葡萄糖六磷酸去氫酵素缺乏症（又名「蠶豆症」）知多點

電話熱線：2361 9979
（24 小時：提供葡萄糖六磷酸去氫酵素缺乏症資訊的電話聲帶）
網址：http://www.cgs.dh.gov.hk

Genetic Screening Unit
Clinical Genetic Service
Department of Health

此單張是專為患有葡萄糖六磷酸去氫酵素缺乏症孩子的家庭而編寫。雖然此症並無根治方法，但只要孩子採取一些預防措施，他們一般能正常生活。此單張提供有關葡萄糖六磷酸去氫酵素缺乏症的重要資訊及建議。由於篇幅所限，資訊未能盡錄；孩子及其家人可向醫生、撥打電話熱線及瀏覽衛生署網站以查詢更多資訊。

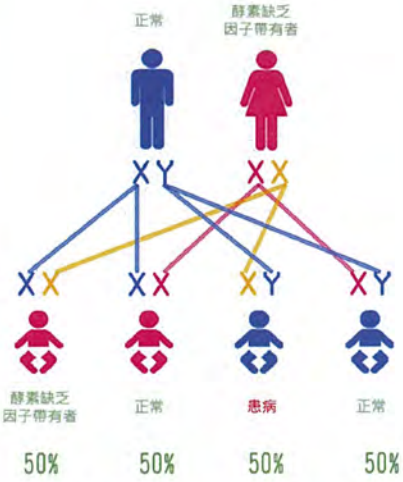
甚麼是葡萄糖六磷酸去氫酵素缺乏症（蠶豆症）？

葡萄糖六磷酸去氫酵素缺乏症為一種遺傳病症，亦常叫蠶豆症。引致此症的基因位於X染色體，所以此症是一個X連鎖的隱性遺傳。X和Y都是決定性別的染色體。女嬰（XX）會遺傳分別來自父親和母親的一條X染色體。而男嬰（XY）則會遺傳母親的X染色體和父親的Y染色體。

男嬰一旦遺傳了帶有引致此症基因的X染色體就會患有此症；而女嬰同時遺傳兩條帶有該基因的X染色體才會患有此症。

下圖顯示其兄弟會有一半機會患有此症；其姊妹則有一半機會成為此症的遺傳基因帶有者（即：沒有表徵但遺傳此症給其子女）。

X連鎖隱性遺傳



與其他遺傳病症一樣，沒有患此症的父母亦可能會將引致此症的基因遺傳予下一代。

葡萄糖六磷酸去氫酵素缺乏症的成因和影響是甚麼？

葡萄糖六磷酸去氫酵素是一種保護紅血球的酵素，協助紅血球維持正常的功能。造成此症的原因是由於這種酵素的份量低於正常水平，繼而引起所謂的「溶血性貧血」。

溶血性貧血是一種血液疾病。當沒有足夠的葡萄糖六磷酸去氫酵素保護紅血球時，大量紅血球會被破壞，溶血性貧血便會發生。而紅血球分解時會導致黃疸，對初生嬰兒來說，這是一個嚴重的情況。若得不到及時的治療，過量的膽紅素（一種可在膽液內找到的黃色色素）會對腦部造成求久的破壞，引致弱智、大腦麻痺、聽覺受損，甚至死亡。

但只要此症患者避免某些藥物（中藥及西藥）、食物、化學品質及避免感染會引起溶血性貧血的疾病，他們一般都能健康地生活。

誰會患有葡萄糖六磷酸去氫酵素缺乏症？

葡萄糖六磷酸去氫酵素缺乏症在東南亞及香港十分常見。男性較女性容易患有此症。在香港，大約每22個男性中有1個（百分之四點五）及每200個女性中就有1個（千分之五）人患有此症。

