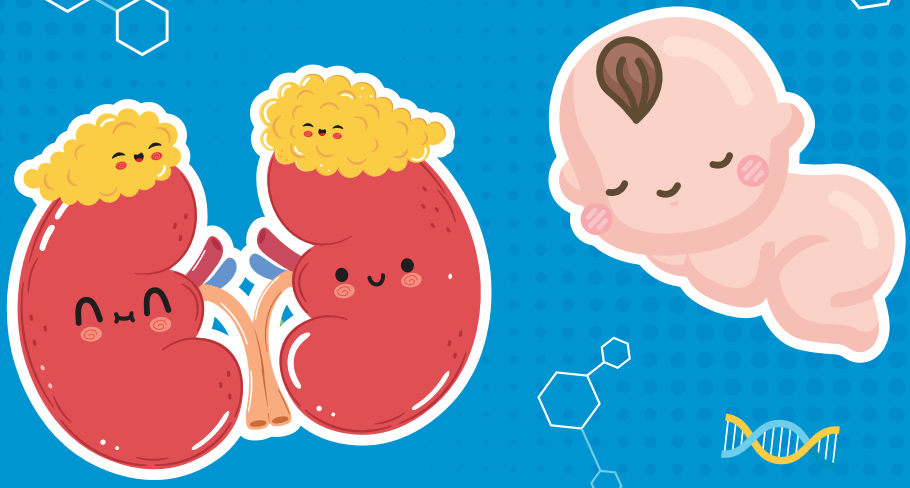
香港兒童醫院
Hong Kong Children's Hospital

認識先天性腎上腺增生症 (CAH)
Understanding Congenital Adrenal Hyperplasia (CAH)



從新生兒篩查到健康成長
From Newborn Screening to Lifelong Care

家庭實用手冊
A Practical Guide for Families

前言 Preface

每一位父母都希望孩子健康成長，而新生兒篩查正默默守護著這份起點。其中，針對先天性腎上腺增生症(Congenital Adrenal Hyperplasia - 簡稱CAH)的篩查，正是這些項目的關鍵之一。

先天性腎上腺增生症(CAH)是一組因腎上腺製造激素功能出現障礙而引起的疾病。CAH是一種常染色體隱性遺傳的疾病 — 也就是說, 只有同時攜帶兩個相關變異基因的情況下，才會發病。

激素是人體重要的訊息傳導物質。當CAH患者腎上腺激素分泌異常時，便會出現不同的臨床表徵與症狀。CAH可分爲多種類型，其中最常見的是21-羥化酶缺乏症。全球平均大約每一萬至兩萬人中，便有一人患病。

雖然CAH是終身疾病，但透過早期篩查和診斷、配合規範治療和定期覆診，絕大多數CAH的孩子都能正常生長發育、學習、工作，並享受美滿的人生。本家庭實用手冊旨在爲您提供清晰實用的CAH資訊。由於每個患有CAH的兒童的臨床表現可能有所不同，以下內容將以最常見的21-羥化酶缺乏症爲例進行說明。

Every parent wishes for their child to have a healthy life. Newborn screening programs help make this possible by checking certain health conditions early on. Among the conditions included in this programme is Congenital Adrenal Hyperplasia (CAH).

CAH is a group of disorders causing problems in hormone production from the adrenal glands. These conditions are inherited in an autosomal recessive way – meaning that only individuals who have two altered copies of a gene are affected with the condition.

Hormones are important messengers in our body. In CAH, faulty adrenal hormone production leads to different presentations and symptoms. There are several forms of CAH and the most common form is 21-hydroxylase deficiency, which affects about 1 in 10000 to 20000 people in the world.

