MetaScreen™

安康檢

100+
代謝病篩查

FDA 認證・GC-MS 檢測技術

簡易・無痛

cordlife

MetaScreen™ 安康檢

聯絡安康檢™

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安康檢™為以下公司的註冊品牌:

cordlife

參考資料:
1. Newborn screening programme for IEM 2015-2017
2. Evaluation of the 18-month "Pilot Study of Newborn Screening for Inborn Errors of Metabolism" in Hong Kong, HK J Paediatr (New Series) 2020;25:16-22
3. Two Siblings Born With Isovaleric Acidemia: One Caught by Newborn Screening, One Wasn't, Posted on May 21, 2013, By Jana Monaco, Newborn Screening Parent Advocate
4. Family-stories_ben. Save Babies Through Screening Foundation

DCR No. 2358 Meta_LF_V2

代謝病 - 孩子的無聲殺手

每1,682名香港嬰兒，便有1人是代謝病(先天性代謝缺陷)患者[1]，遠高於中國及台灣約1/5,800的發病率[2]。現時代謝病不可以完全根治，一般無法在產前檢查出來。部分代謝病患者在出生時或幼兒階段都沒有明顯病徵。即使察覺到有病徵，這些病徵或被誤診為其他疾病，例如食慾不振、嘔吐、肌肉無力、發展遲緩及昏睡等。患者病發時身體會突然急速轉差或可導致智商每周下跌一度，嚴重者可導致終身殘缺甚至死亡。

代謝病是身體缺乏部份酵素引致

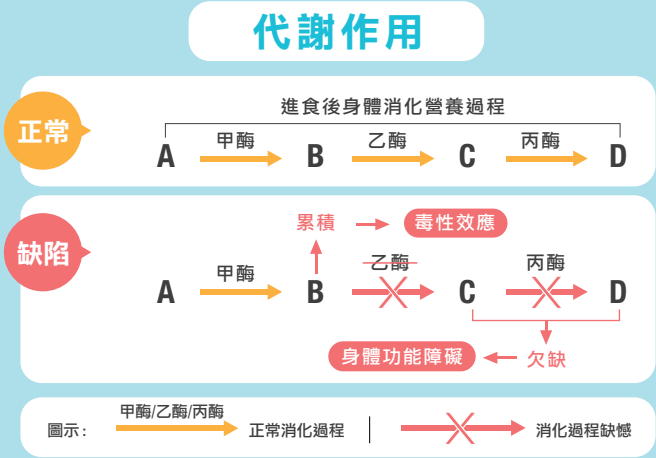
有代謝病的嬰幼兒由於缺乏部分酵素以進行正常的新陳代謝功能，令身體積聚過多的有毒物質，造成毒性效應；另外亦有機會令身體缺乏重要營養，引致身體功能障礙。部份代謝病有機會數年後才病發而期間沒有明顯病徵，往往令父母難以及時察覺和跟進，造成不可挽回的傷害。

患有代謝病的嬰幼兒表面上是正常的

由於缺乏部份酵素，無法正常消化飲食中的營養素（包括母乳、配方奶粉等）

引發毒性效應並出現身體功能障礙

代謝作用



及早檢測 把握黃金治療時機

儘管某些嚴重的代謝病很早就會表現出症狀，但仍存在一些較遲發病的代謝病，患者平均年齡可去到7歲。

其實大部份代謝病可利用飲食治療和藥物輔助控制病情，所以透過早期診斷，可以有助提高代謝病治療的成效。例如普遍的乳糖不耐症只需於日常飲食中減少乳製品攝取便可減輕病症，而其他較嚴重的代謝病包括苯丙酮尿症、異戊酸血症及生物素酶缺乏症等，則要在發病前避免進食或額外補充特定成份以有效控制病情。

如何管理代謝病？*

* 並不適用於所有代謝病，請咨詢醫生專業意見。

代謝病可令智商每一周下跌一度

“高苯丙氨酸血症(HPA)是最常見影響腦部發育的先天性代謝病，智商會每周下跌1度，嚴重者可跌至50多度，同時會有發展遲緩、自閉、痙攣等不可逆轉的後遺症。”


篩查防智障代謝病
社會可年省近千萬
英國驗9病 港只測兩種

明報 09/06/2014

代謝病出生後幾年沒有明顯病狀

“代謝病與遺傳因子有關，一般無法在產前檢查出來，要靠嬰兒出生後做篩查，而且出生頭幾年沒有明顯病狀，待發病時才確診已錯失黃金治療時間。專家建議所有新生兒都要做篩查，及早診斷”


