What is Spinal Muscular Atrophy (SMA)?
什麼是脊髓性肌肉萎縮症 (SMA)？

Spinal muscular atrophy (SMA) is a recessive chromosomal genetic disorder in which the anterior horn nerve cells of the spinal cord (motor neurons) degenerate. Since the motor neurons relay signals from the brain to the skeletal muscles, their degeneration will result in muscle weakness and atrophy. The most common form of SMA is caused by deletions or mutations in the Survival Motor Neuron Gene 1 (SMN1) of the fifth chromosome, which is responsible for making a protein called the Survival Motor Neuron (SMN) protein. This protein is vital for healthy functioning of the motor neurons. It is estimated that 1 in every 6,000 to 10,000 newborns is affected by SMA.

脊髓性肌肉萎縮症 (SMA)是一種隱性染色體遺傳疾病，患者的脊髓前角神經細胞（運動神經元）衰退，運動神經元負責把大腦的信號傳遞至肌肉，它們的衰退 導致肌肉萎縮無力。最常見的一類SMA由位於第五染色體的遺傳基因缺失或突變所引起，這組基因叫「運動神經元存活基因」(Survival Motor Neuron Gene 1, 簡稱SMN1)，這基因指令製造運動神經元存活蛋白（SMN），維持運動神經元的健康運作。估計每6,000至10,000新生嬰兒中就有1人受到影響。

What are the Symptoms of SMA?
SMA的病徵是什麼？

The disease mainly affects the motor muscles, especially the large muscles. The child's intelligence and communication skills are generally normal. There are different types of SMA. The severity and age of onset can vary. Severely affected patients will develop symptoms within the first six months, presenting with poor muscle tone, delayed motor development and difficulty in chewing and swallowing. Weakness and inability to cough will lead to recurrent pneumonia, which can lead to early death. The fortunate ones may survive infancy but still cannot manage to sit and walk. Some type of SMA may have symptoms later, and are often less severely affected. These lucky ones will have better prognosis.

疾病主要影響運動肌肉，特別是大肌肉，病重的智商和溝通能力一般沒有問題。SMA有不同類型，嚴重程度和病發年齡亦不同。常見的一種SMA在頭六個月便出現病徵，嬰兒的肢體肌肉欠缺張力，發展遲緩，咀嚼和吞嚥也有困難，咳嗽無力常引致反覆肺炎，嚴重的胸肺感染常致早逝，幸運長大了的也無法坐立行走。部份較輕微的患者遲些才出現病徵，預後亦較好。

How Is Newborn Screening for SMA Carried Out?
SMA新生兒篩查如何進行？

The copies of the Survival Motor Neuron Gene 1 (SMN1) can be measured in the same few drops of blood collected on the screening card. Babies with positive or doubtful screening results will be called back to receive further tests. Once the diagnosis is confirmed, early treatment can limit the progression of the disorder and prevent severe disability. Other than intensive physiotherapy and occupational therapy, new drugs are currently available to increase the production of SMN protein and improve the outcome for some patients.

化驗室可同時利用篩查卡上收集到的血點，測量SMN1 基因的數量，陽性或可疑的個案會獲通知接受進一步的檢測。近年有新藥增加SMN蛋白的產生，改善一些患者的預後。積極的物理治療和職業治療亦有幫助。及早確診，接受治療可以緩減疾病的進程。

What is Severe Combined Immunodeficiency (SCID)?
什麼是嚴重複合型免疫缺乏症 (SCID)？

Severe combined immunodeficiency (SCID) is a group of rare inherited genetic disorders affecting the "T" and "B" white blood cells of the immune system. As a result, the patient is incapable of fighting against common infections and has recurrent life-threatening infections by bacteria, viruses and fungi. Some may have chronic diarrhea and growth failure. The most severely affected patients need to live in a sterile cabin to be protected from all environmental micro-organisms and may succumb within a year. T-cell receptor excision circles (TREC) are circular DNA segments generated in T and B cells during their maturation. Patient with SCID will have low levels of TREC.

