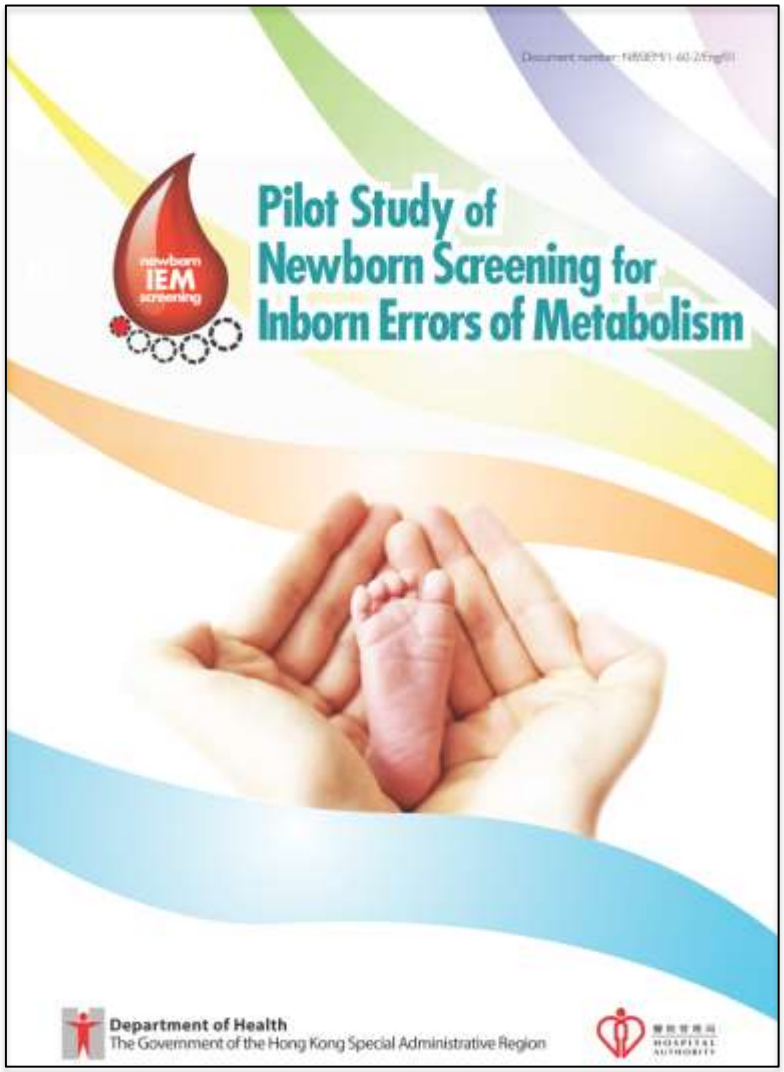
Screening results within the scope of this pilot study available.