東方日報
新生嬰篩查 揪出孤兒症
把握黃金治療期 免危害器官損智力

東方日報 21/01/2017

代謝病篩查 令兄妹踏上不同人生路[3]

哥哥沒有接受檢測 - 永久損害

- 哥哥Stephen於三歲時昏迷，確診患上異戊酸血症
- 由於錯過黃金治療時間，出現智力和發育遲緩、癲癇發作，造成永久傷害
- 視覺亦受損，現時需利用胃管進食

妹妹有接受檢測 - 正常發展

- 妹妹Caroline透過篩查發現同樣患上此症
- 能夠把握黃金治療期間接受藥物管理及治療
- 成長與正常小孩無異

感冒發燒 竟被揪出隱藏7年的遺傳代謝病*

- 一名7歲的小童因發燒、咳嗽、嗜睡、精神狀態差及血細胞下降入院。綜合病情，初步懷疑患血液性疾病及顱內感染
- 住院期間，雖然醫生進行完善檢查及治療，但小童病情反覆，出現低血糖、肺炎、酸中毒、電解質紊亂及部分內臟功能紊亂等情況，有生命危險
- 經遺傳代謝與內分泌專科會診後，最終考慮診斷小童患上甲基丙二酸血症 (Methylmalonic Aciduria)。醫生利用加強營養、低蛋白飲食等治療，成功令小童體內的甲基丙二酸水平明顯下降，情況好轉，最後順利出院

* 新浪育兒網 29/08/2018

去世的2歲半代謝病患童病發前沒有明顯病徵[4]

- Ben在短暫的2年半人生中，一直如其他幼兒一樣正常成長
- 某天，Ben毫無徵兆下突然發燒及嘔吐。僅僅數個小時便不省人事，這個表面正常的孩子在發病的12個小時內不幸去世
- 醫生耗時一個月並經解剖後才得知Ben的真正死因是患有脱氫酶缺乏症(MCAD)。
- 最令人痛心的是父母發現Ben的病症可以通過簡單代謝病篩查試驗即可被檢測，避免造成無可挽回的傷害

代謝病教室

為何孩子出生時沒有異樣並一直健康成長，卻突然會患上代謝病呢？

原來部份代謝病(如甲基丙二酸血症、丙酸血症及同型半胱氨酸血症等)會出現延遲發病的情況，即這些代謝病除了會在嬰兒時期發病之外，亦有機會到幼兒、青少年甚至青年時期才發病，病狀與嬰兒發病相似。

雖然患有代謝病的孩子表面上看起來很健康，但體內是處於一種代償期與失代償期的平衡狀態(即發病與不發病的中間狀態)，當孩子受到刺激時(如劇烈運動及感冒發燒等)，其體內的“平衡”就會被打破並出現症狀，病情更有機會急轉直下。

