Joshua Hellmann Foundation
Newborn Metabolic Screening Program
Clinical Protocol

The CUHK – BCM Joint Centre for Medical Genetics

The Chinese University of Hong Kong

Version 9

Last update: January 2020

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Introduction

Joshua Hellmann Foundation - Newborn Metabolic Screening Program is a newborn screening program targeting at a panel of inborn errors of metabolism (IEM) disorders and congenital adrenal hyperplasia (CAH) (added to the program on 15th February 2016). The goal of the screening program is to identify affected infants at the earliest instance, often before they develop any signs or symptoms of the diseases and treat them early so as to ensure the best possible treatment outcome.

Screening for cystic fibrosis (CF) is added to the program on 1st February 2014. It is recommended if both parents are Caucasians. Samples are sent to an overseas laboratory for analysis. CF screening result is reported separately, and the turnaround time is longer than IEM + CAH screening. For more details, please see the section "Screening for cystic fibrosis".

This screening program is complementary to the conventional cord blood screen for congenital hypothyroidism and glucose-6-phosphate dehydrogenase (G6PD) deficiency provided by the Department of Health in Hong Kong.

Referring doctors can choose either options:

- Option 1: IEM + CAH screening
- Option 2: IEM + CAH + Cystic Fibrosis

Workflow of the screening program is shown in Appendix 1.

Newborn screening for Inborn Errors of Metabolism (IEM)

Background

Inborn Errors of Metabolism (IEM) is a large group of genetic disorders with a collective incidence of 1 in 4000. Infants affected by IEM can appear normal at birth. If not identified and treated early, these IEMs may result in permanent neurological damages and even mortality.

Screening method

We use tandem mass spectrometry (MS/MS) to measure a number of amino acids, free carnitine and acylcarnitines in the dried blood spot cards. Quantities of these analytes and their relationship with each other are used to screen for 30 IEM of amino acid, organic acid and fatty acid oxidation metabolisms. A list each of the IEM target by the screening test is set out in Appendix 2.

Screening result interpretation and reporting

The IEM newborn screening test by MS/MS is a screening but not a diagnostic test. Clinicians must not diagnose or prescribe treatment based solely on the screening test results.

As with any laboratory test, both false positive and false negative results may occur. This means unaffected infants may be falsely identified by the screening test. Thus it is extremely important that all abnormal screening results be followed by proper diagnostic tests.

After analysis by MS/MS, three types of results are possible:

1. **Normal** – this means all metabolites measured are within the pre-defined cut-offs. Normal reports will be sent to the referring doctor.

2. **Positive** – this means some of the metabolites measured are significantly deviated from the pre-defined cut-offs and the pattern of abnormalities suggest an underlying IEM. Clinical assessment and follow-up diagnostic tests (Appendix 3) are necessary. The referring doctor will be informed about the abnormal results by phone as soon as possible. Reports will be faxed and mailed to the referring doctor. Metabolic Paediatrician at the Centre of Inborn Errors of Metabolism will also be informed. ***All positive screening results must be dealt with immediately and without delay.***

3. **Uncertain** – this means some of the metabolites measured fall slightly outside the pre-defined cut-offs. Repeat analysis using a second DBS sample is needed. For some babies, additional blood and/or urine tests are also needed. The referring doctor will be

informed about the abnormal results by phone as soon as possible. Reports will be faxed and mailed to the referring doctor. It is the responsibility of the referring doctor to contact the parents for a repeat DBS sample as soon as possible. It is estimated that around 1 in 600 infants screened may require a repeat DBS sample. Majority of repeat DBS will have a normal analysis result. Babies with persistent abnormal results will require clinical assessment and immediate follow-up diagnostic testing.