與其他的遺傳病症相似的是，此症尚未有根治方法。同樣地，此症不會影響患者的整體健康。

葡萄糖六磷酸去氫酵素缺乏症有甚麼症狀？

病發時，患者皮膚和眼白會泛黃。患者也有可能排出深色尿液。**初生嬰兒**會顯得面色癯白，沒有精力和過度眼睏。**小童**或**成人**可能覺得疲倦，呼吸和心跳變得急促。

如發生以上症狀，患者需立即求醫接受治療。

你應該採取以下措施：

- 如你是葡萄糖六磷酸去氫酵素缺乏症
  - o 一旦發現以上任何症狀，應立即求醫。
  - o 求診時，應通知醫生有關患有此症的病歷。
  - o 應隨身攜帶綠卡。

- 如你已懷孕，應通知醫生有關你的家庭遺傳此症的病歷。
- 如你的孩子患有此症，而你決定餵哺母乳，你應避免進食蠶豆，亦應在服用中西藥前向醫生查詢。
- 應養成習慣，查閱食品、家居用品及藥物（中藥和西藥）上的標籤及留意產品的成份。

你應避免：

只要葡萄糖六磷酸去氫酵素缺乏症患者及其家人注意下列事項，患者可以享受健康的正常生活。

- 避免進食蠶豆及其製成品。

- 避免接觸臭丸（萘類）及含有萘的製成品。

- 避免服用未經醫生處方的藥物。
- *避免使用中藥，例如：
  - o 黃蓮
  - o 臘梅花
  - o 金銀花

  - o 牛黃
  - o 珍珠末
    （某些成藥如保嬰丹內含珍珠末）
- *避免使用某些西藥
  - o 某些退熱藥物
  - o 抗生素（例如：呋喃妥英、萘啶酸片、磺胺甲噁唑）
  - o 抗癌疾藥（例如：伯氨喹）
  - o 鎮痙藥（例如：非那吡啶）

*此清單未完全涵蓋所有藥物，請向醫生查詢詳情

如有任何疑問，
請向醫生或藥劑師查詢。

Knowing more about G6PD deficiency (also called favism)

Hotline: 2361 9979
(24 hours; prerecorded tape information about G6PD deficiency provided)
Website: http://www.cgs.dh.gov.hk

Genetic Screening Unit
Clinical Genetic Service
Department of Health

This leaflet is designed for families who have children diagnosed with G6PD deficiency, which is a genetic condition. There is no cure for this condition, but the children can lead as normal a life as possible if certain precautions are taken. This leaflet provides the essential information and advice concerning G6PD deficiency. The information and advice provided here is not exhaustive, so it is important that families contact their doctors or the hotline as well as access the website to find out more.

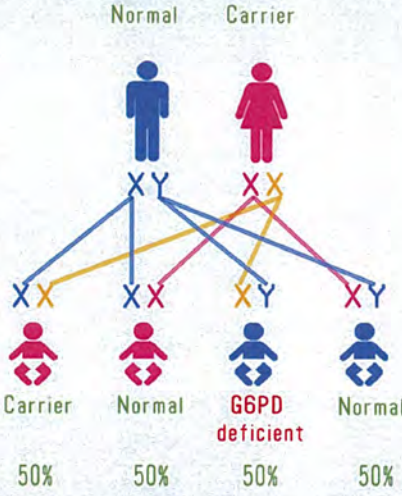
What is G6PD deficiency?

G6PD (Glucose-6-Phosphate Dehydrogenase) deficiency is a genetic condition, also commonly known as favism (after fava beans). The gene causing G6PD deficiency is located on an X chromosome, so it is an X-linked recessive genetic condition. XY are chromosomes that determine the sex of an individual. A girl (XX) will get each X chromosome from her mother and father respectively. A boy (XY) will receive an X chromosome from his mother and a Y chromosome from his father.

A boy will have G6PD deficiency if one X chromosome is defective, while a girl will have the condition only when both X chromosomes are defective.

The figure below shows that a male sibling has a 50% chance to be affected, while a female sibling has a 50% chance to be a carrier (i.e. unaffected but can pass on the condition to their children).