嚴重複合型免疫缺乏症（SCID）是一類影響免疫系統的遺傳罕見病。不同器官經常遭受細菌、病毒和真菌的反覆感染，危及性命，亦可引起慢性腹瀉及生長遲緩，嚴重的病人不能和外界接觸，需生活於無菌艙中，一年內便可致命。SCID患者血液中的 "T" 和 "B" 白細胞都出現問題，TREC （T細胞受體切除環）是T細胞和B細胞成熟過程中產生的環狀DNA片段，檢查血液內的 TREC 可助診斷SCID。

What are the symptoms of SCID?
SCID的病徵是什麼？

Infants with SCID usually develop symptoms after 2 to 6 months, when maternal antibody protection is waning. Patients have frequent and recurrent respiratory infections, chronic otitis media, sinusitis, and infections of the skin and oral mucosa. Peeling rash on the skin appears due to uncontrolled infection. Other common symptoms include ulcers in the mouth and tongue, chronic diarrhea, malnutrition, and growth retardation.

患有SCID的嬰兒的健康通常在2至6個月大之後才出現問題，這時由母體胎盤供應的抗體開始下降，孩子出現反覆呼吸道感染、慢性中耳炎、鼻竇炎等疾患。皮膚因長期受感染出現剝落性皮疹，口舌黏膜亦見潰瘍，長期腹瀉，營養不良，生長遲緩亦是常見癥狀。

How Is Newborn Screening for SCID Carried Out?
新生兒SCID篩查如何進行？

This can be measured in the same few drops of blood collected on the screening card. Babies with a positive or doubtful screening result will be called back for more tests to confirm the diagnosis. If SCID is confirmed, protective measures can be employed to prevent debilitating infections. Regular antibody infusions may also be prescribed by doctors to boost their immunity. In selected patients, SCID can be treated with bone marrow transplantation.

篩查SCID，可通過在測量篩查卡上血點的TREC，患者的濃度會比常人低。如果結果屬陽性或可疑，嬰兒將被召回進行確認性的檢測，一旦確診，可以及早採取保護措施，避免反覆感染導致身體衰弱。醫生也可以為病人定期處方抗體注射，提高免疫力，另有些SCID患者可以通過骨髓移植根治頑疾。

How are Results Reported?
篩查結果如何報告？

Results from the screening will be sent directly from CUHK laboratory to HKAH-TW. This can take from 7 to 10 working days. Once we have received the results, the report will be scrutinized by your pediatrician and conveyed to you by phone or at newborn follow up. Most babies (98-99%) will have a normal result. This indicates that the baby has a very low chance of having the type of disease screened.

If the test is positive, the baby is at risk of having the type of disease screened and immediate referral should be arranged to a specialist/pediatrician for further diagnostic workup and management.

Sometimes the results can be uncertain and a new sample is needed. This may happen in about 1% of all screened babies. Parents of babies with an 'Uncertain' result will be contacted to arrange for taking another sample within 14 days of life.

篩查結果會由中文大學的化驗室直接送到本院，過程通常約需7至10個工作天，我們收到報告後便會通知孩子的兒科醫生及閣下。絕大多數嬰兒 (98-99%) 的篩查結果都是正常的，這意味嬰兒患有已篩查的疾病風險很低。

萬一篩查結果呈陽性，這意味嬰兒有風險患有已篩查的疾病，應立即接受專科/ 兒科醫生評估及進一步的檢查和治療。

有時因種種技術問題，篩查結果為不確定，不能作出結論，需要再取新樣本重複篩查。一般約1%左右的篩查可出現這情況，我們會聯絡父母，安排新生兒於出生十四天之內重再取樣本。

How accurate is the screening?
篩查的結果準確嗎？

The accuracy of Screening for Rare Diseases and IEM is extremely high. However, as in all laboratory tests, there is a small chance (though extremely low) that some affected infants may be missed (false negatives), while unaffected infants may be wrongly identified (false positives). Therefore, it is extremely important that all "Uncertain" or "Positive" screening results should be followed by standard diagnostic tests for confirmation.