Newborn screening for congenital adrenal hyperplasia (CAH)

Background

Congenital adrenal hyperplasia (CAH) is a group of genetic disorders, in which the body cannot produce enough cortisol. Classic CAH describes patients with no or minimal residual enzyme activity who present at birth or soon after birth. There are two different forms of classic CAH – salt-wasting form and simple virilization form. The incidence of classic CAH is around 1 in 10,000 to 1 in 20,000. The most common (>90%) cause of CAH is 21-hydroxylase deficiency. In this form of CAH, 75% of affected patients cannot produce adequate cortisol and aldosterone. Low levels of these hormones may cause nausea and vomiting, tiredness, dehydration, and weight loss. In most severe case, CAH can lead to "salt-losing crisis" with low blood pressure, shock or even death. Low level of cortisol also stimulates the production of ACTH from the pituitary gland. This can lead of hyperpigmentation of the skin and overproduction of adrenal androgens. In female babies, exposure to excessive androgens in utero may result in abnormal genital development. In the remaining 25% of affected patients, production of aldosterone is adequate and affected babies present with virilization at birth or precocious pubertal development in childhood.

Affected baby girls are much more readily detected at birth than affected baby boys because of abnormal genitalia. However, both genders have the same risk of developing salt-losing crisis. The aim of newborn screening for CAH is to detect babies affected by salt-losing form of CAH early so that prompt treatment can prevent the development of salt-losing crisis. The milder form of CAH is not the target of this screening testing.

Screening method

The CAH screening test measures the level of 17-hydroxyprogesterone (17-OHP) in the dried blood spot cards by time-resolved fluoroimmunoassay. All newborns have high levels of

17-OHP at birth. In healthy babies, the levels of 17-OHP decrease with time but for babies affected by CAH 17-OHP concentrations remain high. Premature and stressed babies (otherwise not affected CAH) have higher 17-OHP levels than full-term healthy babies.

Screening result interpretation and reporting

The CAH newborn screening test is a screening but not a diagnostic test. Clinicians must not diagnose or prescribe treatment based solely on the screening test results.

As with any laboratory test, both false positive and false negative results may occur. This means unaffected infants may be falsely identified by the screening test. Thus, it is extremely important that all abnormal screening results be followed by proper diagnostic tests.

Babies with 17-OHP results fall outside the pre-defined cut-off are at risk of CAH and require further investigations. For most babies, a repeat analysis using a second DBS sample is all that is needed. More investigations (e.g. electrolytes) may be required and this depends on the actual 17-OHP results and the clinical condition of the babies. The referring doctor will be informed about the abnormal results by phone as soon as possible. Reports will be faxed and mailed to the referring doctor. It is the responsibility of the referring doctor to contact the parents for a repeat DBS sample as soon as possible.

The false positive rate of CAH screening is around 0.5% in general but is higher in premature and stressed infants. Majority of repeat DBS will have a normal 17-OHP result. Babies with persistent elevated 17-OHP levels will require clinical assessment and immediate follow-up diagnostic testing.

Target Babies

Dried blood spots collection for well babies

All healthy newborns born at or after 34 weeks of gestation[1] who have completed oral feeding for 24 hours are suitable for the screening test. Dried blood spots (DBS) should be

[1]During Phase I (15 July 2013 – 31 August 2013), we accepted DBS from healthy term newborns (>/=37 weeks of gestation). Phase II was started from 1 September 2013, we had extended our service to include slightly preterm baby with body weight >2kg.

collected after completion of oral feeding for 24 hours and up to the 7th day after birth. In general, the best time for screening for the majority of IEM and CAH is between 24 and 72 hours of life. This means that the sample collection is best done in hospital before the babies are discharged.

Dried blood spot collection for premature (<34 weeks of gestation), low birth weight, and sick newborns

For newborns who fit into one of the following criteria, repeated dried blood spots collection at different time points are needed for a better analysis as a newborn screening test for inborn errors of metabolism.