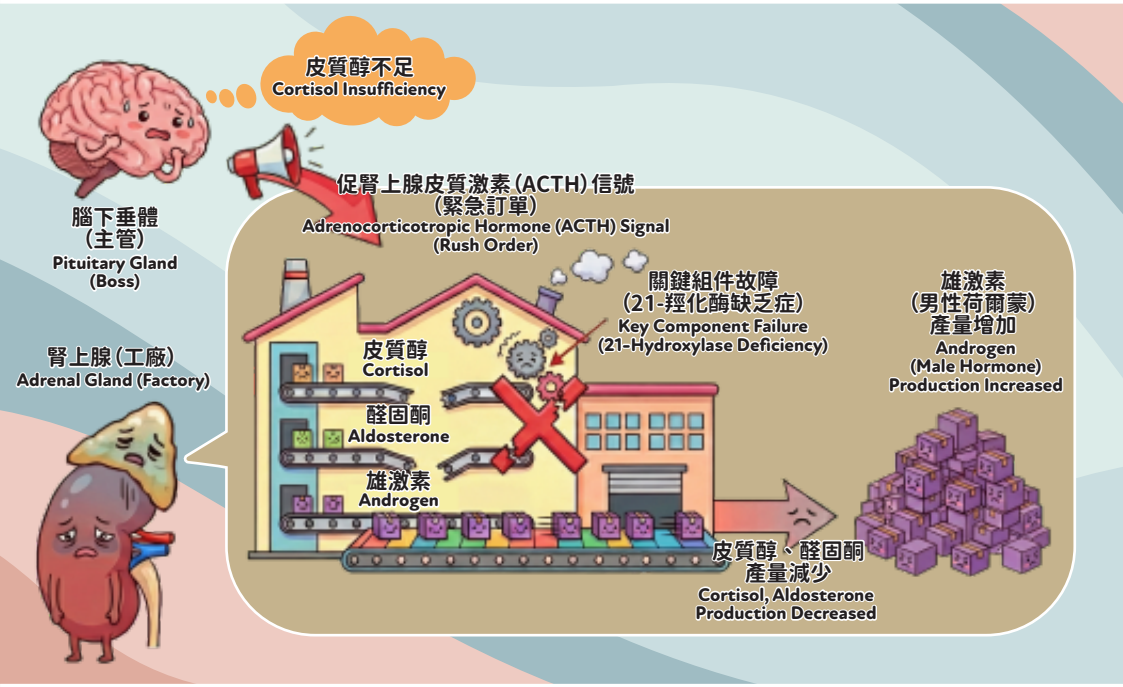
Although CAH is a lifelong condition, with early screening and diagnosis, standardized treatment, and regular follow-ups, most children with CAH can grow, study and work normally, and lead fulfilling lives. This practical handbook aims to provide families with a clear understanding of CAH. As each child's symptoms may differ, the following sections focus mainly on the most common type – 21-hydroxylase deficiency CAH.

第一部分 Part 1

什麼是CAH？ What is CAH?

腎上腺位於腎臟的上方，是一個生產重要激素（皮質醇、醛固酮、性激素）的「工廠」。CAH是因爲「工廠」裡某條特定生產線上的「關鍵零件」（即21-羥化酶）出了問題，導致機器故障，從而使生產線的產品（皮質醇和醛固酮）產量下降。當大腦的垂體感應到皮質醇不足時，會分泌大量的促腎上腺皮質激素（ACTH）。這種信號激素會不斷催促腎上腺更努力工作。然而，由於缺乏21-羥化酶，腎上腺無法生產更多的皮質醇。相反，這會迫使腎上腺製造更多雄激素（男性荷爾蒙）。

The adrenal glands sit on top of the kidneys and act as a “factory” that produces essential hormones (cortisol, aldosterone, sex hormones). CAH occurs because one “key component” in a specific production line (21-hydroxylase enzyme) is faulty, leading to decreased production of cortisol and aldosterone. The brain’s pituitary gland senses that there is not enough cortisol and pumps out extra ACTH—a signaling hormone that pushes the adrenal glands to work harder. This does not fix the cortisol shortage because the blocked enzyme step prevents normal cortisol production; instead, it overloads the pathway, causing the adrenals to churn out even more male hormones (androgens).



皮質醇 Cortisol

對於維持正常的血壓、血糖和能量至關重要。當身體生病或應對創傷時，皮質醇能幫助身體穩定血壓，血糖和電解質水平。

Essential for maintaining normal blood pressure, blood sugar and energy level. It also helps the body cope with illnesses and physical stress.

醛固酮 Aldosterone

有助於維持體內水和鈉的平衡。當醛固酮分泌不足時，水份和鈉會通過尿液流失，導致脫水和低鈉的情況。

Helps regulate the body's water and salt balance. When levels are too low, the body loses water and salt through urine, leading to dehydration and low blood salt level.