罕見病及代謝病篩查的準確性非常高，可是任化驗技術都可能有誤差，在極少的情況下，一些患病的嬰兒未能被檢出（假陰性），或有健康的嬰兒被懷疑患病（假陽性）。當篩查結果是「不確定」或「陽性」時，便應覆檢或進行更多檢測作出診斷。

Appointment & Enquiries
預約及查詢

Please call us for appointment & enquires during office hour.

如有任何疑問或預約，歡迎於辦工時間致電與本中心職員聯絡，或瀏覽下列網址：

電話：(852) 2275 6688
網址：www.twah.org.hk

Location 醫院位置

Bus 巴士

39A	Tsuen Wan West Station	荃灣西站	
39M	Tsuen Wan MTR	荃灣港鐵站	
30X	Whampoa Gardens	黃埔花園	
30	Cheung Sha Wan	長沙灣	
A38	Hong Kong International Airport	香港國際機場	

Minibus 專線小巴

95M	Tsuen Wan MTR	荃灣港鐵站
95	Nina Tower	如心廣場
84	Tsuen Wan Chung On Street	荃灣眾安街

To the Hospital Via Castle Peak Road
所有行經青山公路荃灣段之巴士：

+ Alight at Tsuen King Circuit Flyover
+ Take the bus in front of Tsuen King Circuit Police Station
+ 於荃景圍天橋站下車，
+ 然後步行至荃景圍警署轉乘巴士到醫院。

Adventist 港安 Health
Hong Kong Adventist Hospital • Tsuen Wan
香港港安醫院 • 荃灣

Address 地址：199 Tsuen King Circuit, Tsuen Wan, N.T.
新界荃灣荃景圍199號
Telephone 電話：(852) 2275 6688
Fax 傳真：(852) 2275 6767
Website 網址：www.twah.org.hk

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Hong Kong Adventist Hospital - Tsuen Wan

Valid date 有效期至：2022/12/31
The Hospital reserves the right to change or amend details without prior notification.
For the latest information, please visit our website at www.twah.org.hk
如有任何資料調整或變更，本院毋須另行通知。
如欲查閱最新資料，請瀏覽本院網站www.twah.org.hk。

N-2209

Extending the Healing Ministry of Christ
延續基督的醫治大能

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香港港安醫院 • 荃灣

Extended Screening for Rare Diseases and Inborn Errors of Metabolism in Newborn Babies
新生兒罕見病及代謝病擴展篩查計劃

Introduction
簡介

To enhance the neonatal screening program, Hong Kong Adventist Hospital-Tsuen Wan is now offering an extended neonatal screening for rare diseases and inborn errors of metabolism (IEM) for babies born in the hospital. The package of tests is provided by the Joshua Hellmann Foundation Newborn Metabolic Screening Program from the Chinese University of Hong Kong (CUHK). The screening will cover 30 metabolic diseases as well as other rare diseases recommended by experts to be worth screening for our population, so as to allow for early diagnosis and optimization of treatment to improve outcomes.

For more detailed information please visit the official website of CUHK at http://www.fetalmedicine.hk/en/iem.asp

為了進一步擴展初生嬰兒篩查，香港港安醫院－荃灣與香港中文大學合作，提供新一代的新生兒罕見病及代謝病篩查服務。經學者專家們審查，選出本地30種值得篩查的代謝病及多種罕見病，及早診斷，有利達致最佳的治療效果。全套化驗由香港中文大學夏的喬—新生兒代謝病篩查計劃提供及支援。

如欲獲取更多詳情，可瀏覽中文大學網頁：
http://www.fetalmedicine.hk/en/iem.asp

What Kinds of Diseases are Screened?
篩查計劃話哪些疾病?

