Joshua Hellmann Foundation
Newborn Metabolic Screening Program

Clinical Protocol

Centre of Inborn Errors of Metabolism
The Chinese University of Hong Kong

Version 7

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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) and congenital adrenal hyperplasia (CAH) (newly 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 of the target IEM 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 400 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. The most common (>90%) cause of CAH is 21-hydroxylase deficiency. Classic CAH describes patients with no or minimal residual enzyme activity who present at birth or soon after birth. Its incidence is around 1 in 10,000 to 1 in 20,000. There are two different forms of classic CAH – salt-wasting form (75%) and simple virilization form (25%). For the salt-wasting form, affected patients cannot produce adequate cortisol and aldosterone. Low levels of these hormones may cause nausea and vomiting, tiredness, dehydration, and weight loss. In the most severe cases, 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. For the simple virilizing form, 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 affected babies early so that salt-losing crisis can be prevented by prompt treatment.

**Screening method**

The CAH screening test measures the level of 17-hydroxyprogesterone (17-OHP) in 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 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 elevation of 17-OHP require clinical assessment and immediate follow-up diagnostic testing.

Target babies

All healthy newborns born at or after 34 weeks of gestation\(^1\) who have completed oral feeding for 24 hrs are suitable for the screening test. Dried blood spots (DBS) should be collected after completion of oral feeding for 24 hrs and up to the 7th day after birth. The best time for screening for the majority of IEM and CAH is between 48 and 72 hours of life. This means that DBS collection is best done in hospital before the babies are discharged.

Premature (< 34 weeks of gestation), very low-birth-weight (< 2,000 g) and sick newborns who require admission to Neonatal Intensive Care units, parenteral nutrition, blood transfusion or

---

\(^1\)During Phase I (15 July 2013 – 31 August 2013), we accepted DBS from healthy term newborns (≥37 weeks of gestation). Starting from 1 September 2013, we have extended our service to include preterm babies with gestation ≥ 34 weeks
other medical treatment are NOT SUITABLE for the screening test at the current stage of development of the program.

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 prior logistics arrangement. Please contact our laboratory at 2632 2313 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 **University Pathology Service**, the address is set out at the end of this document.

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. Parents 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 **University Pathology Service**, 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.
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 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 cards to the laboratory**

1. Send all completely dried collection cards to the laboratory as soon as possible.
2. If DBS cards cannot be delivered to the laboratory on the day of collection, store them in a cool dry place at room temperature for no more than four days.
3. Transport each collection card in a separate envelope. DO NOT use plastic bag.
4. Deliver collection cards and completed request forms to **University Pathology Service**, the address is set out at the end of this document.
5. Collection cards reception time:
   Mondays to Fridays (except for public holidays) 9:00am – 4:00pm (closed between 1:00 – 2:00 pm)
6. Collection cards received before 11:00 am will be analyzed on the same day.
7. 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. Blood collected before 24 hours of age.
2. Incomplete information on request form or collection card, making it impossible to determine the baby’s identity or age at the time of collection.
3. Collection card arrived at the laboratory more than one week after it is collected.
4. Insufficient quantity of blood on the card to perform the analysis.
5. 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 Centre of Inborn Errors of Metabolism, The Chinese University of Hong Kong at Prince of Wales Hospital or other designated clinics for blood collection.

**Turnaround time**

A report will be issued within three working days after the collection card is received by the laboratory.

**Storage and disposal of collection cards**

All collection cards will be stored in the laboratory for a minimum of two years according to laboratory accreditation guideline. The laboratory will ensure appropriate and proper protection of sensitive personal information. With informed consent from parents, the laboratory may store the collection cards for more than two years and use them for internal study 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.(1) Newborn screening and early treatment can improve the nutritional, growth and intellectual outcomes in CF patients.(2)

The incidence of CF varies in different ethnic groups (3-6):
- 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.(7-9) 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.(10)

**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 4 completely filled circles are required.*

**Testing laboratory**
Testing algorithms
Tier 1 test: immunoreactive trypsinogen (IRT)
Tier 2 test: mutation analysis of 23 CFTR mutations

