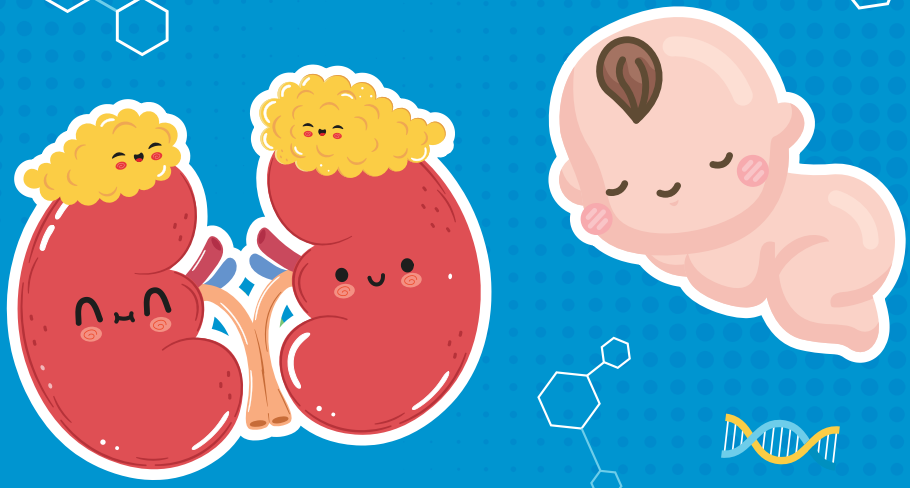
香港兒童醫院
Hong Kong Children's Hospital

认识先天性肾上腺增生症 (CAH)
Understanding Congenital Adrenal Hyperplasia (CAH)



从新生儿筛查到健康成长
From Newborn Screening to Lifelong Care

家庭实用手册
A Practical Guide for Families

前言 Preface

每一位父母都希望孩子健康成长，而新生儿筛查正默默守护着这份起点。其中，针对先天性肾上腺增生症（Congenital Adrenal Hyperplasia - 简称CAH）的筛查，正是这些项目的关键之一。

先天性肾上腺增生症（CAH）是一组因肾上腺制造激素功能出现障碍而引起的疾病。CAH是一种常染色体隐性遗传的疾病 — 也就是说，只有同时携带两个相关变异基因的情况下，才会发病。

激素是人体重要的讯息传导物质。当CAH患者肾上腺激素分泌异常时，便会出现不同的临床表征与症状。 CAH可分为多种类型，其中最常见的是21-羟化酶缺乏症。全球平均大约每一万至两万人中，便有一人患病。

虽然CAH是终身疾病，但透过早期筛查和诊断、配合规范治疗和定期覆诊，绝大多数CAH的孩子都能正常生长发育、学习、工作，并享受美满的人生。本家庭实用手册旨在为您提供清晰实用的CAH资讯。由于每个患有CAH的儿童的临床表现可能有所不同，以下内容将以最常见的21-羟化酶缺乏症为例进行说明。

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.

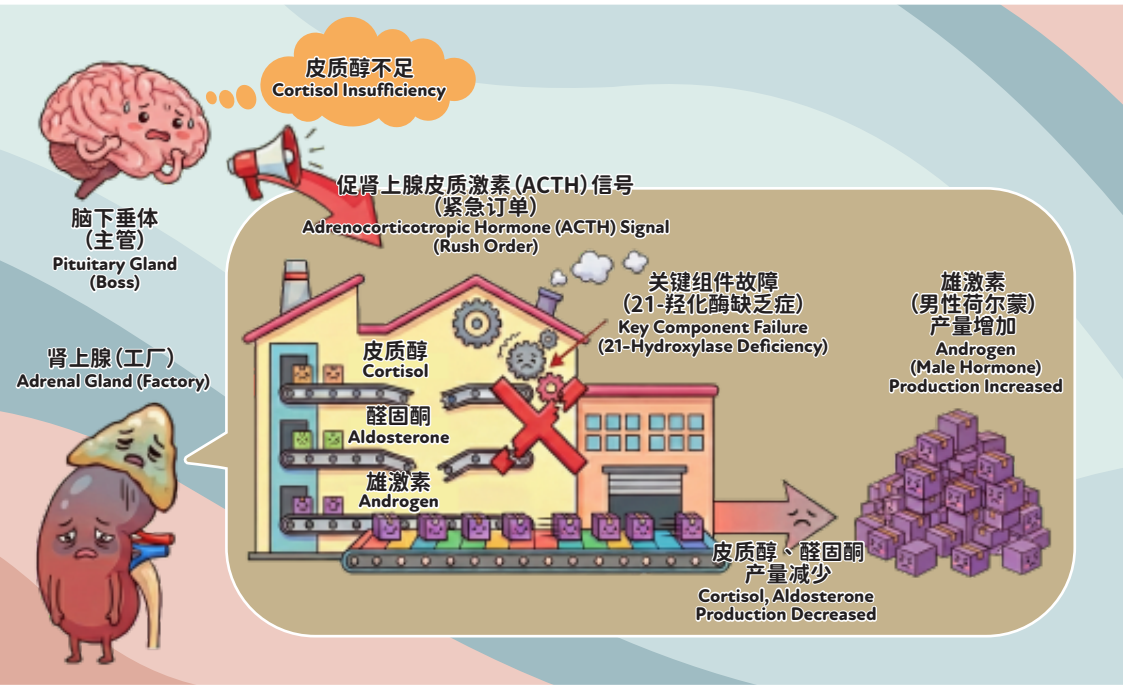
Although CAH is a lifelong condition, with early screening and diagnosis, standardized treatment, and regular follow-ups, most children with CAH can grow, study and work normally, and lead fulfilling lives. This practical handbook aims to provide families with a clear understanding of CAH. As each child's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