- Premature (< 34 weeks of gestation)
- Low birth weight of < 2,000 g
- Sick newborns who require admission to Neonatal Intensive Care Units
- Newborn who was kept nil by oral, or has received parenteral nutrition, blood transfusion

Schedule of blood sample collection

- First blood sample: blood should be drawn at 24 to 72 hours of life at the unit, and preferably before transfusion of red cell or whole blood.
- For babies requiring immediate whole blood or red cell transfusion soon after admission to NICU, an additional blood sample should be drawn before transfusion of such products is instituted. (Note: Transfusion of platelet and/or plasma does not warrant an additional blood sampling.)
- Second blood sample: blood should be drawn at day 28 of life or before discharge whichever comes first.

Recommendation:

We recommend proceeding directly to diagnostic testing for the following cases:

- If there is clinical suspicion for IEM or metabolic derangements such as recurrent hypoglycaemia, hyperammonaemia, or ketonuria.
- If there is clinical suspicion for CAH.
- Infants with a family history of IEM (e.g. affected older siblings) or CAH.

Referral sites

1. Hospital-based Obstetrics or Paediatrics ward or clinic
   To ensure correct timing of blood sampling and adequate follow-up of results, we accept samples from Obstetric or Paediatric departments in local Hong Kong hospitals.
2. Private clinic
   We also accept samples from private obstetrics or paediatrics clinic with their own logistics arrangement. Please contact our laboratory at 6806 4590 for further details.

Collection card

A special filter paper (referred to as "collection card") is used to collect a few drops of blood obtained by heel pricking. This kind of sample is commonly referred to as Dried Blood Spot (DBS).

Collection cards can be obtained from the **Prenatal Genetics Diagnosis Laboratories at Prince of Wales Hospital.** (Address: 4/F, Block K, DTB, Prince of Wales Hospital, 30-32 Ngan Shing Street, Shatin, N.T., Hong Kong)

Unused collection cards should be stored properly. Do not place heavy objects on top of unused collection cards as this will cause compression on the cards.

Request form

(Appendices 4 and 5)

Please make sure that all necessary information on the request form is completed. ***It is vital to provide name and contact of referring doctor, who will be contacted directly by phone should there be any positive or uncertain results.***

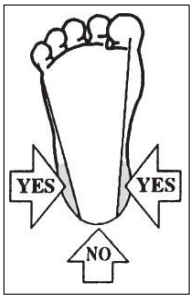
It is the responsibility of the referring doctors to explain the screening test in details to parents before blood collection. Mothers who agree to the screening should give their consent in writing by signing at the end of the request form.

Request form can be obtained from the **Prenatal Genetics Diagnosis Laboratories,** the address is set out at the end of this document.

Dried blood spot (DBS) collection and handling guide

1. Collect sample after completion of oral feeding for 24 hrs, and up to the 7th day after birth. The best time for collection is between 48 and 72 hours. DO NOT collect samples before 24 hours of age. (For preterm, low birth weight, sick babies, please refer to specific blood taking schedule on page 7)
2. The shaded areas (See picture) indicate the puncture sites on the heel where blood is collected from.

3. To prevent specimen contamination, DO NOT touch any part of the circled areas on the filter paper before, during or after blood collection.
4. Complete baby's demographic data (name, date of birth and ID or hospital reference number) or affix baby's GUM label on the collection card before proceeding to collection.
5. Cleanse site with alcohol swap and allow to dry.
6. Puncture heel with sterile lancet designed for heel prick for infants. Blade-type lancet with incision depth of 2 mm is recommended.
7. Wipe away the first drop of blood with sterile gauze and allow the next large blood drop to form.
8. Place collection card over this large blood drop and allow it to soak through and completely fill the circle area in one single application. DO NOT apply blood to both sides of the collection card. DO NOT layer several blood drops on top of each other.



9. Fill remaining circles on the collection card with successive blood drops. A minimum of four circles is necessary for each collection card. *(A minimum of 5 completely filled circles are required if Cystic Fibrosis Screening is requested.)*
10. Allow blood spots to dry thoroughly in a horizontal position on a non-absorbent surface for at least 3 to 4 hours at room temperature. DO NOT leave wet collection cards in a hanging position as this will cause the heavier red cells to migrate to the dependent end of the circle resulting in uneven saturation. (See picture)

11. Keep collection card away from direct sunlight and heat. DO NOT dry blood spots on a heater, in a microwave, with a hair dryer or under sunlight. DO NOT stack collection cards on top of each other before thorough drying.