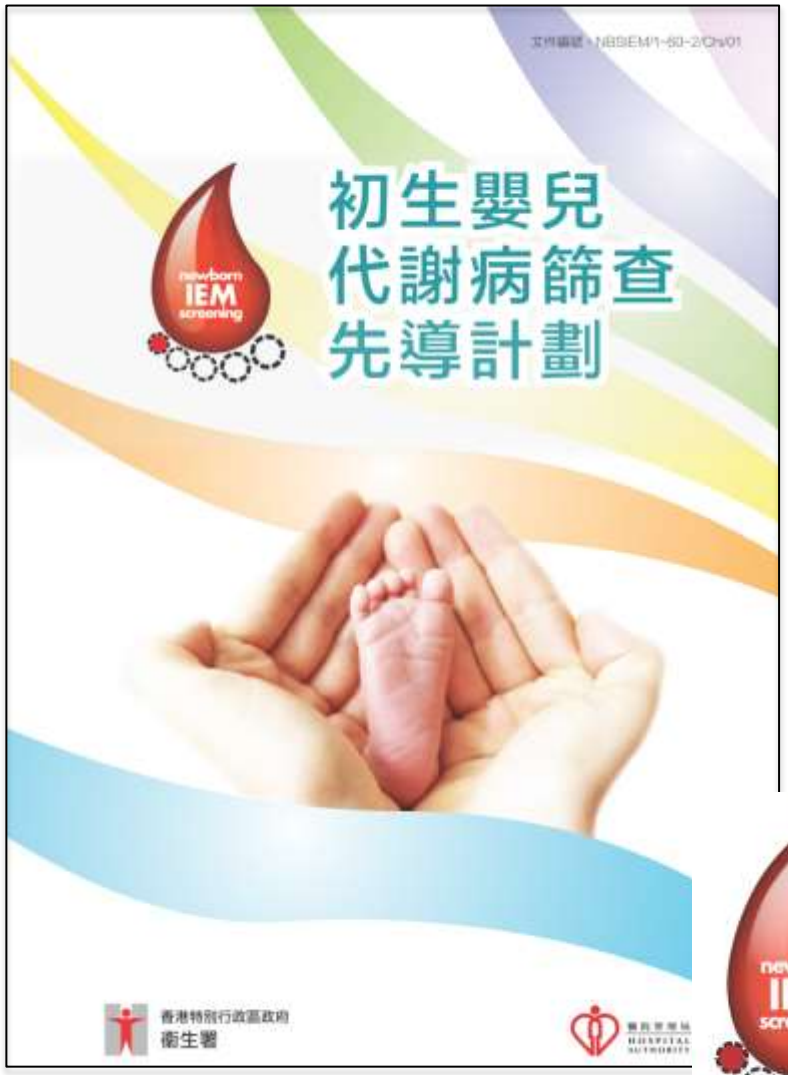
6 Report

Normal results :
Parents will not receive notification.

Education to enrich

Parental Knowledge





Education to enrich
Parental Knowledge

Informed consent

衞生署
香港特別行政區政府

醫院管理局
HOSPITAL AUTHORITY

初生嬰兒代謝病篩查先導計劃

同意書

1. 我已閱讀並理解小冊子內所提供的資料（文件編號：NBSIEM/1-60-2/Chi/v1.0）。
2. 我明白參與這個先導計劃是自願的。
3. 我明白這個先導計劃所涵蓋的先天性代謝病僅限於小冊子內附錄一所列（文件編號：NBSIEM/1-60-2/Chi/v1.0）。
4. 我明白我的嬰兒的個人資料會提供予衞生署，以為先導計劃進行數據分析。
5. 我會讓我的嬰兒參加這個先導計劃。

父/母親簽名 | **聯絡電話號碼**

父/母親姓名 | **日期**

貼上嬰兒標貼

請寫下您的**聯絡電話號碼**，以方便通知不正常或不確定之結果。
謝謝您的合作!

Department of Health
Hong Kong SAR
Government

HOSPITAL AUTHORITY

Pilot Study of Newborn Screening for Inborn Errors of Metabolism (IEM)
Consent Form

1. I have read and understood the information provided in the pamphlet (Ref no: NBSIEM/1-60-2/Eng/v1.0).
2. I understand that the participation of this Pilot Study is voluntary.
3. I understand that this Pilot Study is specific only to the IEM as specified in Appendix 1 to the pamphlet (Ref no: NBSIEM/1-60-2/Eng/v1.0).
4. I understand that my baby's personal data would be made available to the Department of Health for data analysis of this Pilot Study.
5. I will let my baby participate in this Pilot Study.