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. 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 two weeks.
### Enquiries

| Centre of Inborn Errors of Metabolism, The Chinese University of Hong Kong |
| Enquiry hotline for general public (during office hours): 2632 4219 |

| General enquiry (e.g. request form, collection card and DBS reception) |
| University Pathology Service |
| Room 33016, 1/F, Clinical Sciences Building, Old Block |
| Prince of Wales Hospital |
| 30-32 Ngan Shing Street, Shatin, N.T., Hong Kong |
| Tel: 2632 2313  Fax: 2636 0540 |

| Newborn screening laboratory |
| Dr YUEN Yuet Ping Liz |
| Clinical Associate Professor, Department of Chemical Pathology |
| Pager: 7382 2322 |
| Email: lizyuenyp@cuhk.edu.hk |

| Dr LAW Lap Kay Eric |
| Adjunct Assistant Professor, Department of Chemical Pathology |
| Pager: 7382 1195 |
| Email: ericl law@cuhk.edu.hk |

| Paediatricians |
| Dr HUI Joannie |
| Honorary Assistant Professor, Department of Paediatrics, CUHK |
| Pager: 7382 1409 |
| Email: joanniehui@cuhk.edu.hk |

| Dr CHONG Shuk Ching |
| Clinical Professional Consultant, Department of Paediatrics, CUHK |
| Pager: 7382 1400 |
| Email: chongsc@cuhk.edu.hk |
References

Appendix 1. Workflow of the Newborn Metabolic Screening Program

Collect dried blood spot & send card to laboratory

Laboratory analysis (MS/MS for IEM and 17-OHP for CAH)

- Normal result
  - Send out report to referring doctor
- Uncertain result
  - Send out report to referring doctor to arrange repeat dried blood spot
- Positive result
  - Notify referring doctor to arrange referral to Metabolic Paediatrician

Abnormal result
### Appendix 2. Target Inborn Errors of Metabolism

<table>
<thead>
<tr>
<th>Inborn Errors of Metabolism</th>
<th>ACMG# classification</th>
<th>Key metabolites</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Amino Acid Disorders</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Phenylketonuria (PKU)</td>
<td>Core</td>
<td>↑ Phe</td>
</tr>
<tr>
<td>Maple syrup urine disease (MSUD)</td>
<td>Core</td>
<td>↑ Leu/Ile</td>
</tr>
<tr>
<td>Citrullinaemia type 1</td>
<td>Core</td>
<td>↑ Cit</td>
</tr>
<tr>
<td>Argininosuccinic aciduria</td>
<td>Core</td>
<td>↑ Cit</td>
</tr>
<tr>
<td>Homocystinuria</td>
<td>Core</td>
<td>↑ Met</td>
</tr>
<tr>
<td>Tyrosinaemia type 1</td>
<td>Core</td>
<td>↑ Tyr, SA*</td>
</tr>
<tr>
<td>Arginase deficiency</td>
<td>2°</td>
<td>↑ Arg</td>
</tr>
<tr>
<td>Defects of biotin cofactor biosynthesis or regeneration</td>
<td>2°</td>
<td>↑ Phe</td>
</tr>
<tr>
<td>Citrullinaemia type 2 (Citrin deficiency)</td>
<td>2°</td>
<td>↑ Cit</td>
</tr>
<tr>
<td><strong>Organic Acid Disorders</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Propionic acidaemia (PA)</td>
<td>Core</td>
<td>↑ C3</td>
</tr>
<tr>
<td>Methylmalonic aciduria (MUT, cbl A/B)</td>
<td>Core</td>
<td>↑ C3</td>
</tr>
<tr>
<td>Isovaleric acidemia (IVA)</td>
<td>Core</td>
<td>↑ C5</td>
</tr>
<tr>
<td>β-ketothiolase deficiency (BKT)</td>
<td>Core</td>
<td>↑ C5OH, C5:1</td>
</tr>
<tr>
<td>Glutaric acidemia type 1 (GA1)</td>
<td>Core</td>
<td>↑ C5OH</td>
</tr>
<tr>
<td>3-Hydroxy-3-methylglutaryl-CoA lyase deficiency (HMG)</td>
<td>Core</td>
<td>↑ C5OH</td>
</tr>
<tr>
<td>Multiple carboxylase deficiency (MCD)</td>
<td>Core</td>
<td>↑ C5OH</td>
</tr>
<tr>
<td>3-Methylcrotonyl-CoA carboxylase deficiency (3MCC)¶</td>
<td>Core</td>
<td>↑ C5OH</td>
</tr>
<tr>
<td>Malonic aciduria (Malonyl-CoA decarboxylase deficiency)</td>
<td>2°</td>
<td>↑ C3DC</td>
</tr>
<tr>
<td>3-Methylglutaconic aciduria type I (3MGA)¶</td>
<td>2°</td>
<td>↑ C5OH</td>
</tr>
<tr>
<td>Cbl C/D</td>
<td>2°</td>
<td>↑ C3</td>
</tr>
<tr>
<td><strong>Fatty Acid Oxidation Disorders</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Primary carnitine deficiency (Carnitine update defect, CUD)¶</td>
<td>Core</td>
<td>↓ C0</td>
</tr>
<tr>
<td>Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)</td>
<td>Core</td>
<td>↑ C8</td>
</tr>
<tr>
<td>Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)</td>
<td>Core</td>
<td>↑ C14:1</td>
</tr>
<tr>
<td>Long-chain hydroxyl-acyl-CoA dehydrogenase (LCHAD)</td>
<td>Core</td>
<td>↑ C16OH</td>
</tr>
<tr>
<td>Trifunctional protein deficiency (TFP)</td>
<td>Core</td>
<td>↑ C16OH</td>
</tr>
<tr>
<td>Disorder</td>
<td>Type</td>
<td>Primary Metabolite</td>
</tr>
<tr>
<td>------------------------------------------------------------------------</td>
<td>------</td>
<td>-------------------</td>
</tr>
<tr>
<td>Carnitine palmitoyltransferase I deficiency (CPT1)</td>
<td>2°</td>
<td>↑ C0</td>
</tr>
<tr>
<td>Carnitine palmitoyltransferase II deficiency (CPT2)</td>
<td>2°</td>
<td>↑ C16</td>
</tr>
<tr>
<td>Carnitine-acylcarnitine translocase deficiency (CACT)</td>
<td>2°</td>
<td>↑ C16</td>
</tr>
<tr>
<td>Multiple acyl-CoA dehydrogenase deficiency (Glutaric aciduria type 2, GA 2)</td>
<td>2°</td>
<td>↑ C4, C5</td>
</tr>
<tr>
<td>Medium/short-chain hydroxyl-acyl-CoA dehydrogenase deficiency (M/SCHAD)</td>
<td>2°</td>
<td>↑ C4OH</td>
</tr>
<tr>
<td>(3-Hydroxyacyl-CoA dehydrogenase deficiency)</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