If the specimen is improperly collected and handled, the accuracy of the screening test results will be compromised.

Delivery of the collection card to the laboratory

1. Transport each collection card in a separate envelope. DO NOT use plastic bag.
2. Deliver collection cards and completed request forms to **Prenatal Genetics Diagnosis Laboratories**, the address is set out at the end of this document.
3. DBS sample reception time:
   Mondays to Fridays (except for public holidays) 9:00am – 3:00pm (closed between 1:00 – 2:00 pm)
4. DBS received before 10:00am on Friday (or the day before public holiday) will be analyzed on the same day. Reports with normal results will be sent out on the following Monday (or the next working day). If results are positive or uncertain, referring doctors will be contacted as soon as possible.
5. For DBS collected after 10:00am on Friday, Saturday, Sunday and any public holiday, please send them to the laboratory on the following Monday (or the next working day). For DBS samples that cannot reach the laboratory on the day of collection, store them in a cool dry place at room temperature for no more than four days before sending them to the laboratory.
6. Special arrangement for long public holidays (> 3 days) will be announced as necessary.

Unsatisfactory dried blood spot samples

The following DBS are unsatisfactory for the screening test:

1. Incomplete information on request form or collection card, making it impossible to determine the baby's identity or age at the time of collection.
2. Insufficient quantity of blood on the card to perform the analysis.
3. Damaged or contaminated collection card.

When unsatisfactory DBS samples are received, the laboratory will not proceed to analysis. The referring hospital or clinic will be informed as soon as possible for repeat collection.

Out-patient dried blood spot (DBS) collection

If DBS cannot be collected before a newborn is discharged from hospital, the parents may bring their baby to the Newborn Screening Clinic of the CUHK – BCM Joint Centre for Medical Genetics, The Chinese University of Hong Kong at Prince of Wales Hospital for blood collection.

Turnaround time

A report will be issued at around three working days after the collection card is delivered to the laboratory.

Testing laboratory
Xcelom Ltd, Hong Kong

Storage and disposal of dried blood spots

All dried blood spot cards will be stored in the laboratory for a minimum of two years. The laboratory will ensure appropriate and proper protection of sensitive personal information. With informed consent from parents, the laboratory may store residual DBS cards for more than two years and use them for internal study or research studies after all identifying information has been removed. Amino acids and acylcarnitines in DBS cards will deteriorate after prolonged storage which will render retrospective diagnosis of IEM impossible.

Screening for cystic fibrosis

Background
Cystic fibrosis (CF) is the most common autosomal recessive disease in white populations. CF is caused by mutations in the *CFTR* gene, which encodes a chloride channel called cystic fibrosis transmembrane conductance regulator (CFTR) protein. Affected patients develop various gastrointestinal, pulmonary and endocrine problems from neonatal period to adulthood. (3) Newborn screening and early treatment can improve the nutritional, growth and intellectual outcomes in CF patients.(4)

The incidence of CF varies in different ethnic groups (5-8):

- Non-Hispanic Caucasians 1:2,500

- Ashkenazi Jews 1:2,270
- Hispanics 1:13,500
- African Americans 1:15,100
- Asians 1:35,100 – 350,000

A commonly adopted CF screening strategy is the IRT/DNA approach. Immunoreactive trypsinogen (IRT) is first measured in dried blood spot samples (Tier 1 test). IRT is a marker of pancreatic injury and is not specific to CF. If IRT concentration is elevated, a panel of *CFTR* mutations are then tested on the same dried blood spot (Tier 2 test). Babies with elevated IRT concentrations and one or two *CFTR* mutations are reported to have positive screening results. Further laboratory testing and clinical assessment are necessary to confirm the diagnosis of CF.