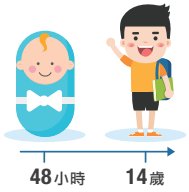
安康檢™ 全面代謝病篩檢

專業可靠

FDA認可GC-MS技術利用尿液進行篩檢，可有效檢測>100種代謝性疾病，提供高準確度及專屬結果

簡易無痛

只需使用專用試紙即可輕鬆收集尿液樣本，不會對孩子造成任何傷害及不適



靈活彈性

由孩子出生48小時直至14歲均可進行檢測，越早進行越快找出孩子健康風險

CAP ACCREDITED
COLLEGE of AMERICAN PATHOLOGISTS

權威認證

香港科學園實驗室榮獲美國病理學會CAP認證，操作流程、管理及檢驗均符合嚴格要求

快速報告

報告可於10-14個工作天內發出，可按情況盡快跟進治療

代謝病 篩檢方法比較

	安康檢™ 氣相色譜-質譜法(GC-MS)	其他平台 串聯質譜(MS-MS)
採樣及風險	尿液: • >3000種可檢測的代謝物，濃度比血液中高 • 非入侵性及安全	血液: • 代謝物含量濃度相對較低 • 入侵性和有痛楚，有感染機會 • 部分代謝病不會影響血液中的代謝物濃度，需要作進一步檢測
代謝病檢測數量	>100種代謝病 • 氨基酸代謝病 • 有機酸代謝病 • 脂肪酸代謝病 • 糖代謝病 • 尿素循環障礙 • 乳酸、丙酮酸+TCA循環不正常 • 嘌呤類，嘧啶代謝病 • 過氧化物酶體症	約40種代謝病 • 氨基酸代謝病 • 有機酸代謝病 • 脂肪酸代謝病
生物指標數量	使用多個標記: 高特異性，由單一測試得出可靠結果	多為單一標記: 非特異性，需作第二輪測試核實
診斷時間	更快: 篩選及確定同步進行，然後到最終診斷	較慢: 先篩選，後確定，然後到最終診斷
驗證檢測	通過單一測試完成篩選及確認，少數情況需要作進一步驗證 • 酵素測定 • 基因檢測	需進行以下測試確認: • GC - MS分析 • 酵素測定 • 基因檢測
外來干擾	氣相色譜可排除藥物和外源性化學物質對分析結果的潛在干擾	某些藥物和外源性化學物質有機會干擾分析結果

安康檢™ 服務計劃

代謝病種類	安康100+
氨基酸症及有機血症	59
糖代謝紊亂	8
脂肪酸代謝紊亂	6
過氧化物酶體病	5
嘌呤、嘧啶代謝紊亂	9
乳酸血症、超丙酮酸血症	7
其他	12
	106

Metascreen™ Service Package

Groups of inborn errors of metabolism (IEMs)	Meta 100+
Amino Acidopathies & Organic Acidemia	59
Disorder of Sugar Metabolism	8
Disorders of Fatty Acid Metabolism	6
Peroxisomal Diseases	5
Disorders of Purine, Pyrimidine Metabolism	9
Lactic academia, Hyperpyruvic Metabolism	7
Other IEMs	12
	106

安康檢™ 服務訂購

1. **訂購安康檢™**
   聯絡您的醫護人員或我們的服務專員，訂購安康檢™
2. **採集樣本**
   請參照收集工具內的指示採集寶寶/孩子的尿液樣本。新生嬰兒於第二天(哺乳至少24小時後)即可進行採樣
3. **收取樣本**
   通知速遞公司，收取樣本
4. **篩檢及分析**
   於CAP認證實驗室進行代謝病篩查分析
5. **報告**
   將會向您發送一份篩查報告
6. **跟進**
   如報告出現陽性結果，我們將盡快通知您或您的醫生

Metascreen™ Service Order

1. **Order Metascreen™**
   Inform your healthcare professional or our representatives for service registration
2. **Collect Urine**
   Please follow the collection procedure guidelines to collect your baby's / child's urine sample. Newborn's one can be done at least 2 days old (with one feed 24 hours ago)
3. **Call Courier**
   Call courier hotline for urine sample pick up
4. **Screen & Analyse**
   Urine sample will be screened and analysed in CAP accredited laboratory
5. **Receive Result**
   A screening report will be sent to you
6. **Escalate to Doctor**
   We will inform you or your doctor if the test result is positive

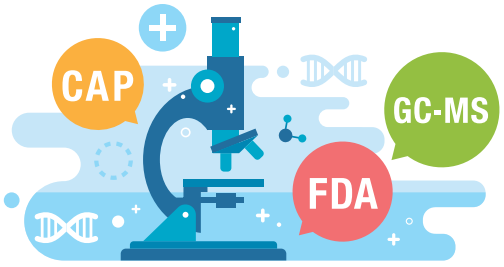
Comparison of Metabolic Disorders Testing Technology

	Metascreen™ GC-MS Tech.	Others MS-MS Tech.
Time to diagnosis	Faster: From screening-cum-confirmation to diagnosis	Slower: From screening to confirmation then finally to diagnosis
Necessity for further confirmation	Screening and confirmation from one single test, though occasionally requires further confirmation via: • Enzyme assays • Genetic testing	Requires confirmatory test via: • GC/MS analysis • Enzyme assays • Genetic testing
Interference	Chromatography separates potential interfering compounds, such as drugs and exogenous chemicals	Certain drugs and exogenous chemicals can interfere with the analysis

