新生兒先天性代謝異常疾病篩檢（簡稱新生兒篩檢）

Newborn Screening for Inherited Metabolic Disorders (Newborn screening)

－可以幫助寶寶早期發現先天性代謝異常疾病，早期接受妥善治療，減少疾病造成身體或智能上的損害。

It can help your baby by detecting inherited metabolic abnormalities early, so your baby can have early treatment to reduce physical or cognitive impairment caused by the disease.

親愛的爸爸、媽媽：

Dear parents,

健康的孩子，是家庭的歡樂泉源。如果孩子在成長過程中發生病痛時，常會帶給家庭及社會不同程度的影響；因此，藉此提醒您，讓孩子儘早接受新生兒篩檢及相關健康檢查服務是非常重要的。透過新生兒篩檢，可以幫孩子早期發現症狀不明顯的先天性代謝異常疾病，及早於黃金治療期間提供妥善之診治，使疾病對身體或智能之損害降至最低。為了確定您的寶寶是否罹患先天性代謝異常疾病，將由醫院（所）對出生 48 小時後之新生兒採取少許的腳跟血液，寄交衛生福利部國民健康署指定之新生兒篩檢中心進行相關檢驗。目前政府提供補助之新生兒篩檢檢查項目，如下：

Healthy children are the source of joy for a family. If a child becomes ill during the growth phase, it will often bring about different degrees of influence on the family and society. Therefore, we would like to remind you that it is essential for your child to receive newborn screening and related health screening services as soon as possible.

Through newborn screening, it can help children by detecting congenital metabolic abnormalities with inconspicuous symptoms and provide proper care in time to minimize damage to the body or cognition.

To determine if your baby is suffering from one of the selective metabolic disorders, the hospital will take a small amount of blood from your baby’s heel when the baby is 48 hours after birth. The sample will then send to the Newborn Screening Center designated by the Health Promotion Administration, Ministry of Health and Welfare for tests. Currently, the government provides subsidize on the following newborn screening conditions:

一、 先天性甲狀腺低能症 Congenital hypothyroidism

約每 3 千個寶寶就會有一個。剛出生的寶寶幾乎無異常症狀，通常在出生 2-3 個月後慢慢出現症狀；主要是寶寶體內缺乏甲狀腺荷爾蒙，影響腦神經及身體生長發育。如到了 6 個月以後才治療，大部分會變成智能障礙、生長發育遲緩、身材矮小。但如能及早發現，在出生後 1-2 個月內給予甲狀腺素治療，可使寶寶有正常的智能及身體生長發育。

Around one in every 3,000 newborns is diagnosed with this disorder. Newborns with hypothyroidism are almost asymptomatic and usually start developing symptoms at two to three months after birth. Hypothyroidism is the lack of thyroid hormones in the infant, thus affecting the brain and the development of the body. If left untreated 6 months after birth, most likely, the baby has severe intellectual disability, growth delay, and small body size. If detecting early and providing synthetic thyroid hormone within one to two months after birth, the baby can have a normal intellectual and physical development.

二、苯酮尿症 Phenylketonuria

約每 3 萬 5 千個寶寶就會有一個。通常在出生後 3-4 個月時出現症狀，如：生長發育遲緩，尿液及身體上有霉臭味，日後會出現嚴重智能不足；主要是寶寶體內無法有效代謝食物中的蛋白質。早期發現，於出生後 1 個月內，給予特殊飲食、定期追蹤，大部分的寶寶可有正常的智能發展。

About one in every 35,000 newborns has is diagnosed with this disorder. Symptoms such as growth delay or the mold smell of the urine and body odor usually appear at three to four months after birth. Phenylketonuria can lead to severe intellectual deficiency. Phenylketonuria is a deficiency of the enzyme that metabolizes dietary protein. If detected early (within two weeks after birth), physicians can provide special diet and regular check-ups; most infants can have normal intellectual development.