第一部分 Part 1

什么是CAH？ What is CAH?

肾上腺位于肾脏的上方，是一个生产重要激素（皮质醇、醛固酮、性激素）的「工厂」。 CAH是因为「工厂」里某条特定生产线上的「关键零件」（即21-羟化酶）出了问题，导致机器故障，从而使生产线的产品（皮质醇和醛固酮）产量下降。当大脑的垂体感应到皮质醇不足时，会分泌大量的促肾上腺皮质激素（ACTH）。这种信号激素会不断催促肾上腺更努力工作。然而，由于缺乏21-羟化酶，肾上腺无法生产更多的皮质醇。相反，这会迫使肾上腺制造更多雄激素（男性荷尔蒙）。

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).



↓ **皮质醇 Cortisol**

对于维持正常的血压、血糖和能量至关重要。当身体生病或应对创伤时，皮质醇能帮助身体稳定血压，血糖和电解质水平。

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**

有助于维持体内水和钠的平衡。当醛固酮分泌不足时，水份和钠会通过尿液流失，导致脱水和低钠的情况。

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)**

用于促进男性生殖器官的发育。

Promote the development of male sexual characteristics

由于激素失衡，CAH患儿可能出现以下的情况：
Because of these hormone imbalances, children with CAH experience the following:

失盐脱水危象 Salt-wasting crisis

新生儿期宝宝可能出现喂食困难、呕吐、体重下降，甚至发生低钠、高钾与脱水等状况，严重时可危及生命。

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.

雄激素过多的症状 Symptoms of excess androgen

女孩可能出现外生殖器男性化的情况(阴蒂异常增大、阴唇黏合，阴道与尿道开口部分闭合等外生殖器变化)。无论男女童也可能出现性早熟、骨龄加速成熟、高度异常增长等情形。若没有接受适时治疗，成年后的身高可能会受影响。

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-羟化酶缺乏程度不同，患儿的临床表现亦有所差异：

There are different severities of enzyme deficiency leading to a wide spectrum of clinical presentation:

经典失盐型 Classic salt-wasting CAH

由于21-羟化酶严重缺乏，体内的皮质醇与醛固酮水平均明显降低。患儿可能在婴儿期出现失盐脱水危象，需终身补充皮质醇和醛固酮。女童出生时亦可能出现外生殖器男性化的情况。

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.

单纯男性化型 Simple virilizing CAH

由于21-羟化酶中度缺乏所致。患儿的皮质醇水平下降，雄激素明显升高，但醛固酮缺乏程度较轻，因此多数患儿在婴儿期不会出现失盐危象。然而，由于体内雄激素过高，可能导致性早熟的表现。

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.

非经典型 Non-classical CAH

由于21-羟化酶的缺乏较轻度，症状通常较轻微。大多在儿童期或青春期后出现过多雄激素的表现。例如女孩可能出现体毛增多、月经不规律或受孕困难；男孩则可能较早出现阴毛及青春痘等现象。

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

新生儿筛查：生命的第一道保护网
Newborn screening – The First Line of Protection

针对CAH的新生儿筛查，已在香港、上海及全球许多地方实施。此项筛查通过采集婴儿的几滴血液，进行快速检测，以判断某些激素水平（尤其是17-OHP）是否异常升高。筛查结果呈阳性意味着婴儿患有CAH的风险较高，需要作进一步确诊性的检查。医疗团队将安排详细的检查，包括基因检测，以确认或排除CAH的诊断。

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.