Comparison of Metabolic Disorders Testing Technology

	Metascreen™ GC-MS Tech.	Others MS-MS Tech.
Specimen and risk	Urine • Contains more than 3,000 detectable metabolites, with many in higher concentration than in blood • Non-invasive, safe, and simple for collection by parents or hospital staff	Blood: • Though contains similar number of detectable metabolites as urine, many not in as high concentration than in urine • Invasive, painful, requires technical expertise, risk of infection • Certain metabolic disoreders do not alter blood amino acid or acylcarnitine profile, hence further testing is required to differentiate type o disorder
No. of disorders detected	100+ IEMs from 9 groups of IEMs • Amino acidemia • Organic acidemia • Disorders of fatty acid metabolism • Disorders of sugar metabolism • Urea cycle disorders • Lactate, pyruvate + TCA cycle abnormalities • Purine, pyrimidine disorders • Peroxisomal disorders	40+ IEMs from 3 groups of IEMs • Amino acidemia • Organic acidemia • Disorders of fatty acid metabolism
No. of biomarkers (analyte profiles) used	Multiple markers used: Highly specific, definitive interpretation from one single test	Mostly single marker used: Non-specific, ambiguous - 2nd tier testing required

Metascreen™ The Most Comprehensive Metabolic Diseases Screening

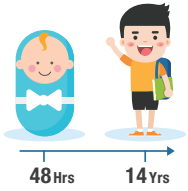


Professional & Reliable

FDA approved GC-MS screening technology using urine to detect >100 types of metabolic disorders with high accuracy and specificity.

Simple & Non-Invasive

The sample can be easily collected by placing a filter paper in urine. This collection is non-invasive and causes no discomfort to your children.



Flexible & Expandable

Screening can be performed from 48 hours after birth or up to 14 years old. Ideally, your children should be screened as early as possible.

International Accreditation

The lab is located at Hong Kong Science and Technology Park and accredited by The College of American Pathologists (CAP), which has stringent standards for laboratory operations, quality and technology.

Timely Report

Results will be available within 10 - 14 working days for prompt follow-up with a paediatrician if necessary.

Late Onset Metabolic Disease Found in a 7-Year-Old Boy Suffered from Fever*

- The boy with fever, cough, lethargy and dropped blood cells was sent to hospital. Preliminary findings suggested he may have blood related diseases and intracranial infection.
- Though comprehensive checking and treatment were given, the boy's life was in danger and showed low glucose level, pneumonia, acidosis, electrolyte disturbances and partial visceral dysfunction etc.
- The boy was diagnosed with Methylmalonic aciduria finally. His condition became better after nutritional treatment and low protein diet, and he could finally discharged from hospital

* http://baby.sina.com.cn/ 29/08/2018

Metabolic Diseases Classroom

Why Children Appeared Normal and Healthy in Childhood but Diagnosed with Metabolic Disorders Suddenly?

Indeed some metabolic disorders (like Methylmalonic aciduria, Propionic acidemia and Hyperhomocysteinaemia) are late onset ones, i.e. they will not only affect infants and babies, but also children, adolescents and even adults with similar symptoms.

Some affected children may appear healthy but their body are actually in an unstable condition (50-50 chance for onset disorders). If they are trigged by external stimulus like strenuous exercises or flu etc, the balance will be broken and symptoms will be shown. Their condition may go worse in a short period of time.

Two Siblings Born with Isovaleric Acidemia - Different Life Paths[3]

No screening test for elder brother - Lifelong Damage

- Stephen was in a coma at the age of 3 and diagnosed with Isovaleric Acidemia
- Missed the optimal treatment period and developed epilepsy, mental and physical retardation
- Experienced visual damage and gastric tube feeding is required

Screening test done for younger sister - Normal Life

- Caroline was diagnosed with same disease through the screening test
- Prompt medical treatment can be arranged
- Same lifestyle as other normal children

No Obvious Symptoms Found in a 2.5 Years Old Dead Child[4]