Not all diseases are worth screening for. Experts need to consider the incidence of the disease and availability of effective treatments for the local population. A screening test with a high call back-rate can cause repeated tests and unnecessary anxiety to the parents. Experts in CUHK have carefully selected the following conditions for this screening program:

並非所有疾病都值得篩查，先要考慮疾病在當地的病發率，及確診後是否可以改善病人的預後。若篩查對某種疾病的準確性不足，會引致覆檢率高、不必要的檢測和父母的焦慮。香港中文大學的專家們經仔細考量，建議將下列兒科疾病加入本篩查計劃：

Inborn Errors of Metabolism (IEM) 先天性代謝病

30 different types of IEM in the following categories:

- Amino acid disorders
- Fatty acid oxidation disorders
- Organic acid disorders

屬不忠義，罕見病就是少見的疾病。美國把罕見病定義作影響少於二十萬份之一人口的疾病，疾病是如此罕見，社會上多數人有時甚至專業人士對它們的所知都甚少。雖然每種罕見病都不常發生，但由於有多達數千種遺傳疾病之多，因此每二千人之中，就有一人因某種罕見病而受困擾。治療方案很有限，因為市場細小，藥廠通常對開發新療法不感興趣，研究對象的招募亦十分困難。

氨基酸障礙
脂肪酸氧化障礙
有機酸障礙

Rare Diseases 罕見病

- Congenital adrenal hyperplasia (CAH)
- X-linked adrenoleukodystrophy (X-ALD)
- Spinal muscular atrophy (SMA)
- Severe combined immunodeficiency (SCID)

- 先天性腎上腺皮質增生症 (CAH)
- 腎上腺腦白質失養症 (X-ALD)
- 脊髓性肌肉萎縮症 (SMA)
- 嚴重複合型免疫缺乏症 (SCID)

The list may be updated periodically as new research appears or when new technologies are available.

隨著新技術的發展或科研結論，該列表可能會不時更新。

Who needs to be screened?
哪些嬰兒需要接受篩查？

Every newborn baby is recommended to be screened unless the baby's health condition is not suitable, e.g., baby is clinically unstable.

香港中文大學的專家們建議，除非嬰寶健康狀況並不適合，例如醫床狀況不穩定，否則每位新生寶寶都應該接受篩查。

What are Diseases?
什麼是罕見病？

Rare diseases are rare, so much so that most people, sometimes even professionals, know very little about them. In the United States, a rare disease is defined as a condition that affects fewer than 1 in 200,000 people. Since there are many thousand types of rare diseases, it is estimated that up to 1 in 2,000 people are debilitated by one or another type of rare disease. Treatment options are limited. Drug companies are generally not interested in developing new treatment for a small market, and research subjects are difficult to recruit.

What are Inborn Errors of Metabolism (IEM)?
什麼是新生兒代謝病？

Inborn errors of metabolism (IEM) are a large group of rare genetic diseases that generally result from a defect in an enzyme or transport protein, causing a block in a metabolic pathway involved in the breakdown of nutrients or the generation of energy. Disruption of the metabolic pathway leads to toxic accumulations of substrates before the block. Different organs may be affected, and patients may present with different disease spectra. Some IEM cause defects in energy production only when the body is under acute stress. Many IEM affect the central nervous system, causing irreversible neurological damage. Although IEM are individually rare, experts estimated that the condition affects 1 in every 4,000 newborn babies in Hong Kong because there are more than hundreds different types of IEM.

先天性代謝病（IEM）是一大群罕見的遺傳疾病，通常由酶或運輸蛋白的缺陷引起，阻斷了分解營養或產生能量的代謝途徑。代謝途徑受阻令有毒物質積聚，可以影響中樞神經系統，導致不可逆轉的損傷。不同的IEM亦可影響不同的器官，引起不同的疾病譜系。有些 IEM 令身體儲存的營養不能轉化成細胞能量，當身體有其他毛病或遇上壓力時才出現問題。雖然 IEM 是罕見的，但因為有數以百種不同類型的 IEM，專家估計，本港每四千名新生兒中，就有一名患有 IEM 。

What are the symptoms of IEM?
IEM有什麼病徵？

Patients with IEM appear healthy at birth, because in the womb the placenta provides nutrients and metabolites and clears wastes for the baby. Symptoms only appear after birth when toxic precursors have accumulated to critical levels or when energy cannot be converted from body stores in times of crisis. Early symptoms are often nonspecific in small infants, such as tiredness, vomiting, poor feeding, abnormal breathing or general weakness. Some patients with IEM may experience sudden deterioration from what appears to be a mild illness to a critical state, with seizures, loss of consciousness, brain dysfunction, unexplained low blood sugar level, or liver failure. Some IEM may present later in childhood, with abnormal facial features, enlarged internal organs, mental retardation or growth failure.