The sensitivity of newborn screening for CF is around 95% in developed countries such as Australia, the United Kingdom and the United States. (9-11) Approximately 15% of infants with CF are born with meconium ileus. These patients may have normal IRT concentrations and thus be missed by the newborn screening. Therefore, neonates with meconium ileus or a history of CF in siblings should always be followed up regardless of the screening result. The same principle applies to patients who develop signs and symptoms suggestive of CF. Nonetheless, there is evidence demonstrating that false negative newborn screening results do not result in delay in diagnosis or poorer outcomes in affected patients. (11,12)

Sample requirement

Two dried blood spot of 12 mm in diameter (i.e. one completely filled circle).

Newborn Metabolic plus Cystic Fibrosis Screening: a minimum of 5 completely filled circles are required.

Testing laboratory

Newborn Screening Laboratory, Wisconsin State Laboratory of Hygiene, Madison, Wisconsin, USA

Testing algorithms

Tier 1 test: immunoreactive trypsinogen (IRT)
Tier 2 test: mutation analysis of 23 *CFTR* mutations (http://www.slh.wisc.edu/clinical/newborn/health-care-professionals-guide/nbs-test-panel-of-diseases/#cf).
Tier 2 test is performed on the highest 4% of the daily IRT results.

Interpretation

Babies with elevated IRT concentrations and one or two CFTR mutations are reported to have positive screening results. Further laboratory testing and clinical assessment are necessary to confirm the diagnosis of CF.

Provision of accurate ethnicities of parents to the laboratory aids interpretation of Tier 2 test (CFTR gene analysis) results.

- Potential false negative IRT results
  - Affected infants with meconium ileus.
  - Affected infants with pancreatic sufficiency.
  - IRT levels in affected infants remain elevated for 2 to 4 weeks and may decline in some patients at 1 month. (13) Thus this newborn screening test is not suitable for older infants or children suspected to have CF.
- Potential false positive IRT results
  - IRT may be falsely elevated in premature or sick infants.
- Mutation detection rate of the 23-*CFTR*-mutation panel (*CFTR* gene analysis)
  - Ashkenazi Jewish 94%
  - Non-Hispanic white 88%
  - Hispanic white 72%
  - African American 64%
  - Asian American 49%