在某些罕见情况或较轻微的病例中，新生儿筛查可能未能检测出CAH。若出现疑似的症状，医疗团队将会进行血液及尿液检测，并配合基因分析，以协助确诊。

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

分阶段管理与治疗：与孩子共同成长
Stage-specific Management: Growing Together with Your Child

一旦确诊CAH，应立即开始治疗。我们的治疗目标是：「替代不足的，抑制过多的」，即是通过药物补充缺乏的激素，抑制过高的雄激素，帮助孩子健康成长。

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.

治疗的主要药物包括 Main Medications

氢化可的松 Hydrocortisone

- 用于补充肾上腺无法制造的皮质醇。
  Replaces the cortisol hormone that the adrenal glands are unable to make.
- 医生通常会以较高剂量的药物，来减低促肾上腺皮质激素(ACTH)水平，减少过量雄激素的产生，并预防相关问题——例如女童的外生殖器男性化，或性早熟与骨龄快速成熟的现象。
  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.
- 由于氢化可的松比其他类固醇对生长的影响最小，因此尤其适用于儿童。
  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.

氟氢可的松 Fludrocortisone

- 用于补充肾上腺无法制造的醛固酮。
  Replaces the aldosterone hormone that the adrenal glands are unable to make.
- 帮助维持体内电解质(钠与钾)的正常水平，并稳定血压。
  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.

大部分婴儿另需额外补充食盐
Most infants need extra salt supplements

生病剂量 Stress Dose Hydrocortisone

- 在孩子发热、感染、进行手术时，必须增加氢化可的松的剂量(通常为常规剂量的2-3倍)，以预防肾上腺危象(即低血压/低血糖/休克)发生。建议您与医疗团队共同制定清晰的「生病应对方案」，以确保能及时处理突发的情况。
  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.

外生殖器矫形治疗 Genital Surgery

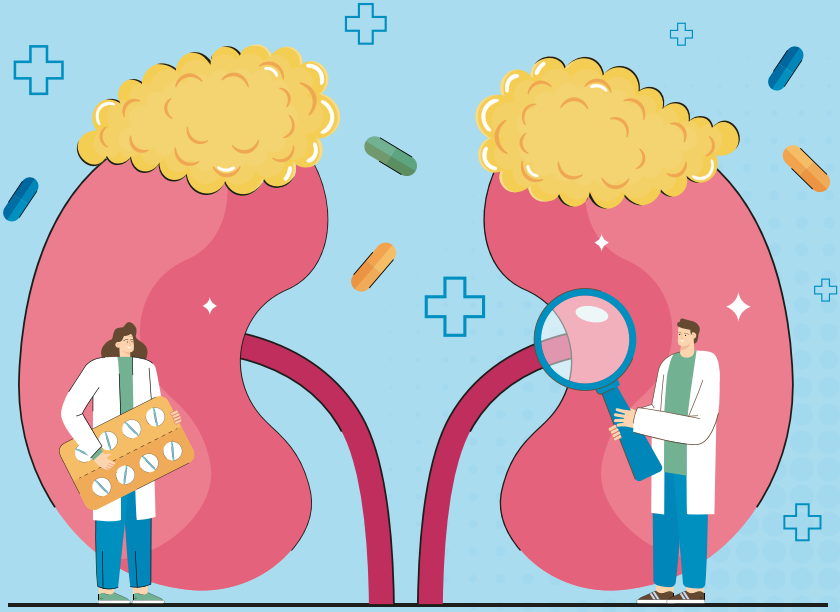
- 对外生殖器男性化(如阴蒂肥大、阴唇融合等)的女性患儿，待体内代谢状况稳定后，患者与家长可与外科团队共同讨论手术的最佳时机。
  For female patients with male-like external genitalia (such as enlarged clitoris or labial fusion), patients and parents should consult the surgical team about the best timing for corrective procedures, once metabolic control is stabilized.