Signature of mother/ father | **Contact phone No.**

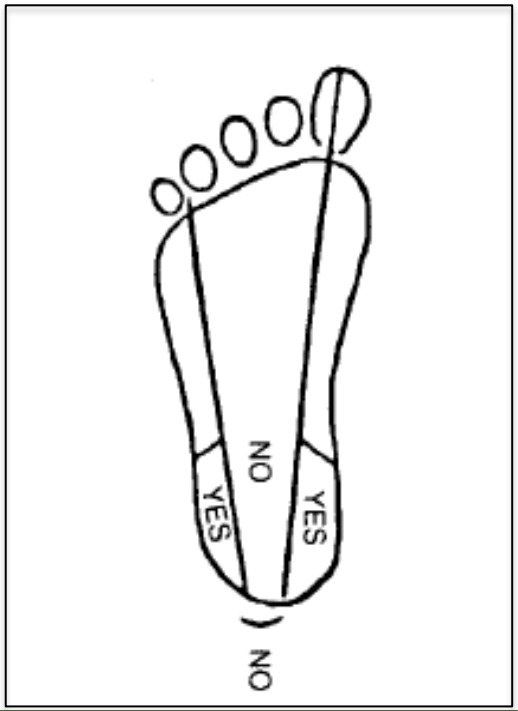
Name of mother/ father | **Date**

Fix a newborn gum label

Please write down your **contact ph**
numbers so that abnormal result c
notified once available.
Thanks foryour co-operation!

newborn IEM screening

Workflow - Specimen Collection



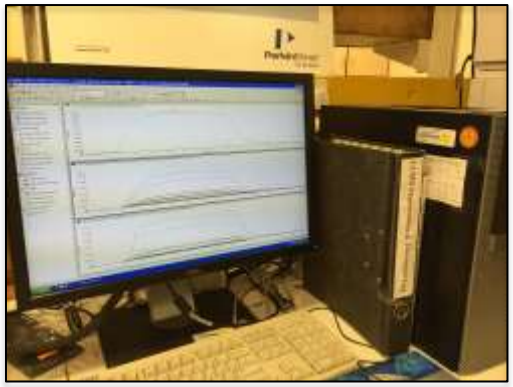
newborn IEM screening

Workflow - Dispatch of Specimens

Drying (3 hours)

Dispatch of specimen

Workflow - Laboratory



Workflow - Reporting

Screening Results		Follow-up Action
Normal	Risk of suffering from the screened metabolic diseases is very low.	Parents will not receive any notification.
Abnormal	Risk of suffering from the screened metabolic disease is high.	Hospital staff will notify parents by telephone within **7 working days**. Babies will be referred to paediatricians for further diagnostic testing and management.
Uncertain	About 1% of the screened specimens will have uncertain results.	

Education of enhance ***Healthcare Professionals Knowledge***

- Four lectures held at QEH and QMH
  - in June 2015
  - before launching 1st phase
- Six lectures held at QEH and QMH
  - in March & April 2016
  - before launching 2nd phase
  - esp. for Paediatrics frontline staff

Newborn Screening
For Inborn Errors of Metabolism
(Pilot Study)

Aim:
- To introduce the Pilot Study
- To enhance the knowledge and understanding of newborn screening programme and inborn error of metabolism

Target audience:
- Healthcare professional

Speakers:
- Dr Hau Wai Lok, SMO, Clinical Genetic Service, DH
- Dr Chloe Mak, Consultant Chemical Pathologist, PMH
- Dr Betty But, QEH/ Grace Poon, QMH (Paediatrician)

Date	Time	Venue
3 June 2015 (Wednesday)	10am - 12noon	Lecture Theatre, M Block, QEH
3 June 2015 (Wednesday)	2:15pm – 4:15pm	K2 Common Room, QMH
5 June 2015 (Friday)	2:30pm - 4:30pm	Lecture Theatre, M Block, QEH
10 June 2015 (Wednesday)	2:15pm – 4:15pm	Class room 1, Block A, NQ, QMH