三、高胱胺酸尿症 Homocystinuria

約每 10-20 萬個寶寶就會有一個。主要是寶寶體內無法有效代謝食物中的蛋白質，若未加以治療，會出現全身骨骼畸形、智能不足、血栓形成等併發症。早期發現，予以特殊飲食及維生素治療，可以防止寶寶智能不足的發生。

About one in every 100,000 to 200,000 newborns is diagnosed with this disorder. Infants with the disease are unable to metabolize dietary protein effectively. If left untreated, the baby may have complications such as skeletal deformity, learning, and intellectual disabilities, and thrombosis. With early detection and introduction of special diet and vitamin treatment, we can prevent that baby from the intellectual disability.

四、半乳糖血症 Galactosemia

約 100 萬個寶寶就會有一個典型的半乳糖血症。主要是寶寶體內無法正常代謝乳糖，通常會出現餵奶後發生嘔吐、昏睡之現象、眼睛、肝臟及腦部損害。早期發現，以不含乳糖及半乳糖之奶製品來代替母乳或一般嬰兒奶粉，可防止疾病之危害。曾經生育過罹患此病症寶寶的媽媽，在懷孕期間最好避免攝取含有乳製品或乳糖的食物，如：牛乳、乳類製品、內臟等，以免造成患有此病症寶寶的可能傷害。

Around one in a million infants is diagnosed with classical galactosemia. Infants with the disease cannot metabolize galactose, and present vomit and drowsy after feeding, and damage to the eyes, liver, and brain. With early detection, dairy products that are free from lactose and galactose will replace breastfeeding or normal infant formula, to decrease the damage made from the disease. Mothers who have given birth to such a baby should avoid foods containing dairy or lactose such as milk/dairy products/offal during her next and following pregnancy. By doing so may eliminate the possibility of harming a fetus with such condition.

五、葡萄糖-六-磷酸鹽脫氫酶缺乏症(G-6-PD 缺乏症，俗稱蠶豆症)

Glucose-6-phosphate dehydrogenase deficiency (G6PD deficiency, favism)

每 100 個寶寶就會有三個。是台灣地區常見的遺傳性疾病，主要是寶寶體內紅血球之葡萄糖新陳代謝發生異常，患有此病症的寶寶在接觸某些藥物時，如：吃蠶豆、接觸萘丸（臭丸）、擦紫藥水，服用磺胺劑及解熱鎮痛劑等，常容易造成急性溶血性貧血，如未及時處理會導致核黃疸、智能障礙，甚至有生命危險。早期確認寶寶健康狀況，避免接觸上述之致病因素，可減少對寶寶的傷害。

Out of every 100 infants, 3 are diagnosed with the disorder. It is the most common hereditary disease in Taiwan. This disease is mainly the abnormal metabolism of glucose in red blood cells. When an infant with this condition is exposed to certain substances (such as broad beans, naphthalene (mothballs), gentian violet, sulfa drugs, antipyretic, analgesic, etc.), he/she is susceptible to acute hemolytic anemia. Failure to receive timely treatment could lead to jaundice and subsequently, intellectual disabilities or death. Early detection and avoiding the substances mentioned above can reduce the damage to your baby.

政府為提供更完善的新生兒篩檢服務，自 95 年 7 月起，除上述 5 項疾病篩檢之外，優先增加 6 項先天性代謝異常疾病篩檢補助項目如下：

To provide a more comprehensive screening service for newborns, since July 2006, in addition to the five conditions mentioned above, the government has prioritized the addition of six screening tests for congenital metabolic abnormalities as follows:

六、先天性腎上腺增生症 Congenital Adrenal Hyperplasia (CAH)

約每 1 萬 5 千個寶寶就會有一個。此症最常見的因素為腎上腺 21-羥化酵素缺乏，臨床表徵因「21-羥化酵素」缺乏的質與量不同而有：(1)「失鈉型」，大部分在新生兒時期因鹽分大量流失會造成緊急危險狀況，若疏於診斷，極可能因而致死。(2)「單純型」，此類女嬰會有異常性徵，成長後無月經、過度男性化、不孕及發育異常。患病之男嬰也會有發育上的問題。如未早期發現，生理與心理矯治均甚困難。(3)「晚發作型」，患者在嬰兒期以後才出現症狀。先天性腎上腺增生症，除了晚發型以外，可經由篩檢早期診斷，早期治療可以避免新生兒生命危險，依其缺乏予以適量補充藥物，可使之正常發育及成長。