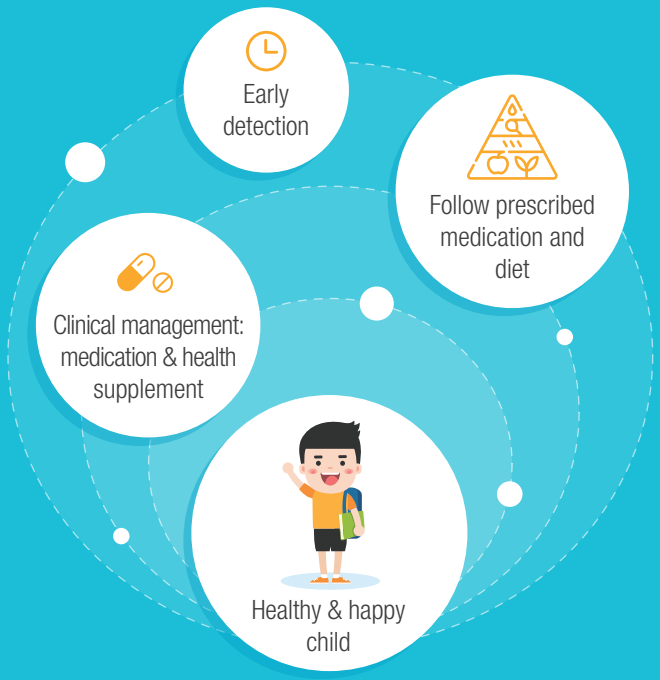
- Ben was growing and developing normally during his brief 2 .5 years of life
- With no other signs such as fever, Ben became ill with vomiting. In just hours, Ben became unresponsive, stopped breathing and his heart stopped beating. The apparent healthy child had passed away within 12 hours
- Doctor had speculated about the cause of Ben's death for a month and concluded Ben's real cause of death, MCAD
- Sadly, his parents learnt that this disorder could have been detected at birth, or any other time before his fatal crisis, with a simple metabolic test

Early Detection Allows for Timely Intervention

Although some serious metabolic diseases are more obvious, there are still some late-onset ones and the average age of children is up to 7.

Most metabolic disorders be controlled by introducing appropriate diet restriction and medical treatment. The outcome will become better through early diagnosis. For example, lactose intolerance can be treated by restricting dairy product intakes. In contrast, some diseases like PKU, Isovaleric aciduria and Biotinidase deficiency would require strict dietary control or specific supplements intake in the treatment.

How Do We Manage The Metabolic Disorder?*



*This do not apply to all IEMS, please consult your doctor for medical advice

Metabolic Disorders: The Silent Killer

In Hong Kong, about 1 in every 1,682 babies is expected to be born with metabolic disorder[1] (inborn errors of metabolism, or IEMs). The prevalence rate is much higher than that of China and Taiwan[2] (about 1 in every 5,800 babies). Meanwhile, metabolic disorders cannot be completely treated and screened through antenatal check-up. Some affected children appear normal without obvious symptoms. The found symptoms like loss of appetite, vomiting, acratia, developmental retardation and lethargy may be misdiagnosed. The health condition of these babies may become worse in a short time or the IQ will be dropped by 1 point per week, and even with lifelong disabilities and have risk of death.

Metabolic Disorders are Caused by Enzymes Deficiency

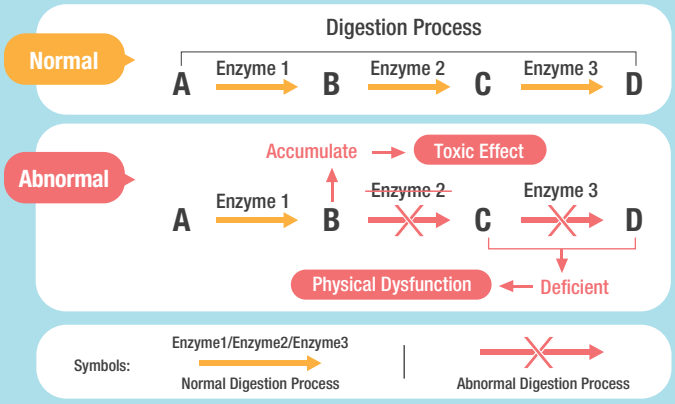
Babies with metabolic disorders lack certain enzymes to maintain the normal metabolic function, causing the build-up of toxic substances or deficiency of critical nutrients. The onset of some metabolic disorders may by delayed by several years after the baby is born with no obvious symptoms, making it difficult for parents to take action and may cause irreversible damage to the children.

Affected child appears normal

Lacks the enzymes to breakdown the nutrients (incl. breast milk, baby formula etc)

Causes toxic effect and physical dysfunction

Metabolic Function



MetaScreen™ 安康檢

Contact Metascreen™

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Service Hotline
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biotech.cordlife.com.hk

The services under Metascreen™ are marketed by:

cordlife

Reference:
1. Newborn screening programme for IEM 2015-2017
2. Evaluation of the 18-month "Pilot Study of Newborn Screening for Inborn Errors of Metabolism" in Hong Kong, HK J Paediatr (New Series) 2020;25:16-22
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4. Family-stories_ben. Save Babies Through Screening Foundation

DCR No. 2358 Meta_LF_V2

MetaScreen™

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100+

Metabolic Disorders Screening

FDA Approved • GC-MS Technology

Easy · Non-Invasive

cordlife