IEM的患者在出生時看起來很健康，因為在媽媽的子宮內，胎盤會提供胎兒所有營養和代謝物的需要及為嬰兒清除廢物。出生後，當毒素積累到臨界水平，或當孩子生病或身體遇上代謝壓力時，問題才出現。嬰兒的早期癥狀可包括嗜睡、嘔吐、餵養不良、呼吸異常或全身無力等，這些病癥在初生時期未必容易察覺。一些 IEM 患者平時十分健康，但得到一些看似輕微的疾病時便突然危急惡化，出現驚厥、喪失意識、腦功能障礙、不明原因的低血糖或肝功能衰竭等現象。一些 IEM 亦可在孩子較大時出現，例如有些可引致面部特徵異常、內臟器官增大、智力遲鈍或生長障礙等。

How to Screen for and Diagnose IEM?
怎樣篩查和診斷代謝病？

A few drops of blood are collected onto a screening card by pricking the baby's heel after completing oral feeding for 1 day. The test can be carried out up to the 7th day of life. Results will be available within a few days.

篩查可在嬰兒吃奶滿1天後至出生後第7天之內進行，只需在嬰兒腳跟後針刺一下，收集數滴血液在篩查卡上，進行檢測，數天後便可得知檢測結果。

What is Congenital Adrenal Hyperplasia (CAH)?
什麼是先天性腎上腺皮質增生症 (CAH)？

The adrenal glands are responsible for producing cortisol, a hormone that regulates our body's stress response to illness or injury. Cortisol is structurally and chemically related to two other types of hormones that regulate salt and water balance and sexual differentiation. Many enzymes are needed in the production pathway of cortisol, and congenital adrenal hyperplasia (CAH) is caused by deficiency of one of the enzymes. Affected patients will not be able to cope with metabolic stress caused by mild illnesses or injury. Accumulation of the hormone's precursors also causes overproduction of other chemically related hormones, which cause undesirable effects. Increase in sex hormones may cause abnormal genitalia in the newborn, or causes precocious puberty later.

腎上腺負責產生皮質醇，它負責調節我們有病或身體受傷時的壓力反應。皮質醇的產生需要不同的酶，先天性腎上腺皮質增生症（CAH）由其中一種酶的缺乏引起。皮質醇的化學結構跟雄性激素及調節鹽份和水份平衡的激素類似，產生的代謝途徑亦有連繫。產生途徑受阻令皮質醇不足，阻斷的化合物積累也會導致結構相關的激素過高。雄性激素增多可令新生兒的生殖器異常發育，也可在將來引起性早熟。

What are the Symptoms of CAH?
CAH的病徵是什麼？

Some baby girls with CAH are diagnosed soon after birth because of abnormal male looking external genitalia caused by excessive male hormones. More often, patients present with feeding difficulties, weight loss and poor growth after discharge from birth. Sometimes, these babies come in acutely with repeated vomiting, dehydration, hypotension, low blood sugar, shock, or even death. Mild cases of CAH may be completely asymptomatic during infancy but present with early sexual development in later childhood.