Around one in every 15,000 newborn infants is diagnosed with the disorder. The most common defect of this condition is due to adrenal 21-hydroxylase deficiency. Clinical presentation is based on the degree of missing "21-hydroxylase" quality and quantity, and is defined as (1) Salt-wasting type – most newborns of this type will lose too much sodium and result in an emergency. If left undiagnosed, it could lead to death. (2) Simple virilizing type – female newborns of this type have ambiguous genitalia, may experience failure to menstruate, over masculine, difficulties getting pregnant, and abnormal growth. Male newborns of this type would also experience abnormalities in growth. Without early detection, it can be difficult

to treat physical and psychological conditions. (3) Nonclassic CAH – most symptoms of this type will start to show after infancy. Apart from the nonclassic congenital adrenal hyperplasia, the other types can be detected early via the screening and receive treatment to avoid fatality. When treated appropriately with supplements, the child can have normal development and growth.

七、楓漿尿症 Maple syrup urine disease

國內發生率約為 10 萬個寶寶就會有一個。由於患者的體液和尿液會有楓樹糖漿的甜味因而命名為楓漿尿症。罹患典型此症的嬰兒，在開始餵食後數天，會逐漸出現嘔吐、嗜睡、食慾減低、呼吸急促、黃疸、抽搐等現象，嚴重者會意識不清、昏迷甚至死亡。此症是特殊支鏈胺基酸代謝異常的罕見疾病，使得支鏈胺基酸（纈胺酸、白胺酸、異白胺酸）的代謝無法進行順利。早期發現及治療對新生兒是十分重要，可讓寶寶較有正常的生長及智能發展。

About one in every 100,000 newborns is diagnosed with the disorder locally. It is named maple syrup urine disease because the patient's body fluids and urine have the sweetness of maple syrup. Infants with this type of disease usually develop symptoms several days after starting feeding. The symptoms include vomiting, lethargy, loss of appetite, shortness of breath, jaundice, seizures, etc. In severe cases, the affected can experience unconsciousness and even death. This disease is due to the abnormal metabolism of certain branched-chain amino acids. The patient is unable to process branch-chain amino acids such as valine, leucine, and isoleucine. Early detection and treatment are vital for the newborn as it will allow a better chance of normal physical and intellectual growth for the child.

八、中鏈脂肪酸去氫酶缺乏症 Medium-chain acyl-CoA dehydrogenase deficiency

國內發生率低於 30 萬分之一，但歐美約 1 萬 5 千個寶寶有一個，是最常見的一種脂肪酸代謝疾病。通常會在出生後的前兩年出現臨床症狀。寶寶會因缺少中鏈脂肪酸去氫酶，使得脂肪代謝無法順利進行，不完全分解的脂肪堆積在體內產生毒性，對大腦和神經系統造成傷害，引發嘔吐、肝臟腫大、低血酮性低血糖、意識模糊、昏迷及抽搐等現象。雖然有部分病人沒有症狀，這個疾病有 25

%的病例在第一次發作時死亡，也常被誤診為嬰兒猝死症。早期篩檢可預防疾病的發作，在急性期快速治療低血糖症狀，長期治療則是要在就寢前提供碳水化合物點心，避免長時間禁食，以及積極治療感染或胃腸炎等突發狀況。如能妥善預防傷害的發生，最終的預後是相當好的。

Less than one in every 300,000 newborns is diagnosed with the disorder locally. However, there is about one in every 15,000 newborns diagnosed in Europe, and it is one of the most common fatty acids metabolic disorders. Clinical symptoms usually appear in the first two years after birth. The baby is unable to metabolize a group of fats due to the lack of medium-chain acyl-coenzyme A dehydrogenase. The buildup of fats accumulates in the body and becomes toxic, causing damage to the brain and nervous system, triggers vomiting, liver enlargement, hypoglycemia, loss of consciousness, coma, seizures, etc. Although some patients show no symptoms, 25% of the cases may die in the first episode and are often misdiagnosed as sudden infant death. Early screening can prevent the onset of the disease. Combined with rapid treatment of hypoglycemia in the acute phase, and long-term treatment of providing carbohydrate snacks before bedtime, avoid fasting and actively treat unexpected situations such as an infection or gastroenteritis, the disease can be properly managed with good prognosis.