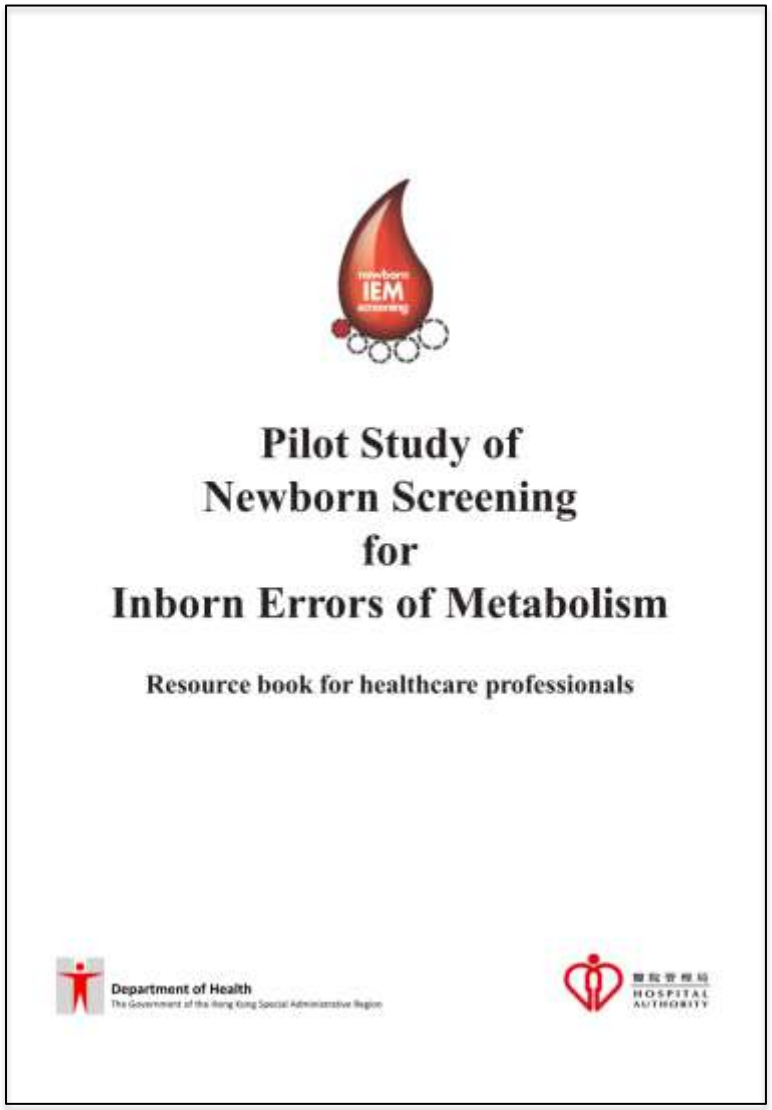
All are welcome

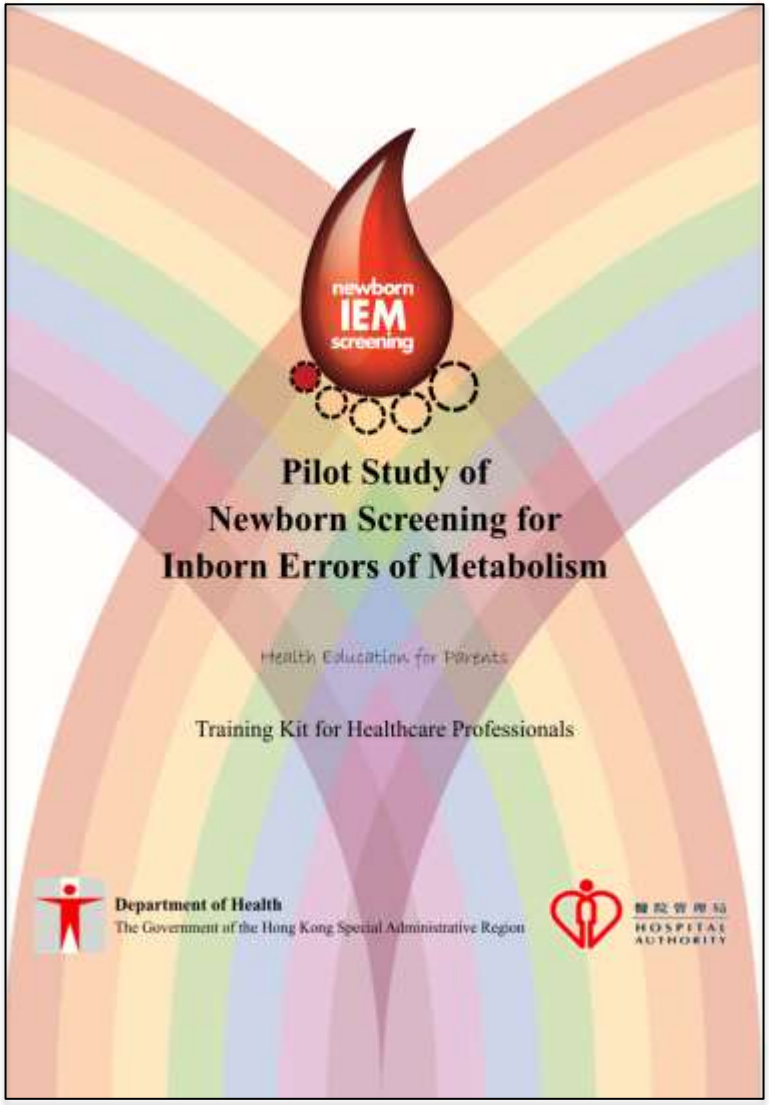
Department of Health
Hong Kong SAR Government