X-linked Recessive Inheritance



It is important to know that while the parents may not be affected by the condition, as in all genetic conditions, the defective gene may be passed to the child from one or both parents.

What are the causes and consequences of G6PD?

G6PD is an enzyme that protects red blood cells and helps them to function normally. G6PD deficiency is therefore the result of having less than usual amount of G6PD in the red blood cells, leading to what is technically called haemolytic anemia.

Hemolytic anemia is a blood disorder in which excessive red blood cells are destroyed because there is not enough G6PD enzyme to protect the red blood cells. The breakdown of the red blood cells leads to jaundice and can be a serious problem as far as newborn babies are concerned. If left untreated, excess bilirubin (a yellowish pigment found in bile) can cause permanent brain damage, leading to neurological complication, which includes mental retardation, cerebral palsy and hearing deficit, or even death.

People with G6PD deficiency enjoy good health as long as they avoid certain medications (Chinese and Western), food and chemical substances, and are not exposed to severe infections which may lead to acute hemolytic anemia.

Who gets affected by G6PD?

This genetic condition is common in South East Asia and in Hong Kong. Males are more likely to be affected by this condition than females. In Hong Kong, about 1 in every 22 males (4.5%) and 1 in every 200 females (0.5%) become affected.

Like all inherited genetic conditions, G6PD deficiency is a life-long condition and currently there is no cure for it. Also, like many genetic conditions, G6PD deficiency does not affect the general health of the affected individuals.

What are the symptoms of G6PD deficiency?

If affected or at risk of G6PD deficiency, the skin and the white part of the eyes will turn yellow. The affected individuals may also pass dark coloured urine. ***Newborn babies*** may look pale and appear lacking energy and excessively sleepy. ***Older children*** or ***adults*** may complain of tiredness, breathlessness and have rapid heartbeats.

If you observe any of the above symptoms, please seek medical advice without delay.

What you should do

- If you are affected or at risk of G6PD deficiency
  - o Consult a doctor without delay when you observe any of the above symptoms.
  - o Inform the doctor about the G6PD diagnosis during consultation.
  - o Always carry the green card.


衛生署醫學遺傳服務遺傳篩選組
Genetic Screening Unit Clinical Genetic Service
Department of Health

葡萄糖六磷酸去氫酵素缺乏症
Glucose-6-phosphate dehydrogenase (G6PD) deficiency

姓名 /NAME 陳大文 / Chan Tai Man 性別 /SEX 男/M
出生日期 /DATE OF BIRTH 01/01/2013

DH 2264 (Revised 2013)
每次見醫生時，請出示此卡
Please present this card when seeing your doctor

SAMPLE

- If you are pregnant, you must inform your doctor about any family history of G6PD deficiency.
- If as a mother you are breastfeeding your G6PD deficient baby, you should avoid eating fava or broad beans yourself and must consult a doctor before taking any Chinese or Western medicine.

What you should avoid

G6PD deficient individuals will enjoy normal growth and health if the following precautions are taken.

- Avoid eating fava or broad beans and their products.

- Avoid close contact with mothballs (Naphthalene) and Naphthalene-containing products.

- Avoid taking any drugs unless prescribed by a doctor

- *Avoid Chinese herbal medicines such as
  - o Rhizoma Coptidis (Huang Lien)

  - o Flos Chimonanthi Praecocis (Leh Mei Hua)

  - o Flos Lonicerae (Kam Ngan Fa)

  - o Calculus Bovis (Niu Huang)
  - o Margaritas (e.g. Counter drugs such as Bo Ying Compound which contains Margaritas, etc.)
- *Avoid Western medicines such as
  - o Certain Antipyretics
  - o Antibiotics (e.g. Nitrofurantoin, Nalidixic acid, Sulfamethoxazole)
  - o Antimalarials (e.g. Primaquine)
  - o Antispamsmodics (e.g. Phenazopyridine)

*This list is not exhaustive. Please consult a doctor.

If you have any further questions or concerns, please contact us or consult your doctor or pharmacist.