九、戊二酸血症第一型 Glutaric acidemia type 1

國內發生率約 10 萬個寶寶有一個，是一種有機酸代謝異常的的罕見疾病。寶寶因為缺乏戊二基輔酶 A 去氫酶無法正常分解離胺酸與色胺酸，有毒產物(如戊二酸等)過量堆積於血液與組織中，造成漸進的神經症狀及急性的代謝異常。通常寶寶在出生幾個月內可能沒有異常或僅有無症狀的巨腦，但在嬰兒期的晚期逐漸呈現出運動困難、漸進式的舞蹈徐動症、肌肉低張到僵硬、麻痺、角弓反張（四肢向外翻轉，身體呈弓狀）等症狀，也可能會有癲癇或昏睡昏迷的急性發作。早期發現及治療對新生兒是十分重要，可讓寶寶較有正常的生長及智能發展。

About one in every 100,000 newborns is diagnosed with the disorder locally. It is a rare disease related to the abnormalities of certain organic acids. Due to the lack of glutaryl-CoA dehydrogenase, the infant is unable to breakdown lysine and tryptophan normally. The buildup of toxins, such as glutaric acid in the bloodstream and tissues can cause progressive neurological symptoms and acute metabolic abnormalities. Usually the affected newborn may have no abnormalities or only asymptomatic enlarged head within a few months of birth. But in late infancy, the baby gradually shows symptoms of difficulties in movements, progressive athetosis, hypotonic, muscle stiffness, numbness, opisthotonos

(backward arching of the limbs), etc. There may also be an acute episode of epilepsy or slumbering coma. Early detection and treatment are vital for the newborn. It allows the affected to have more normal physical and intellectual development.

十、異戊酸血症 Isovaleric acidemia

國內發生率約為 8 萬個寶寶就會有一個。是一種有機酸代謝異常的罕見疾病。寶寶因為缺乏異戊醯輔酶 A 去氫酶，無法正常分解白胺酸，有毒產物異戊酸過量堆積，進而侵犯神經與造血系統。根據症狀嚴重程度以及發病早晚分為典型及非典型兩種。典型病患在出生後可能與一般嬰兒無異，但逐漸會出現倦怠、噁心、嘔吐、嗜睡、胃口不佳及抽筋等症狀，大量的異戊酸堆積在體內，寶寶身體和尿液會有明顯臭腳汗味道。此時若沒有正確的診斷治療，病患將會逐漸昏迷。而非典型患者發病時間較晚且症狀輕微不明顯，往往在出生後一年才會被診斷出來，有時會被誤判為其他類似疾病。早期篩檢發現後，利用飲食控制和定期追蹤，有良好的治療成效。

About one in every 80,000 newborns is diagnosed with this disease locally. This is a rare disease that is related to the abnormalities of metabolizing a certain organic acid. The affected cannot normally breakdown leucine due to the lack of isovaleryl-CoA dehydrogenase. The buildup of isovaleric acid accumulates and damages the nervous system and hematopoietic system. According to the severity of the symptoms and the onset of the disease, they are divided into typical and atypical. The typical patient may be asymptomatic at birth but will gradually show symptoms of nausea, vomiting, lethargy, poor appetite and cramps. With a large amount of isovaleric acid accumulates in the body, the infant will have a distinctive odor of sweaty feet from the body or urine. If there is no correct diagnosis and treatment at this time, the patient will gradually become unconscious. The atypical patients have a late-onset and mild symptoms that are not obvious. It is often diagnosed one year after birth and sometimes misdiagnosed as other similar diseases. With early screening, the use of diet control and regular monitoring, the treatment has effective results.