Turnaround time

Samples with normal IRT results: around three to four weeks

Enquiries

Office hours: Monday to Friday 9:00am – 5:00pm
The CUHK – BCM Joint Centre for Medical Genetics , The Chinese University of Hong Kong
Enquiry hotline for general public (during office hours): 6806 4590
General enquiry (e.g. request form, collection card and DBS reception)
Prenatal Genetics Diagnosis Laboratories 4/F, Block K, DTB, Prince of Wales Hospital 30-32 Ngan Shing Street, Shatin, N.T., Hong Kong Tel: 6806 4590 Fax: 3505 4810
Obstetrician
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Newborn screening laboratory
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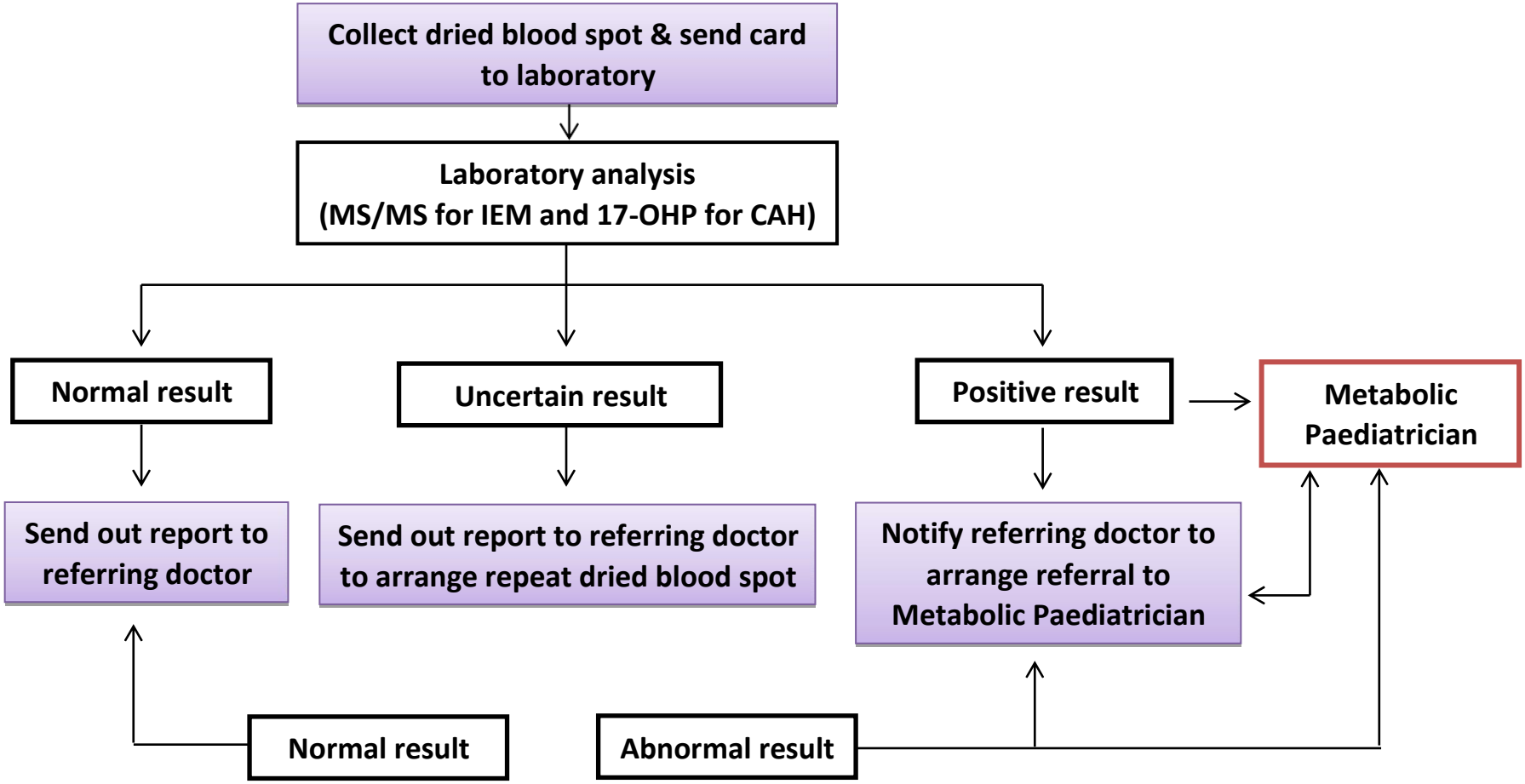
Clinical laboratory and standard institution. October 2009

Appendix 1. Workflow of the Newborn Metabolic Screening Program



Appendix 2. Target Inborn Errors of Metabolism

Inborn Errors of Metabolism	**ACMG# classification**	**Key metabolites**
Amino Acid Disorders		
Phenylketonuria (PKU)	Core	↑ Phe
Maple syrup urine disease (MSUD)	Core	↑ Leu/Ile
Citrullinaemia type 1	Core	↑ Cit
Argininosuccinic aciduria	Core	↑ Cit
Homocystinuria	Core	↑ Met
Tyrosinaemia type 1	Core	↑ Tyr, SA*
Arginase deficiency	2°	↑ Arg
Defects of biopterin cofactor biosynthesis or regeneration	2°	↑ Phe
Citrullinaemia type 2 (Citrin deficiency)	2°	↑ Cit
Organic Acid Disorders		
Propionic acidaemia (PA)	Core	↑ C3
Methylmalonic aciduria (MUT, cbl A/B)	Core	↑ C3
Isovaleric acidaemia (IVA)	Core	↑ C5
ß-ketothiolase deficiency (BKT)	Core	↑ C5OH, C5:1
Glutaric acidaemia type 1 (GA1)	Core	↑ C5DC
3-Hydroxy-3-methylglutaryl-CoA lyase deficiency (HMG)	Core	↑ C5OH
Multiple carboxylase deficiency (MCD)	Core	↑ C5OH
3-Methylcrotonyl-CoA carboxylase deficiency (3MCC)¶	Core	↑ C5OH
Malonic aciduria (Malonyl-CoA decarboxylase deficiency)	2°	↑ C3DC
3-Methylglutaconic aciduria type I (3MGA)	2°	↑ C5OH
Cbl C/D	2°	↑ C3
Fatty Acid Oxidation Disorders		
Primary carnitine deficiency (Carnitine update defect, CUD)¶	Core	↓ C0
Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)	Core	↑ C8
Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)	Core	↑ C14:1
Long-chain hydroxyl-acyl-CoA dehydrogenase (LCHAD)	Core	↑ C16OH
Trifunctional protein deficiency (TFP)	Core	↑C16OH