少數患有CAH的女嬰會因男性荷爾蒙過高，令外生殖器男性化，在初生時期便被診斷出來。但更多病兒往往院後才出現餵食困難、體重降低、生長遲滯，嚴重的會不停嘔吐、脫水至低血壓、休克、甚至死亡。亦有部分輕微的患者在三、四歲後，出現早熟才被發現。

How Is Newborn Screening for CAH Carried Out?
CAH新生兒篩查如何進行？

The screening test is done with the same few drops of blood collected on the screening card. The laboratory will measure a chemical called 17-hydroxyprogesterone (17-OHP) which is elevated in patients with CAH. If the screening is positive, the baby will be called back for more tests to confirm the diagnosis. Once confirmed, patients with CAH can be treated with oral medications to avoid life threatening symptoms. This may also reduce the chance of early puberty and short stature in future.

篩查可用篩查卡上收集的血點進行。實驗室會檢測一種名為17-羥基孕酮（17-OHP）的化學物質，它在CAH患者中會升高。如果篩查呈陽性或不確定，嬰兒將被召回重檢或進行更多診斷性的檢測，一旦確診，便可以在出現癥狀之前以口服藥物治療，如此亦可減少將來出現性早熟和身材矮小的機會。

What is X-linked Adrenoleukodystrophy (X-ALD) ?
什麼是腎上腺腦白質失養症 (X-ALD) ?

Adrenoleukodystrophy is a hereditary condition which results in damages to the membrane (myelin sheath) that insulates nerve cells in the nervous system. The most common type, called X-linked adrenoleukodystrophy (X-ALD), is caused by a genetic defect on the X chromosome and is more common and severe in males. Patients with this disease cannot break down very long-chain fatty acids (VLCFAs), causing them to build up in the adrenal glands and the nervous system especially the brain.

腎上腺腦白質失養症是一種遺傳性疾病，會令患者神經細胞的總繞膜（髓鞘）受損。最常見的一類是因X染色體上的遺傳缺陷所引起的，在男性中更常見和較嚴重，簡稱 X-ALD。患者缺乏一種蛋白，這蛋白在細胞內負責運輸極長鏈脂肪酸（VLCFA），細胞不能運輸和分解VLCFA，積聚在大腦，神經系統和腎上腺中，造成破壞。

What are the Symptoms of X-ALD?
X-ALD的病徵是什麼？

The symptoms of X-ALD can vary among patients and may begin at different ages. The most severe type occurs between the ages of 4 and 8. Affected children are found to deteriorate in school performance, develop attention deficits, behavioral problems, double vision, visual and hearing impairment, or epilepsy. The disease usually progresses rapidly in the next 6 months to 2 years, causing the patient to lose bodily control and become bed-ridden, finally early death. Patients with a milder form of X-ALD may develop symptoms later in adulthood or middle age. Others may present with adrenal insufficiency.

X-ALD患者的癥狀因人而異，可在不同年齡出現。最嚴重的類型常發生在4至8歲之間。兒童的學習表現急劇退步，失去以往的專注力，開始時常被誤會，以為是行為問題，直到出現複視、視力和聽覺衰退、行動不便、失禁或癲癇才被診斷出來。病情常在6個月至2年內便迅速惡化，患者失去身體的控制能力，臥床不起，最後早逝。一些輕度患者可能在成人或中年時才出現癥狀，亦有些病人只患腎上腺皮質功能不全。

How Is Newborn Screening for X-ALD Carried Out?
X-ALD新生兒篩查如何進行？

The X-ALD screening aims to detect babies at risk of severe X-ALD by measuring the concentration of VLCFA, which accumulates in the body of patients with X-ALD. The test can be done with the same drops of blood collected on the screening card. If positive or uncertain, the baby will be called back for more confirmatory tests. When X-ALD is diagnosed before the onset of symptoms, patient's outcome can be improved by early treatment including steroid replacement therapy and bone marrow transplant. It should be noted that the test is not designed to detect mild forms of the disease.

患上嚴重X-ALD的嬰兒，體內積聚VLCFA，化驗室可從篩查卡上收集得的血點，測量VLCFA的濃度。如果結果呈陽性或不確定，嬰兒將被召回進行確認性的檢測，一旦確診，及早治療，包括類固醇替代療法和骨髓移植，可改善患者的預後。注意篩查並不檢測輕度的X-ALD。