十一、甲基丙二酸血症 Methylmalonic acidemia

國內發生率約為 10 萬個寶寶就會有一個。是一種有機酸代謝異常的罕見疾病。寶寶因為甲基丙二醯輔酶 A 變位酶功能異常或鈷胺素代謝異常，導致體內甲基丙二酸、丙酸等有機酸蓄積，造成神經系統損害，嚴重時引起酮酸中毒、低血糖、高血氨、高甘胺酸血症。新生兒、

嬰幼兒期死亡率很高,早期篩檢發現可預防急性發病,適時補充液體、避免酸中毒。對於VitB12有效型的病患,須給予維生素B12治療。對於VitB12無效型的病患,給予特殊配方奶粉及高熱量飲食,可使血液、尿液中的甲基丙二酸濃度維持在理想範圍。

About one in every 100,000 newborns is diagnosed with this disease locally. It is a rare disease that is related to the abnormalities of metabolizing certain organic acids. The affected infant has abnormal function of methylmalonyl-CoA mutase or abnormal cobalamin metabolism that leads to a buildup of organic acids like methylmalonic acid and propionic acid etc. Such buildup can damage the nervous system, and in severe cases, it could cause ketoacidosis, hypoglycemia, hyperammonemia, and hyperglycemia. There is high mortality among newborns and infants. Early screening could detect and prevent the acute onset, replenish fluids at the right time, and avoid acidosis. For patients with abnormal cobalamin metabolism, vitamin B12 should be administered as treatment. For those not responding to vitamin B12, special infant formula and high-calorie diet are given to control the level of methylmalonic acid in the blood and urine at an acceptable range.

為提升新生兒的照護品質，除上述11項新生兒篩檢項目，其他尚未納入衛生福利部公告而可透過串聯質譜儀同時檢驗之項目，因考量其檢測效力是否足供臨床判定、檢驗準確性、有無確診後續發展及有效的治療方法等，經國民健康署檢視10年新生兒篩檢先趨計畫成果，並依照世界衛生組織(WHO)疾病全面篩檢十大「原則」，決定可同時以串聯質譜儀篩檢項目，於108年10月1日起再增加10項。新增補助之新生兒篩檢檢查項目，如下：

In order to improve the quality of newborn care, apart from the 11 aforementioned diseases that are included in newborn screening, other diseases tested simultaneously by tandem mass spectrometry are considered to be included in the panel announced by the Ministry of Health and Welfare. In order to understand the test effectiveness, test accuracy, the status of follow-up diagnosis, and the effective treatment, the Health Promotion Administration reviewed the results from a 10-year newborn screening pilot program. Based on the ten principles of disease screening published by World Health Organization (WHO), another 10 diseases screened by tandem mass spectrometry are included in the government subsidizing conditions starting from October 1, 2018 as follows:

十二、瓜胺酸血症第Ⅰ型 Citrullinemia type I

瓜胺酸血症第Ⅰ型為尿素循環障礙的疾病之一；患者常因無法代謝血氨，而造成高血氨症。在新生兒期發病，早期的一般症狀是餵食欠佳、嘔吐、昏睡、焦躁不安、呼吸急促等，他們的

病情通常會快速地變化，而表現出更嚴重的神經學與自律神經的問題，如果沒有適當的治療，大部份的病人都會死亡或產生併發症。在長期的治療上，患者需限制蛋白質的攝取，使用特殊奶粉補充生長發育所需，並使用特殊藥物以幫助血氨的排出，並定期監測血氨等數值。

Citrullinemia type I is one of the urea cycle disorders. The patient often cannot metabolize ammonia and presents hyperammonemia. During infancy, the early symptoms are poor appetite, vomiting, drowsiness, restlessness, shortness of breath, etc. The condition usually changes rapidly, showing more serious neurological and autonomic nervous system problems. If not treated appropriately, most patients will die or have complications. In the long-term treatment, patients need to limit protein intake, use special infant formula to supplement growth and development, use medication to remove excess ammonia from the blood, and regularly monitor blood ammonia and others.