雄激素 Androgen (male hormone)

用於促進男性生殖器官的發育。

Promote the development of male sexual characteristics

由於激素失衡，CAH患兒可能出現以下的情況：
Because of these hormone imbalances, children with CAH experience the following:

失鹽脫水危象 Salt-wasting crisis

新生兒期寶寶可能出現餵食困難、嘔吐、體重下降，甚至發生低鈉、高鉀與脫水等狀況，嚴重時可危及生命。

Salt-wasting crisis: During the newborn period, babies may present with poor feeding, vomiting, weight loss, with low sodium and high potassium levels in blood, and dehydration. This can be life-threatening.

雄激素過多的症狀 Symptoms of excess androgen

女孩可能出現外生殖器男性化的情況(陰蒂異常增大、陰唇黏合，陰道與尿道開口部分閉合等外生殖器變化)。無論男女童也可能出現性早熟、骨齡加速成熟、高度異常增長等情形。若沒有接受適時治療，成年後的身高可能會受影響。

Girls may be born with male-like changes in their external genitalia (eg. clitoris may be bigger, the labia may be joined together and the openings of the vagina and urethra may be partly closed). Both boys and girls may have early pubertal onset, and have bones that age too quickly with a fast growth spurt during childhood. Without proper treatment, they may end up shorter as adults.

因21-羥化酶缺乏程度不同，患兒的臨床表現亦有所差異：
There are different severities of enzyme deficiency leading to a wide spectrum of clinical presentation:

經典失鹽型 Classic salt-wasting CAH

由於21-羥化酶嚴重缺乏，體內的皮質醇與醛固酮水平均明顯降低。患兒可能在嬰兒期出現失鹽脫水危象，需終身補充皮質醇和醛固酮。女童出生時亦可能出現外生殖器男性化的情況。

Severe enzyme deficiency - this leads to greatly reduced cortisol and aldosterone levels. Infants may experience salt-wasting crisis and require lifelong replacement of cortisol and aldosterone. Girls may also be born with male-like changes in their external genitalia.

單純男性化型 Simple virilizing CAH

由於21-羥化酶中度缺乏所致。患兒的皮質醇水平下降，雄激素明顯升高，但醛固酮缺乏程度較輕，因此多數患兒在嬰兒期不會出現失鹽危象。然而，由於體內雄激素過高，可能導致性早熟的表現。

Moderate enzyme deficiency - there is reduced cortisol production and excess androgen levels, but aldosterone deficiency is not as severe, so most infants do not experience a salt-wasting crisis. However, excess androgen may cause early puberty.

非經典型 Non-classical CAH

由於21-羥化酶的缺乏較輕度，症狀通常較輕微。大多在兒童期或青春期後出現過多雄激素的表現。例如女孩可能出現體毛增多、月經不規律或受孕困難；男孩則可能較早出現陰毛及青春痘等現象。

Mild enzyme deficiency - Symptoms are usually mild, with presentation in childhood related to excess androgen levels. For example, girls may have excess hair growth, irregular menstruation, or difficulty conceiving, while boys may have early pubic hair and acne.

第二部分 Part 2

新生兒篩查：生命的第一道保護網
Newborn screening – The First Line of Protection

針對CAH的新生兒篩查，已在香港、上海及全球許多地方實施。此項篩查通過採集嬰兒的幾滴血液，進行快速檢測，以判斷某些激素水平（尤其是17-OHP）是否異常升高。篩查結果呈陽性意味著嬰兒患有CAH的風險較高，需要作進一步確診性的檢查。醫療團隊將安排詳細的檢查，包括基因檢測，以確認或排除CAH的診斷。

Newborn screening for CAH is implemented in many places around the world, including Hong Kong, Shanghai etc. It involves a quick check of a few drops of your baby's blood to see if certain hormone levels, especially 17-OHP, are unusually high. A positive screening result means that the baby has a higher risk of CAH and needs further confirmatory tests. The medical team will arrange detailed investigations, including a genetic test, to confirm or rule out CAH.

在某些罕見情況或較輕微的病例中，新生兒篩查可能未能檢測出CAH。若出現疑似的症狀，醫療團隊將會進行血液及尿液檢測，並配合基因分析，以協助確診。

In rare occasions or in milder cases, CAH may not be detected through newborn screening. With symptoms suggestive of CAH, the medical team will use blood and urine tests, as well as genetic analysis, to make a definitive diagnosis.