Carnitine palmitoyltransferase I deficiency (CPT1)	2°	↑ C0
Carnitine palmitoyltransferase II deficiency (CPT2)	2°	↑ C16
Carnitine-acylcarnitine translocase deficiency (CACT)	2°	↑ C16
Multiple acyl-CoA dehydrogenase deficiency (Glutaric aciduria type 2, GA 2)	2°	↑ C4, C5
Medium/short-chain hydroxyl-acyl-CoA dehydrogenase deficiency (M/SCHAD) (3-Hydroxyacyl-CoA dehydrogenase deficiency)	2°	↑ C4OH

#ACMG: American College of Medical Genetics (Ref 13, 14). Core conditions: newborn screening for these disorders are mandated in the United States. 2° conditions: they are part of the differential diagnosis of a core condition, they are clinically significant and revealed with the screening technology but lack an efficacious treatment, or they represent incidental findings for which there is potential clinical significance.

*SA: Succinylacetone. A second tier test for dried blood spots with tyrosine concentration above the pre-defined cut-off.

¶ A positive screening test result may suggest inborn errors of metabolism in the mother or baby.

Appendix 3. First line diagnostic tests for Inborn Errors of Metabolism

Amino acid profile	
Details:	Diagnostic test for amino acid disorders Abnormal in some organic acid disorders
Sample requirement:	Heparin blood 3 mL (minimum 1 mL) or Plasma 1 mL (minimum 0.2 mL)
Carnitine and acylcarnitine profile	
Details:	Diagnostic test for fatty acid oxidation disorders Abnormal in some organic acid disorders
Sample requirement:	Clotted blood in plain bottle 1 mL or Serum 0.5 mL
Urine metabolic screen	
Details:	Diagnostic test for organic acid disorders Abnormal in some amino acid and fatty acid oxidation disorders
Sample requirement:	Fresh spot urine in plain bottle 20 mL

Appendix 4. Request form (consent in Chinese)

THE CHINESE UNIVERSITY OF HONG KONG

The CUHK – BCM Joint Centre for Medical Genetics

Joshua Hellmann Foundation Newborn Metabolic Screening Program

Tel: 6806-4590 Fax: 3505-4810

Affix Baby's GUM label here or fill in the following Name: DOB: ID / Hospital no.:	Affix Mother's GUM label here or fill in the following Name: ID / Hospital no.:

Gestation age at delivery :_____week_____day Hospital / Place of birth: _____

Birth weight :_____grams Date and time of birth :____/____/_____:____am/pm

Ethnicity: ☐ Chinese ☐ Caucasian ☐ Indian ☐ Pakistani ☐Other (please specify:_____)

Feeding type: ☐ Breast ☐ Formula ☐ Mixed *(please wait until feeding for > 24 hrs before blood collection)*

Baby / Mother: ☐ Antibiotics (_____) ☐ Steroids

Family history: ☐ IEM ☐ Congenital adrenal hyperplasia ☐ Cystic fibrosis

Screening Test:	**☐ IEM & CAH** Minimum 4 blood spot circles	**☐ IEM, CAH and Cystic Fibrosis** Minimum 5 blood spot circles

Referring doctor information (contact person for positive results)

Name: Dr_____ Signature: _____

Phone: _____ Fax:_____

Hospital or Clinic: _____

Dried Blood Spot (DBS) Information

See back of collection card for DBS collection and handling guide.
Provide baby's name, ID / hospital reference no, and date of birth on the collection card.