十三、瓜胺酸血症第 II 型 Citrullinemia type II

瓜胺酸血症第 II 型因為體內 Citrin 蛋白功能缺乏所引起。新生兒期發作型的患者在出生 1~5 個月間便會發生膽汁鬱積性黃疸，肝臟功能不正常，出現多種高胺基酸血症，半乳糖血症及脂肪肝等症狀。嚴重者會導致生長遲緩，異常出血不止或貧血、低血糖，肝臟腫大，甚至肝臟衰竭。治療包括補充脂溶性維生素，盡量維持高蛋白高脂肪飲食。

Citrullinemia type II is caused by citrin deficiency. The neonatal onset of type II citrullinemia will experience cholestatic jaundice, abnormal liver function, elevation of several amino acid levels in the blood, galactosemia and hepatic steatosis, etc. Severe cases could present with delayed growth, abnormal bleeding or anemia, hypoglycemia, hepatomegaly, and even liver failure. Treatment includes taking fat-soluble vitamins, maintaining a high protein, and high-fat diet.

十四、三羥基三甲基戊二酸尿症 3-Hydroxy-3-Methylglutaric Aciduria (HMG CoA lyase deficiency)

白胺酸代謝異常係先天性遺傳性疾病，屬於有機酸血症之一種。患者由於體內無法合成酵素來分解白胺酸，以致體內堆積有害人體的有機酸，並導致血氨值攀升，寶寶將因酸中毒及血氨過高而致智障甚或死亡。除無法代謝白胺酸之外，第二項生理缺陷在於無法製造酮體，以因

應長期飢餓狀況。若能及早限制蛋白質攝取，輔以特殊奶粉配方，並避免長期飢餓，則患者身心發展仍可趨於正常。

It is a congenital disorder in which the body is unable to metabolize leucine normally. It is one of the organic acidemias. The affected is unable to produce the enzyme to process leucine, The accumulation of such organic acid in the body leads to a high blood ammonia level, causing acidosis and hyperammonemia that may kill or intellectually disable an infant. Apart from being unable to metabolize leucine, the affected is also unable to make ketones. This causes the patient is not able to cope with long-duration of fasting. If detected early, babies can have less dietary protein, have supplement with special infant formula, and avoid long duration of fasting. Therefore patient's physical and intellectual development can still be normal.

十五、全羧化酶合成酶缺乏 Holocarboxylase synthetase deficiency

為利用生物素作為輔酶的羧化酶功能不足；寶寶常出現進食困難、呼吸困難、皮疹、脫髮和嗜睡等症狀；在代謝上，患者會有酮乳酸中毒，有機酸血(尿)症和高氨血症。及時補充生物素可以阻止許多併發症發生；但若沒有進行治療，此症可能會導致發育遲緩、癲癇發作及昏迷，甚至可能會危害到生命。

It is a carboxylase deficiency due to inborn error of biotin metabolism. The affected infants often have symptoms such as difficulties in feeding, tachypnea, exfoliative dermatitis, loss of hair, lethargy, etc. In terms of metabolism, the patient can experience ketolactic acidosis, organic acidemia, organic aciduria, and hyperammonemia. Timely supplementation with biotin can prevent many complications. If left untreated, this condition can cause delayed development, seizures, coma, and may even become life-threatening.

十六、極長鏈醯輔酶 A 去氫酶缺乏症 Very Long Chain Acyl CoA Dehydrogenase Deficiency (LCAD)

此症會造成身體無法將特定脂肪轉換成能量，尤其是禁食狀態。其典型症狀出現在嬰兒或兒童早期，包括低血糖、昏睡、以及肌肉無力。患者可能會併發肝臟或危及性命的心臟問題。若症狀始於青春期或成年，通常會有肌肉痛及橫紋肌溶解。當肌肉組織被破壞會釋出稱為肌紅素的蛋白，其經過腎臟進到尿液中會使尿液變成紅色或褐色。飲食方面以少量多餐避免飢餓為主，限制長鏈脂肪酸的攝取與補充肉鹼是主要的治療原則。