第三部分 Part 3

分階段管理與治療：與孩子共同成長
Stage-specific Management: Growing Together with Your Child

一旦確診CAH，應立即開始治療。我們的治療目標是：「替代不足的，抑制過多的」，即是通過藥物補充缺乏的激素，抑制過高的雄激素，幫助孩子健康成長。

Once CAH is diagnosed, treatment should start immediately. Our treatment goal is: "replace what is lacking, suppress what is excessive". In other words, we use medications to replace the missing hormones while suppressing the high levels of androgen, thus allowing the child to grow normally.

治療的主要藥物包括 Main Medications

氫化可的松 Hydrocortisone

- 用於補充腎上腺無法製造的皮質醇。
  Replaces the cortisol hormone that the adrenal glands are unable to make.
- 醫生通常會以較高劑量的藥物，來減低促腎上腺皮質激素(ACTH)水平，減少過量雄激素的產生，並預防相關問題——例如女童的外生殖器男性化，或性早熟與骨齡快速成熟的現象。
  Doctors often use a higher dose to lower the ACTH levels, which helps cut down excess androgen and prevent problems like male-like genital changes in girls, or early puberty and fast-aging bones in both boys and girls.
- 由於氫化可的松比其他類固醇對生長的影響最小，因此尤其適用於兒童。
  Hydrocortisone has the least effect on growth suppression among other steroid options, making it the first choice for children.
- 每天需要服用3-4次。
  Taken 3-4 times a day.

氟氫可的松 Fludrocortisone

- 用於補充腎上腺無法製造的醛固酮。
  Replaces the aldosterone hormone that the adrenal glands are unable to make.
- 幫助維持體內電解質(鈉與鉀)的正常水平，並穩定血壓。
  Helps maintain normal electrolyte (sodium and potassium) levels and blood pressure.
- 每天需要服用1-2次。
  Usually taken 1-2 times a day.

大部分嬰兒另需額外補充食鹽
Most infants need extra salt supplements

生病劑量 Stress Dose Hydrocortisone

- 在孩子發熱、感染、進行手術時，必須增加氫化可的松的劑量(通常爲常規劑量的2-3倍)，以預防腎上腺危象(即低血壓/低血糖/休克)發生。建議您與醫療團隊共同制定清晰的「生病應對方案」，以確保能及時處理突發的情況。
  During stressful situations such as fever, infection, or surgery, hydrocortisone doses must be increased (usually 2-3 times the normal dose) to prevent an adrenal crisis (low blood pressure / low blood sugar / coma). Always discuss and make a clear “stress dose” plan with your medical team.

外生殖器矯形治療 Genital Surgery

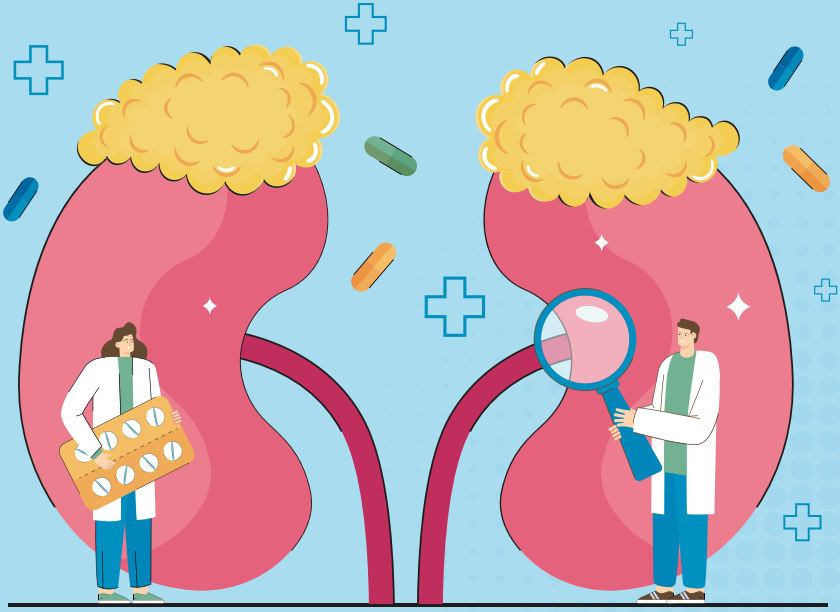
- 對外生殖器男性化(如陰蒂肥大、陰唇融合等)的女性患兒，待體內代謝狀況穩定後，患者與家長可與外科團隊共同討論手術的最佳時機。
  For female patients with male-like external genitalia (such as enlarged clitoris or labial fusion), patients and parents should consult the surgical team about the best timing for corrective procedures, once metabolic control is stabilized.