#ACMG: American College of Medical Genetics (Ref 12, 13). 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

<table>
<thead>
<tr>
<th>Amino acid profile</th>
<th>Details: Diagnostic test for amino acid disorders Abnormal in some organic acid disorders</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sample requirement:</td>
<td>Heparin blood 3 mL (minimum 1 mL) or Plasma 1 mL (minimum 0.2 mL)</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Carnitine and acylcarnitine profile</th>
<th>Details: Diagnostic test for fatty acid oxidation disorders Abnormal in some organic acid disorders</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sample requirement:</td>
<td>Clotted blood in plain bottle 1 mL or Serum 0.5 mL</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Urine metabolic screen</th>
<th>Details: Diagnostic test for organic acid disorders Abnormal in some amino acid and fatty acid oxidation disorders</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sample requirement:</td>
<td>Fresh spot urine in plain bottle 20 mL</td>
</tr>
</tbody>
</table>
Appendix 4. Request Form (consent in Chinese)

CHEMICAL PATHOLOGY
Newborn Metabolic Screening Program
Tel: 2632-2313 Fax: 2636-0540

| 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 4 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  □ Repeat sample □ Other samples (please specify: ___________________)

全家同意書:

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，剩餘的血樣經刪除所有身份信息後，可能會被保留並用於實驗室內部的試驗。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“X”號：□

母／父親姓名: ___________________  電話號碼: ___________________
母／父親簽署: ___________________  簽署日期: ___________________
Appendix 5. Request Form (consent in English)

CHEMICAL PATHOLOGY
Newborn Metabolic Screening Program
Tel: 2632-2313 Fax: 2636-0540

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: ____________________________  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 □ Repeat sample □ Other samples (please specify: ____________________________

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 leftover samples may be retained for laboratory internal use after all identifying information has been removed. Refusal to permit the use of my child’s sample will not affect the screening test result. If you don’t want your child’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 4 (effective date: 15th February 2016)