This is a condition in which the body is unable to properly breakdown certain fats and transforms into energy, particularly during periods without food (fasting). Signs and symptoms can occur during infancy or childhood and include hypoglycemia, drowsiness, and muscle weakness. The affected babies are also at risk of serious complications such as liver abnormalities and life-threatening heart problems. Signs and symptoms during puberty or adulthood often include intermittent rhabdomyolysis (breakdown of muscle), muscle cramps, and pain. When the muscle is breakdown, a protein called myoglobin is released. Myoglobin passes through the kidney and excretes into the urine, thus making the urine red or brown. In terms of diet, frequent meals with smaller portions are recommended to avoid hunger. The main treatment principles include limiting the intake of long-chain fatty acids and carnitine supplementation.

十七、原發性肉鹼缺乏症 Primary carnitine deficiency

或稱肉鹼運輸障礙。國內發生率約 3 萬分之一。肉鹼(或稱卡尼丁)負責將脂肪酸運送到粒腺體，以進行氧化產生能量。原發性肉鹼缺乏症患者發病時，在一歲以前可以發生高血氨性腦病變，一歲以後發生心肌病變。只要能早期診斷，定時服用肉鹼即可避免發病。

This disease is also known as carnitine transporter deficiency. About one in every 30,000 babies is diagnosed with the disease. Carnitine is responsible for bringing fatty acids into mitochondria to produce energy. Patients with primary carnitine deficiency may experience hyperammonemic encephalopathy before the age of one, and cardiomyopathy after the age of one. As long as the disease is diagnosed early, taking carnitine regularly can avoid the onset of those unwanted symptoms.

十八、肉鹼棕櫚醯基轉移酶缺乏症第Ⅰ型 Carnitine Palmitoyltransferase I deficiency

此症會阻礙人體利用脂肪酸產生能量，尤其是在食物攝取不足時。症狀嚴重程度因人而異，通常於童年早期發病，患者因無法有效利用脂肪酸產生能量，而導致低酮酸性低血糖，也常伴隨肝腫大、肝功能異常、等症狀，並存在神經系統受損、肝衰竭、癲癇、昏迷和猝死的風險；預防低血糖可降低神經受損之風險。為了預防低血糖，嬰兒白天需增加餵食頻率，晚上則接續補充玉米粉；患者在生病或接受手術等特定醫療處置時，不可超過 12 小時未進食；成年患者需以高醣低脂為飲食原則，以確保身體以醣類作為能量的主要來源。

Carnitine Palmitoyltransferase I (CPT I) deficiency is a condition that prevents the body from using certain fats for energy, particularly during periods without food (fasting). The

severity of this condition varies among affected individuals. Signs and symptoms of CPT I deficiency often appear during early childhood. The affected cannot effectively produce energy from fatty acids and leads to hypoketotic hypoglycemia. Patients with CPT I deficiency can also have an enlarged liver (hepatomegaly), liver malfunction, etc. Individuals with CPT I deficiency are at risk for nervous system damage, liver failure, seizures, coma, and sudden death. The prevention of hypoglycemia reduces the risk of nervous system damage. In order to prevent hypoglycemia, infants need to increase the feeding frequency during the day, and then add corn starch during the night. Patients should not go on for more than 12 hours fasting especially when they are sick or undergoing specific medical treatment such as surgery. Adult patients need to be on high sugar and low-fat diet to ensure that the body uses sugar as the main source of energy.