治療的監測 Monitoring during treatment

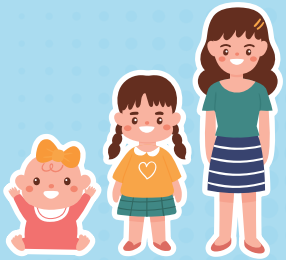
您的孩子需要定期的覆診，監測生長情況、青春期發育和血壓變化。此外，還需進行血液檢查以監測電解質和激素水平（包括17OHP及雄激素），並需定期進行骨齡評估，確保治療效果正常。

Your child will need regular follow-up visits to monitor growth, pubertal development, and blood pressure changes. In addition, blood tests are required to check electrolyte and hormone levels (including 17OHP and androgen). Bone age assessment should also be performed regularly to ensure that treatment remains effective and appropriate.

- 治療初期需密切監測臨床情況，每2周至1個月監測一次
  Close monitoring according to clinical condition in the early treatment phase: every 2-4 weeks
- 情況穩定後，約3~4個月監測一次
  Once stable: around every 3 - 4 months
- 醫療團隊會根據臨床需要，調整監測的頻率
  The frequency of monitoring may be adjusted as per clinical need



不同成長階段的主要照護重點
Key care points in different stages of life



嬰兒/幼兒期 Infant/Toddler period

- 重點在於預防體內鹽分與水分的過度流失。醫生將密切監測幼兒的生長、血壓、電解質平衡及激素水平。父母是孩子最重要的守護者，應學會識別食慾不振、嘔吐、脫水、嗜睡、甚至昏迷等的徵兆，並及時就醫。

  The main focus is to prevent excessive loss of salt and water from the body. Doctors will closely monitor your child's growth, blood pressure, electrolyte balance, and hormone levels. Parents are the child's most important caregivers and should learn to recognize warning signs such as poor appetite, vomiting, dehydration, drowsiness, or even coma, and seek medical attention promptly.

兒童期 School Age

- 關注正常生長和學習需要，並與學校老師保持溝通(如日常用藥和應急處理)，鼓勵孩子正常參與體育活動。

  Focus on supporting normal growth and learning needs. Maintain good communication with teachers regarding the child's daily medication and emergency management. Encourage participation in regular physical activities to promote a healthy and balanced lifestyle.

青春期 Adolescence

- 關注青春期進程、骨齡和身高變化，並提供心理支持，幫助孩子理解並管理自己的疾病。青春期的荷爾蒙變化可能令病情管理變得較不穩定。因此醫療團隊會安排更頻密的檢查，並依需要調整用藥劑量。

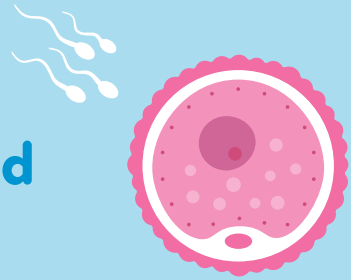
  Pay attention to the progression of puberty, bone age, and height changes, while also providing psychological support to help the child understand and manage their condition. Hormonal changes during this stage can make disease management trickier. Therefore, the medical team will arrange more frequent follow-up visits and adjust medication dosages as needed.

已完成生長的青少年及成年患者，可考慮將氫化可的松調整爲更長效的類固醇，如潑尼松龍 (Prednisolone) 或地塞米松 (Dexamethasone)，以減少每日服藥次數。

For adolescents and adults who have completed growth, the doctor may consider switching hydrocortisone to a longer-acting steroid, such as prednisolone or dexamethasone, to reduce the number of daily doses.

第四部分 Part 4

生殖健康與終身監測
Reproductive health and Lifelong Follow-up



生殖健康 Reproductive health

女性患者 Female Patients

- 雄激素過高可能干擾下丘腦-垂體和卵巢的功能，導致月經不規則、多囊卵巢症、甚至不孕。長期穩定的激素控制是維持生育能力的基礎。計劃妊娠前，您應該進行孕前諮詢，並在多學科團隊(內分泌、婦科、遺傳)共同指導下制定診療方案。

  High levels of androgens can affect the hypothalamic-pituitary-ovarian axis, causing irregular menstruation, polycystic ovary syndrome, or even infertility. Maintaining good hormone control over long term is the key to preserving fertility. Before planning a pregnancy, you should consult a multidisciplinary team (endocrinology, gynaecology, and genetics) for individualized care.

