香港兒童醫院
Hong Kong Children's Hospital

醫學遺傳科遺傳篩選診所
新生嬰兒篩選服務

先天性甲狀腺功能不足



醫院管理局
HOSPITAL AUTHORITY

甲狀腺

甲狀腺是人體的主要內分泌器官之一。它所分泌的甲狀腺素主要功能是促進新陳代謝，而新生嬰兒需要足夠的甲狀腺素刺激生長和腦部發育。

主要成因

- 甲狀腺發育不全
- 甲狀腺位置偏差
- 甲狀腺本身未能正常地製造甲狀腺素

病徵

初生時病徵**不明顯**，直至數月或數年後才出現病徵。

由於嬰兒腦部發育在出生後兩年內非常倚賴甲狀腺素，如未能及早確診和治療，會引致身型矮小及智力發展遲緩。

血液檢驗

醫院管理局轄下醫學遺傳科的遺傳篩選診所現正為所有公立醫院出生的新生嬰兒安排血液檢驗，測試嬰兒的甲狀腺功能。

首次測試主要是抽取新生嬰兒的臍帶血，提供檢驗服務。檢視測試結果之後，如有需要，醫護人員安排嬰兒抽取血液樣本，作進一步檢驗，以確定其甲狀腺功能。如嬰兒最終確定患有先天性甲狀腺功能不足，便可及早接受治療及跟進。

治療

- 患者有可能需要終生服藥
- 每日補充適量的甲狀腺素
- 定期覆診

如能及早治療，按時服藥及覆診，嬰兒的發育與智力發展將不會受到影響

常見問題

先天性甲狀腺功能不足的主要成因是什麼？

主要原因有三：
一. 甲狀腺發育不全
二. 甲狀腺位置偏差
三. 甲狀腺本身未能正常地製造甲狀腺素

先天性甲狀腺功能不足是不是遺傳病？

現時醫學證據只能顯示先天性甲狀腺功能不足有遺傳性，但仍需要更多研究結果，才能準確評估其遺傳風險。

如果患有先天性甲狀腺功能不足，需要吃藥多久？

用藥安排往往依據患病原因及需要。患者需遵從醫生指示，有可能需要終生服藥。

先天性甲狀腺功能不足可以根治嗎？

雖然先天性甲狀腺功能不足或未能完全根治，但只要遵從醫生指示，按時服藥及覆診，嬰兒的發育與智力發展將不會受到影響。

如果覆驗時促甲狀腺激素已經回落，是否代表甲狀腺功能正常？會否再變不正常？

促甲狀腺激素回落後，一般不會再變不正常。如果對篩選正常結果有擔心，家長可再諮詢家庭醫生或兒科醫生。

促甲狀腺激素升高是否因為媽媽懷孕時吃錯東西？

除了媽媽曾於懷孕期間不按醫生指示服用抗甲狀腺藥外，促甲狀腺激素升高通常與媽媽的飲食習慣無關。

為什麼剖腹生產的嬰兒也會受到壓力？

無論是順產或剖腹生產，嬰兒出生時，生產過程及環境轉變，例如接觸冷空氣，都可以是壓力的來源，導致促甲狀腺激素升高。

* 此單張未能完全盡錄所有問題，如有其他問題，請諮詢家庭醫生或兒科醫生。

如果有其他查詢，歡迎您聯絡我們。

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香港兒童醫院
Hong Kong Children's Hospital

Genetic Screening Clinic
Department of Clinical Genetics

Newborn Screening Programme

Congenital Hypothyroidism



Thyroid Gland

The thyroid gland is one of the major endocrine organs in our body. It secretes a hormone called thyroxine, which is vital for our body metabolism, and is required for growth and mental development.

Causes

- Absence or underdevelopment of thyroid gland
- Thyroid gland is not in the right position
- Impaired thyroxine production from the thyroid gland

Symptoms

No obvious **early symptoms** until months or years later. Thyroxine is crucial for normal mental development in the first two years of life. If undiagnosed and untreated, short stature and irreversible developmental delay will be resulted.

Blood Testing

The Genetic Screening Clinic of Department of Clinical Genetics, Hospital Authority provides newborn screening for all babies born in public hospitals.

The first screening test uses baby's umbilical cord blood for thyroid function testing. Baby might be required to have re-testing to confirm the thyroid function if the screening result is abnormal. For those with confirmed congenital hypothyroidism, early management and follow-up can then be provided.

Treatment

- Affected baby might require lifelong treatment
- Daily thyroxine supplement
- Regular follow-up

With early treatment, good drug compliance and regular follow-up, baby's growth and mental development will not be affected.

Common Questions & Answers

What are the main causes of Congenital Hypothyroidism?

There are 3 main causes:
i. Absence or underdevelopment of thyroid gland
ii. Thyroid gland is not in the right position
iii. Impaired thyroxine production from the thyroid gland

Is Congenital Hypothyroidism a genetic problem?

It is now evident that genetic cause is contributable to Congenital Hypothyroidism. More research is, however, necessary to substantiate the risk to the offspring.

How long does the baby need to take medications if the baby has Congenital Hypothyroidism?

The duration of treatment depends on the cause of Congenital Hypothyroidism and the baby's condition. Diseased babies need to follow doctor's instruction and some might need to take lifelong medication.

Is Congenital Hypothyroidism curable?

Congenital hypothyroidism might not be curable but is treatable. As long as your baby follows doctor's instruction in taking medication and having regular follow up, your baby's growth and mental development will not be affected.

After the raised Thyroid Stimulating Hormone (TSH) has become normal, does it mean the thyroid is functioning properly? Would TSH become abnormal again?

Generally, it is unusual for TSH to raise again after becoming normal. You may consult your family doctor or paediatrician if you have worry about the baby's result and condition.

Is raised Thyroid Stimulating Hormone (TSH) related to any improper food consumption of mother during pregnancy?

Apart from poor adherence to anti-thyroid drugs as prescribed by doctor, raised TSH is seldomly related to mother's dietary habits during pregnancy.

Why is there stress when baby is delivered by Caesarean Section?

Baby is usually born with stress, whether through normal delivery or by caesarean section. There are many factors, such as the process of labour and cold external environment, which may affect the TSH level.

* This pamphlet does not cover all the questions. You may consult your family doctor or paediatrician if you have further enquiries.

For further enquires, you may contact us.

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