十九、肉鹼棕櫚醯基轉移酶缺乏症第 II 型 Carnitine Palmitoyltransferase II deficiency

此症會阻礙人體利用脂肪酸產生能量，尤其是在食物攝取不足時。最嚴重的新生兒致死型患者在出生後短時間內即可發病，相關症狀包括呼吸衰竭、癲癇、肝衰竭、心肌病變、心律不整及低酮酸性低血糖。晚發型則可能到孩童期才發病。治療主要以減少長鏈脂肪酸的攝取，防止飢餓，避免低血糖，並讓患者有需要時多補充醣類來產生能量。

Carnitine Palmitoyltransferase II (CPT II) deficiency is a condition that prevents the body from using certain fats for energy, particularly during periods without food (fasting). The lethal neonatal form of CPT II deficiency becomes ill soon after birth. Infants with this form develop respiratory failure, seizures, liver failure, a weakened heart muscle (cardiomyopathy), an irregular heartbeat (arrhythmia), and hypoketotic hypoglycemia. The other forms can appear later during childhood. Treatment is mainly based on restricted intake of long-chain fatty acids, avoidance of hunger, preventing hypoglycemia, as well as adding more sugar to the diet to produce energy when needed.

二十、戊二酸血症第 II 型 Glutaric Acidemia type II

此病症主要成因為多發性醯基輔酶 A 去氫酶缺乏所導致，因而造成脂肪酸及支鏈氨基酸代謝出現問題。新生兒可出現低血糖、酸血症、肌肉無力、肝臟腫大等，另外，腳底會有臭味。晚發型則可能到青年期才發病。治療可以補充核黃素與肉鹼，並以高碳水化合物、低脂肪低蛋白為主，防止飢餓，避免低血糖，並讓患者有需要時多補充醣類來產生能量。

This disorder is due to the lack of multiple acyl CoA dehydrogenase and causes metabolic problems with fatty acids and branched-chain amino acids. The affected newborns may have symptoms of low blood sugar (hypoglycemia), acidemia, muscle weakness, enlarged liver (hepatomegaly), etc. In addition, glutaric acidemia type II may also present with an odor resembling that of sweaty feet. Some affected individuals have less severe symptoms that begin later in childhood or adulthood. Treatment includes riboflavin and carnitine supplement, using a high carbohydrate, low fat and low protein diet, avoiding hunger, preventing hypoglycemia, as well as adding more sugar to the diet for energy when needed.

二十一、丙酸血症 Propionic Acidemia

最嚴重的新生兒型在出生後幾個星期即產生症狀，餵食情況差，出現嘔吐、癲癇、肌肉張力低下、脫水、嗜睡、呆滯及腦部病變等症狀。晚發型相對較少見。此症患者需限制蛋白質攝取，尤其是會產生丙酸的胺基酸，因此除少量一般飲食之外，另可給予特殊配方奶粉以提供足夠生長所需的蛋白質及熱量供應。

Babies with the most severe form of propionic acidemia become apparent ill within a few weeks after birth. The signs and symptoms include poor feeding, vomiting, seizures, weak muscle tone (hypotonia), dehydration, lethargy, sluggishness, and brain damage, etc. The late-onset form of this condition is less common. The affected patients need to restrict protein intake, especially the amino acids that produce propionic acid. Therefore, in addition to a small amount of normal diet, special formulas are supplied to provide a sufficient supply of protein and calories for growth.

- 在尚未知道篩檢結果之前，請不要讓寶寶接觸萘丸（俗稱臭丸），也不可任意服用藥物；如有健康上的問題，請務必詢問您的小兒科醫師。如寶寶的篩檢結果為（疑）陽性時，並不代表寶寶已經確定罹患該項疾病，原採血院所或確認檢查醫院，會在最短的時間內協助您的寶寶接受進一步之確認檢查。初(複)檢之篩檢結果，可在採血後約 2 星期獲知，請洽詢原採血院所，或查詢_**新生兒篩檢中心**網站（網址：_）；疾病相關問題，請洽詢衛教諮詢專線：_。
- Before you receive the screening results, please do not let your baby get in contact with naphthalene (mothballs), and do not medicate the baby at will. If the baby has a health problem, be sure to consult your pediatrician. If the screening result of your baby comes back as a positive, it does not mean that the baby has the disease. The original blood collection hospitals or the confirmational referred hospital will assist your baby to undergo

further testing within the shortest time. The screening results of the initial (re-) examination can be obtained in about 2 weeks after the blood collection. Please contact the original blood collection hospitals or search online (www.). For disease-related information, please call the Hotline:

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