男性患者 Male Patients

- 部分患者可能出現睾丸內腎上腺殘餘瘤，影響精子功能，所以需定期進行睾丸超聲波檢查。

  Some may develop testicular adrenal rest tumors (TART), which can potentially affect sperm production. Regular testicular ultrasound examination is recommended.

終身監測與成年照護 Lifelong monitoring and transition to adult care

當孩子長大成人後，將會轉由成人內分泌科醫生繼續跟進與定期覆診。這包括定期監測身體的代謝狀況(例如血壓、血糖與骨骼健康)、激素水平，以及生育能力的評估。適量的運動有助於預防長期健康問題，並維持整體身心健康。

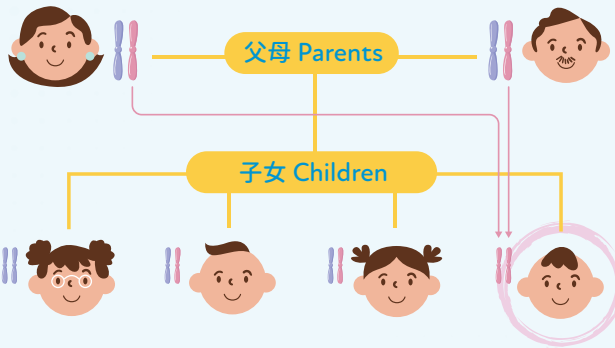
As your child enters adulthood, they will transition to lifelong monitoring with an adult endocrinologist. This includes regular checks for metabolic health (like blood pressure, blood sugar, and bone health), hormone balance, and reproductive function. Staying active helps prevent long-term health issues and promotes overall physical and mental well-being.

第五部分 Part 5
遺傳諮詢 Genetic Counseling

CAH是一種常染色體隱性遺傳病，當患者遺傳了兩個病變的CYP21A2基因（一個來自父親，一個來自母親），身體就不能製造所需的酶而會發病。如果一對基因中只有一個病變基因，他們則被稱爲基因攜帶者，不會有任何病徵。

CAH is an autosomal recessive disorder. A child develops the disease only when they inherit two abnormal CYP21A2 genes — one from each parent. Individuals carrying only one abnormal gene are called carriers and show no symptoms.

假如父母都是CAH的基因攜帶者，他們每次懷孕的孩子（不分男女）會有：
If both parents are carriers of a CAH gene mutation, for each pregnancy (regardless of the child's sex), there is a:



- 25%（即四分之一）的機會遺傳了兩個正常的基因，即沒有CAH。
  25% (1 in 4) chance the child inherits two normal genes and does not have CAH.
- 50%（即二分之一）的機會遺傳了一個正常基因和一個病變基因，即與父母一樣屬於基因攜帶者。
  50% (1 in 2) chance the child inherits one normal and one abnormal gene, making them a carrier like the parents.
- 25%（即四分之一）的機會遺傳了兩個病變基因，成爲CAH患者。
  25% (1 in 4) chance the child inherits two abnormal genes and is affected by CAH.

CAH患者的孩子患病的概率與患者配偶是否攜帶CAH病變基因有關。因此，CAH患者及其家人在計劃生育時，建議進行遺傳諮詢，了解後代風險及產前診斷等事宜。

For patients with CAH, the risk of the child being affected depends on whether the partner carries the abnormal gene. Therefore, genetic counseling is recommended during family planning to understand the risk to future children and to consider prenatal genetic diagnosis.

第六部分 Part 6

妊娠期的診斷與治療
Diagnosis and treatment during pregnancy

如果您有一個患有CAH的孩子，可以考慮在孕10-11週進行絨毛膜取樣或孕12-14週進行羊膜穿刺取樣，進行妊娠期診斷，評估胎兒是否患病。這兩種手術均會輕微增加流產風險。此外，在某些具備高度專門設施的產前診斷中心，可考慮進行無創產前基因檢測 (NIPT)，即從母體血液中分離胎兒的DNA，去篩查胎兒是否患病。

If you have one child with CAH, prenatal diagnosis can be performed — chorionic villus sampling at 10–11 weeks or amniocentesis at 12–14 weeks — to check whether the fetus has CAH. Both procedures carry a small risk of miscarriage.

In addition, at highly specialized prenatal diagnostic centres, non-invasive prenatal testing (NIPT) may be considered. This test uses a sample of the mother's blood to look at the baby's DNA and check if the baby may have CAH.