治疗的监测 Monitoring during treatment

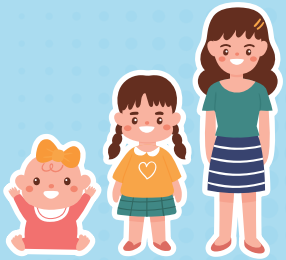
您的孩子需要定期的覆诊，监测生长情况、青春期发育和血压变化。此外，还需进行血液检查以监测电解质和激素水平（包括17OHP及雄激素），并需定期进行骨龄评估，确保治疗效果正常。

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.

- 治疗初期需密切监测临床情况，每2周至1个月监测一次
  Close monitoring according to clinical condition in the early treatment phase: every 2-4 weeks
- 情况稳定后，约3~4个月监测一次
  Once stable: around every 3 - 4 months
- 医疗团队会根据临床需要，调整监测的频率
  The frequency of monitoring may be adjusted as per clinical need



不同成长阶段的主要照护重点
Key care points in different stages of life



婴儿/幼儿期 Infant/Toddler period

- 重点在于预防体内盐分与水分的过度流失。医生将密切监测幼儿的生长、血压、电解质平衡及激素水平。父母是孩子最重要的守护者，应学会识别食欲不振、呕吐、脱水、嗜睡、甚至昏迷等的征兆，并及时就医。

  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.

儿童期 School Age

- 关注正常生长和学习需要，并与学校老师保持沟通(如日常用药和应急处理)，鼓励孩子正常参与体育活动。

  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

- 关注青春期进程、骨龄和身高变化，并提供心理支持，帮助孩子理解并管理自己的疾病。青春期的荷尔蒙变化可能令病情管理变得较不稳定。因此医疗团队会安排更频密的检查，并依需要调整用药剂量。

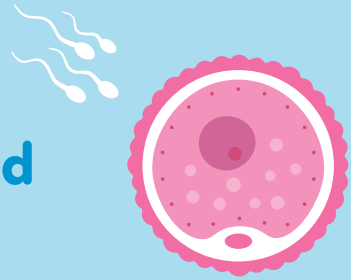
  Pay attention to the progression of puberty, bone age, and height changes, while also providing psychological support to help the child understand and manage their condition. Hormonal changes during this stage can make disease management trickier. Therefore, the medical team will arrange more frequent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

已完成生长的青少年及成年患者，可考虑将氢化可的松调整为更长效的类固醇，如泼尼松龙 (Prednisolone) 或地塞米松 (Dexamethasone)，以减少每日服药次数。

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

生殖健康与终身监测
Reproductive health and Lifelong Follow-up



生殖健康 Reproductive health

女性患者 Female Patients

- 雄激素过高可能干扰下丘脑-垂体和卵巢的功能，导致月经不规则、多囊卵巢症、甚至不孕。长期稳定的激素控制是维持生育能力的基础。计划妊娠前，您应该进行孕前咨询，并在多学科团队（内分泌、妇科、遗传）共同指导下制定诊疗方案。

  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

- 部分患者可能出现睾丸内肾上腺残余瘤，影响精子功能，所以需定期进行睾丸超声波检查。

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

终身监测与成年照护 Lifelong monitoring and transition to adult care

当孩子长大成人后，将会转由成人内分泌科医生继续跟进与定期覆诊。这包括定期监测身体的代谢状况（例如血压、血糖与骨骼健康）、激素水平，以及生育能力的评估。适量的运动有助于预防长期健康问题，并维持整体身心健康。

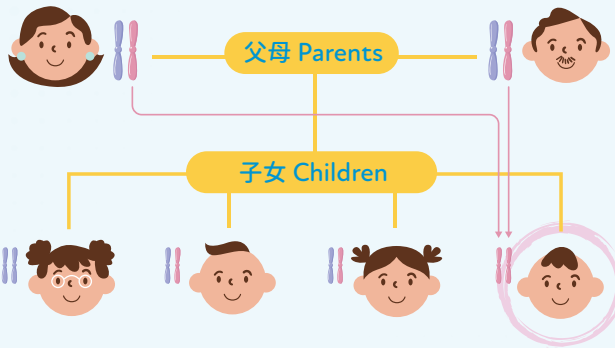
As your child enters adulthood, they will transition to lifelong monitoring with an adult endocrinologist. This includes regular checks for metabolic health (like blood pressure, blood sugar, and bone health), hormone balance, and reproductive function. Staying active helps prevent long-term health issues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

第五部分 Part 5
遗传咨询 Genetic Counseling

CAH是一种常染色体隐性遗传病，当患者遗传了两个病变的CYP21A2基因（一个来自父亲，一个来自母亲），身体就不能制造所需的酶而会发病。如果一对基因中只有一个病变基因，他们则被称为基因携带者，不会有任何病征。