DBS collection date:_____ DBS collection time:_____

Test information: ☐ 1st sample ☐ 2nd sample ☐ Pre-transfusion sample ☐ Others:_____)

家長同意書:

- 我明白這個新生兒代謝病篩查計劃的目的和可能出現的結果。
- 我明白如果寶寶的第一個血樣顯示不確定或陽性結果，我們將會被安排重新抽取血樣本再作篩查。
- 我明白在極少數情況下，患病的寶寶有可能未被檢出。
- 我明白寶寶的篩查報告會出現在醫院管理局的電腦系統內。
- 我明白，剩餘的血樣經刪除所有的身份信息後，可能會被保留並用於實驗室內部試驗及醫學研究。
  拒絕允許使用寶寶的剩餘血樣將不會影響寶寶的篩查結果。
  如果您不希望寶寶的剩餘血樣被保留作以上用途，請在這裡劃上✓號：☐

母 / 父親姓名:_____ 電話號碼:_____

母 / 父親簽署:_____ 簽署日期:_____

Version 8 (effective date: 26th February 2020)

Appendix 5. Request Form (consent in English)

THE CHINESE UNIVERSITY OF HONG KONG

The CUHK – BCM Joint Centre for Medical Genetics

Joshua Hellmann Foundation Newborn Metabolic Screening Program

Tel: 6806-4590 Fax: 3505-4810

Affix Baby's GUM label here or fill in the following Name: DOB: ID / Hospital no.:	Affix Mother's GUM label here or fill in the following Name: ID / Hospital no.:

Gestation age at delivery :_____week_____day Hospital / Place of birth: _____

Birth weight :_____grams Date and time of birth :____/____/_____:____am/pm

Ethnicity: ☐ Chinese ☐ Caucasian ☐ Indian ☐ Pakistani ☐ Other (please specify:_____)

Feeding type: ☐ Breast ☐ Formula ☐ Mixed *(please wait until feeding for > 24 hrs before blood collection)*

Baby / Mother: ☐ Antibiotics (_____) ☐ Steroids

Family history: ☐ IEM ☐ Congenital adrenal hyperplasia ☐ Cystic fibrosis

Screening Test:	☐ **IEM & CAH** Minimum 4 blood spot circles	☐ **IEM, CAH and Cystic Fibrosis** Minimum 5 blood spot circles

Referring doctor information (contact person for positive results)

Name: Dr_____ Signature: _____

Phone: _____ Fax:_____

Hospital or Clinic: _____

Dried Blood Spot (DBS) Information

See back of collection card for DBS collection and handling guide.
Provide baby's name, ID / hospital reference no, and date of birth on the collection card.

DBS collection date:_____ DBS collection time:_____

Test information: ☐ 1st sample ☐ 2nd sample ☐ Pre-transfusion sample ☐ Others:_____)

Parent consent:

1. I understand the purpose and possible results of the newborn metabolic screening test.
2. I understand that my baby will be called back for a second heel prick if the first sample showed uncertain or positive results.
3. I understand that, in rare circumstances, variant forms of target diseases may escape screening.
4. I understand that my baby's screening report will appear in the Hospital Authority computer system.
5. I understand that leftover samples may be retained for laboratory internal use and medical research after all identifying information has been removed. Refusal to permit the use of my baby's sample will not affect the screening test result.
   If you don't want your baby's sample to be kept for these purposes, please tick here: ☐

Name of mother / father:_____ Tel no.:_____

Signature of mother / father:_____ Date of signature:_____

Version 8 (effective date: 26th February 2020)

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