若胎兒確診患有CAH，醫療團隊可能會在大概在孕6週給予類固醇藥物(如地塞米松)，以降低女嬰外生殖器出現男性化變化的風險與嚴重程度。由於生殖器官在懷孕初期已經開始發育，此治療必須及早開始才能發揮效用。然而，這項治療方法在醫學上仍存在爭議，並可能給母親帶來不良的身體變化，如妊娠糖尿病、高血壓和體重增加等。因此我們建議您與婦產科醫療團隊仔細評估治療的利弊。

From around the 6th week of pregnancy, the medical team may prescribe a steroid (dexamethasone) to reduce the risk and severity of male-like changes to a baby girl's genitals if the fetus has CAH. This treatment must start early because genital development happens very early in pregnancy. However, this practice remains controversial and may carry side effects for the mother, such as gestational diabetes, high blood pressure, and weight gain. The potential benefits and risks should be carefully discussed with the obstetrics team.

第七部分 Part 7

CAH治療的新進展
New Horizons in the Treatment of CAH

近年來，研究人員在CAH的治療方面取得了以下突破性進展：
In recent years, researchers have made strides in the management of CAH:

緩釋型氫化可的松製劑
Modified-release hydrocortisone formulations

緩釋製劑能更好地模擬人體皮質醇分泌的生理節律，達致更好的控制。
Slow-release formula that better replicates the natural cortisol production rhythm in the body, leading to improved control

靶向抑制促腎上腺皮質激素(ACTH)產生
Targeting ACTH production

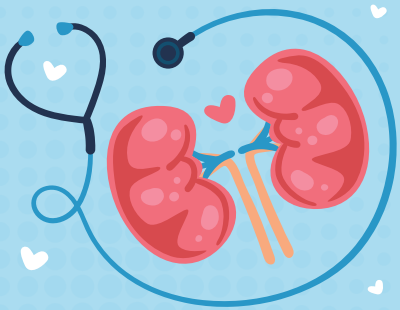
有助於降低過量雄激素水平，並減少所需藥物的劑量。
Helps reduce excess androgen levels and lower the required medication dose

基因療法(現階段尚屬實驗性)
Gene therapy (still experimental at this stage)

透過病毒載體將正常的*CYP21A2*基因送入體內，幫助腎上腺恢復製造皮質醇與醛固酮的功能。
Gene therapy for CAH uses a viral vector to deliver functional copies of the *CYP21A2* gene into the body, enabling the adrenal glands to properly make cortisol and aldosterone.

這些嶄新的醫療進展，可以爲CAH患者帶來更精準、個人化的治療，也可以改善患者長期的健康。

These innovations bring hope for improved, more personalized care and better long-term outcomes for people with CAH.



給家長的重要信息
Key messages for parents

CAH的管理猶如一場「馬拉松」，而非「短跑」。它需要醫療團隊、患者和家庭彼此合作，才能支援孩子健康、快樂地成長。

Taking care of CAH is like a “marathon”, not a “sprint”. It takes teamwork between the medical team, your child, and the family to help your kid grow up strong and happy.

- 請信任您的醫療團隊，並與他們保持良好的溝通與穩定的關係。
  Trust your medical team and chat with them often to stay on the same page.
- 持續記錄孩子的用藥情況、身高體重變化及任何不適，並於覆診時帶給醫生作參考。
  Jot down your child’s medication, growth updates, and any discomfort, and share this information at check-ups.
- 隨著年齡增長，逐步讓孩子了解自己的疾病和用藥的安排。
  As your little one grows, slowly explain their condition and medicines in simple words.
- 留意孩子的心理狀況；心理健康與身體健康同樣重要，都需要細心關懷和支持。
  Watch their emotional well-being too; mental health is just as important as physical health and needs your support .
- 如果家長發現任何的情況，或接觸到與疾病相關的新資訊，歡迎主動與醫療團隊分享和討論。
  If you notice anything new or come across helpful information about the condition, feel free to reach out to the medical team.

持之以恆的照顧，能讓孩子在健康、充滿愛的環境中快樂成長。我們之間的互信和配合，對治療效果非常重要。讓我們一起攜手，爲孩子的健康勇往直前！

Love and care help your child bloom and thrive in a healthy, caring environment. Our trust and teamwork make all the difference in treatment. Let’s team up and keep going for your child’s bright future!

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