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的基因携带者，他们每次怀孕的孩子（不分男女）会有：
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）的机会遗传了两个正常的基因，即没有CAH。
  25% (1 in 4) chance the child inherits two normal genes and does not have CAH.
- 50%（即二分之一）的机会遗传了一个正常基因和一个病变基因，即与父母一样属于基因携带者。
  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）的机会遗传了两个病变基因，成为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与患者配偶是否携带CAH病变基因有关。因此，CAH患者及其家人在计划生育时，建议进行遗传咨询，了解后代风险及产前诊断等事宜。

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

妊娠期的诊断与治疗
Diagnosis and treatment during pregnancy

如果您有一个患有CAH的孩子，可以考虑在孕10-11周进行绒毛膜取样或孕12-14周进行羊膜穿刺取样，进行妊娠期诊断，评估胎儿是否患病。这两种手术均会轻微增加流产风险。此外，在某些具备高度专门设施的产前诊断中心，可考虑进行无创产前基因检测 (NIPT)，即从母体血液中分离胎儿的DNA，去筛查胎儿是否患病。

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.

若胎儿确诊患有CAH，医疗团队可能会在大概在孕6周给予类固醇药物（如地塞米松），以降低女婴外生殖器出现男性化变化的风险与严重程度。由于生殖器官在怀孕初期已经开始发育，此治疗必须及早开始才能发挥效用。然而，这项治疗方法在医学上仍存在争议，并可能给母亲带来不良的身体变化，如妊娠糖尿病、高血压和体重增加等。因此我们建议您与妇产科医疗团队仔细评估治疗的利弊。

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治疗的新进展
New Horizons in the Treatment of CAH

近年来，研究人员在CAH的治疗方面取得了以下突破性进展：
In recent years, researchers have made strides in the management of CAH:

缓释型氢化可的松制剂
Modified-release hydrocortisone formulations

缓释制剂能更好地模拟人体皮质醇分泌的生理节律，达致更好的控制。
Slow-release formula that better replicates the natural cortisol production rhythm in the body, leading to improved control

靶向抑制促肾上腺皮质激素(ACTH)产生
Targeting ACTH production

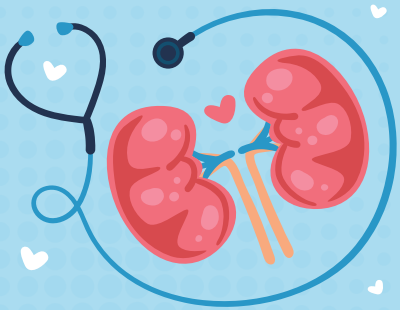
有助于降低过量雄激素水平，并减少所需药物的剂量。
Helps reduce excess androgen levels and lower the required medication dose

基因疗法(现阶段尚属实验性)
Gene therapy (still experimental at this stage)

透过病毒载体将正常的CYP21A2基因送入体内，帮助肾上腺恢复制造皮质醇与醛固酮的功能。
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.

这些崭新的医疗进展，可以为CAH患者带来更精准、个人化的治疗，也可以改善患者长期的健康。

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



给家长的重要信息
Key messages for parents

CAH的管理犹如一场「马拉松」，而非「短跑」。它需要医疗团队、患者和家庭彼此合作，才能支援孩子健康、快乐地成长。

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.

- 请信任您的医疗团队，并与他们保持良好的沟通与稳定的关系。
  Trust your medical team and chat with them often to stay on the same page.
- 持续记录孩子的用药情况、身高体重变化及任何不适，并于覆诊时带给医生作参考。
  Jot down your child’s medication, growth updates, and any discomfort, and share this information at check-ups.
- 随着年龄增长，逐步让孩子了解自己的疾病和用药的安排。
  As your little one grows, slowly explain their condition and medicines in simple words.
- 留意孩子的心理状况；心理健康与身体健康同样重要，都需要细心关怀和支持。
  Watch their emotional well-being too; mental health is just as important as physical health and needs your support .
- 如果家长发现任何的情况，或接触到与疾病相关的新资讯，欢迎主动与医疗团队分享和讨论。
  If you notice anything new or come across helpful information about the condition, feel free to reach out to the medical team.

持之以恒的照顾，能让孩子在健康、充满爱的环境中快乐成长。我们之间的互信和配合，对治疗效果非常重要。让我们一起携手，为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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