HOSPITAL AUTHORITY

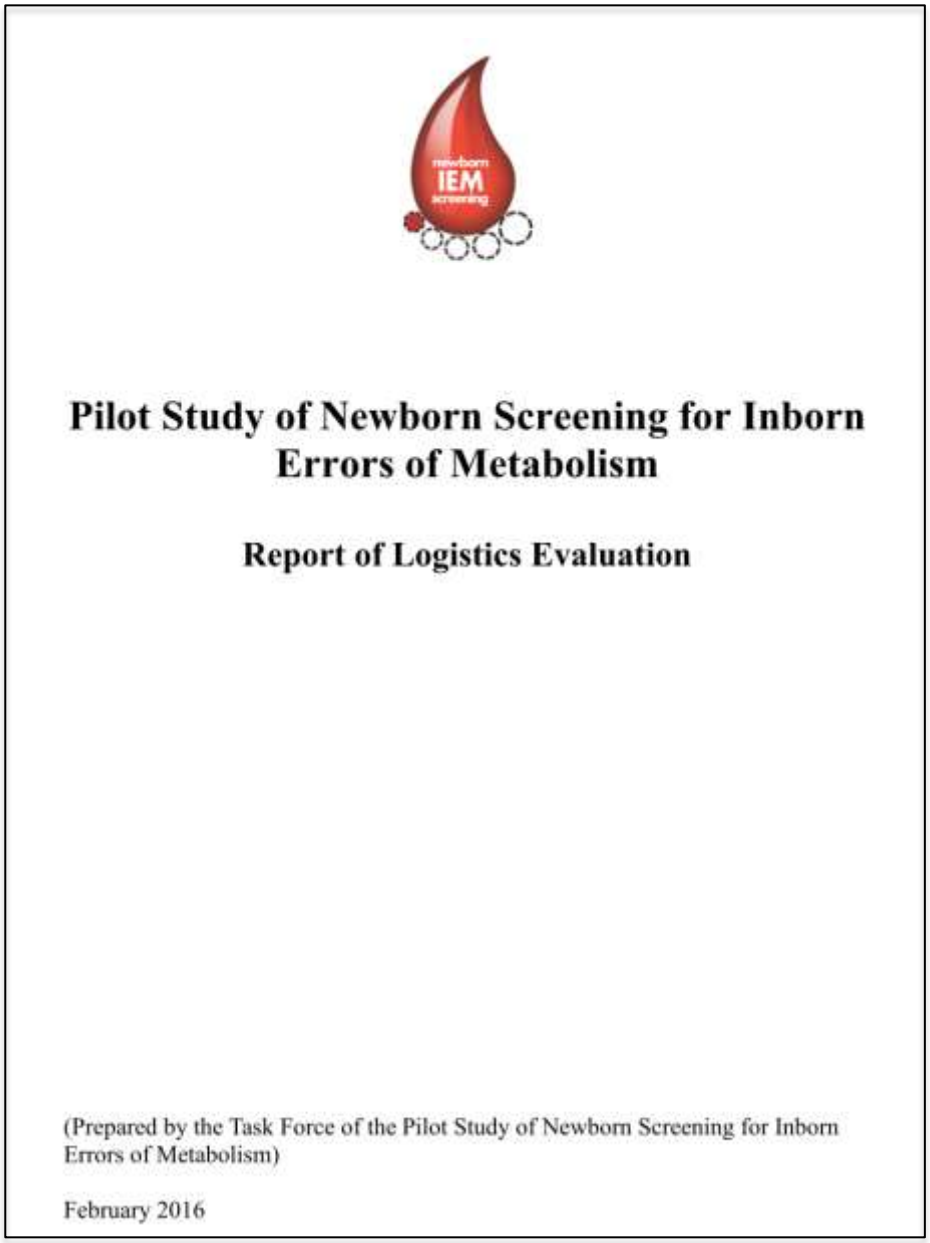
newborn IEM screening

Education of enhance *Healthcare Professionals Knowledge*





Evaluation - Logistics



Pilot Study of Newborn Screening for Inborn Errors of Metabolism

Report of Logistics Evaluation

(Prepared by the Task Force of the Pilot Study of Newborn Screening for Inborn Errors of Metabolism)

February 2016

- Parents being informed
- Encouraging parental consent rate
- Smooth operation
- Effective and efficient communication among involved parties

Challenges

- Enhancement of public education
- Reinforcement of healthcare professional capacity
- Rapid pace of scientific advancement in the detection, diagnosis, and treatment
  - review of included IEM conditions
  - optimisation of mass spectrometry MS/MS-based screening
  - lifelong treatment with specialty and interdisciplinary care
- Resources implication
- Incidental findings

Challenges

- Retention of residual dry blood spots (DBS)

- quality assurance
- test validation
- development of new screening methods
- additional/repeat testing (esp for sudden death)
- forensic purposes
- future research

JOURNAL OF LAW, MEDICINE & ETHICS

Lessons from the Residual Newborn Screening Dried Blood Sample Litigation

Michelle Huckaby Lewis

2014 PUBLIC HEALTH LAW CONFERENCE: INTERSECTION OF LAW, POLICY AND PREVENTION • SPRING 2015

Background

Most babies born each year in the U.S. undergo mandatory newborn screening to detect serious medical conditions that can cause devastating effects if treatment is not initiated prior to the onset of symptoms.[1] Not all of the blood collected from newborns is used during routine newborn screening, and many states retain the residual dried blood samples (DBS).[2] DBS have a broad range of potential uses, from program evaluation to public health and biomedical research unrelated to newborn screening.[3] State laws vary regarding whether parental consent is required to use DBS for secondary research,[4] but federal now requires parental consent for the use of DBS in federally funded research.[5]

The use of DBS for secondary research without explicit parental permission has generated controversy, culminating in lawsuits against health departments in Texas, Minnesota, and Indiana. The issues raised by the lawsuits extend beyond the legal question of whether states had statutory authority to retain DBS for secondary use. Additional aspects of state practices related to the retention and use of DBS have been of concern to some parents.

Acknowledgement

- Healthcare Professional involving in the Pilot Study
- Fellow Task Force Members

The End

Thank you