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

新生嬰兒篩選服務資料小冊子



醫院管理局 HOSPITAL AUTHORITY

醫學遺傳科遺傳篩選診所
提供新生嬰兒篩選服務

服務對象
香港公立醫院出生的嬰兒



篩查疾病
1 葡萄糖六磷酸去氫酵素缺乏症（G6PD缺乏症）
2 先天性甲狀腺功能不足



原因


- 兩者在早期均沒有明顯病徵
- 照顧患有葡萄糖六磷酸去氫酵素缺乏症（G6PD缺乏症）的嬰兒時，有需要額外注意的事項，以預防相關症狀發生
- 如能早發現嬰兒患有先天性甲狀腺功能不足，及早治療，可以預防身體及智力發展遲緩

費用
新生嬰兒臍帶血篩選測試為免費，及後覆驗或檢查需按照醫院管理局門診服務收費。

方法
經同意後，抽取新生嬰兒臍帶血作測試



篩查結果通知
如篩查結果**正常**，家長將不會收到任何通知

如篩查結果**不正常**，家長將會於**兩星期**內收到通知：

1 **嬰兒確診葡萄糖六磷酸去氫酵素缺乏症（G6PD缺乏症）**
遺傳篩選診所或出生醫院會通知嬰兒的父母，提供適當的輔導。

2 **先天性甲狀腺功能不足**
- ★ 嬰兒懷疑患有先天性甲狀腺功能不足，遺傳篩選診所或出生醫院會通知嬰兒的父母安排嬰兒覆驗，包括驗血及身體檢查。
- ★ 如有需要進一步檢驗嬰兒是否患有先天性甲狀腺功能不足，遺傳篩選診所會轉介嬰兒到出生醫院接受跟進。





如果有其他查詢，歡迎您聯絡我們。

醫學遺傳科遺傳篩選診所
地址：香港九龍九龍灣承昌道一號
香港兒童醫院B座一樓專科門診
電話：5741 4280

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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

Information Leaflet on Newborn Screening



醫院管理局 HOSPITAL AUTHORITY

The Genetic Screening Clinic of Department of Clinical Genetics provides newborn screening service

Target Group

Babies born in Hong Kong public hospitals

Conditions Screened

1 Glucose-6-phosphate Dehydrogenase (G6PD) Deficiency

2 Congenital Hypothyroidism

Reason

- No obvious symptoms at the early stage for both conditions
- Parents should take precaution when taking care of a baby with G6PD Deficiency to prevent the occurrence of any related symptoms
- Early detection and treatment could help to prevent physical and mental impairment in babies with congenital hypothyroidism

Fee

Umbilical cord blood screening tests are free. Follow-up investigation or assessment provided are charged as per the Public Charges of Hospital Authority.

Method

Testing the umbilical cord blood of newborn babies with consent.

Getting the results

If result is **normal**, parents will not be informed

If result is **abnormal**, parents will be informed within **2 weeks**:

1 **Confirmed Glucose-6-phosphate Dehydrogenase (G6PD) Deficiency**

Parents will be contacted by the Genetic Screening Clinic or birthing hospital for counselling.

2 **Congenital Hypothyroidism**

- ★ For suspected Congenital Hypothyroidism: Parents will be contacted by the Genetic Screening Clinic or birthing hospital for reassessment, including blood taking and physical examination.
- ★ For further investigation to confirm any diagnosis of Congenital Hypothyroidism: Baby will be referred by the Genetic Screening Clinic to the birthing hospital for further management.

For further enquires, you may contact us.

Genetic Screening Clinic, Department of Clinical Genetics

Address: Specialist Outpatient Clinic, 1/F, Tower B, Hong Kong Children's Hospital, 1 Shing Cheong Road, Kowloon Bay, Kowloon, Hong Kong

Office Tel : 5741 4280

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