Glucose-6-Phosphate Dehydrogenase Deficiency (G6PD Deficiency)
What is Glucose-6-Phosphate Dehydrogenase Deficiency (G6PD deficiency)?

Glucose-6-Phosphate Dehydrogenase (G6PD) is an enzyme which protects the red blood cells and prevents them from being damaged. People with G6PD deficiency can generally enjoy normal health. However, if they suffer from a severe infection or are exposed to oxidant stress such as from certain drugs or chemicals, massive damage of red blood cells may occur, leading to acute hemolysis. Bilirubin will be produced as red blood cells are broken down during acute hemolysis, resulting in jaundice. If newborn babies with severe jaundice do not receive timely treatment, excessive bilirubin may accumulate in the brain, causing irreversible damage to the brain, a condition clinically known as kernicterus. It may result in hearing loss, intellectual disability, spasticity or even death.

How common is G6PD deficiency?

G6PD deficiency is common in Hong Kong. According to the statistical data from the Neonatal Screening Programme, there are 4 to 5 out of 100 male newborns and 3 to 5 out of 1,000 female newborns suffering from G6PD deficiency.

Why the majority of people with G6PD deficiency are male?

G6PD deficiency is a genetic condition with X-linked recessive inheritance.
In human, each cell contains 23 pairs (46 chromosomes) of chromosomes. The 23\textsuperscript{rd} pair are the sex chromosomes. Males have one X and one Y chromosome (XY). Females have two X chromosomes (XX). The gene coding for G6PD enzyme is located on the X chromosome.

As males have only one X chromosome and thus one copy of G6PD gene, they would have G6PD deficiency if the G6PD gene carries a mutation. The G6PD activity in the blood will be reduced.

A female who has a mutant G6PD gene on one of her X chromosomes is a carrier for G6PD deficiency. She usually has a normal copy of G6PD gene on the other X chromosome that
produces sufficient G6PD to protect the red blood cells. Therefore, she may not present any symptoms of G6PD deficiency.

Female carriers for G6PD deficiency have 50% chance to pass on the mutated gene to their next generation. Each son has 50% chance of inheriting the mutant gene and thus suffers from G6PD deficiency. Each daughter has 50% chance of inheriting the mutant gene and thus becomes a carrier of G6PD deficiency.

![X-linked Recessive Inheritance](image)
How do I know if my newborn baby has this disease?

The Department of Health has implemented the Neonatal Screening Programme for all babies born in public hospital since 1984. The programme is free-of-charge and uses cord blood specimen to screen for two common diseases, namely congenital hypothyroidism and G6PD deficiency. To dovetail with the latest development of genomic medicine in Hong Kong, the programme has been transferred to the Clinical Genetics Service Unit of the Hospital Authority from July 1, 2023.

By testing the G6PD enzyme activity in the cord blood specimen, those newborn babies suffering from G6PD deficiency will be identified and diagnosed.

Most of the screening results are normal and parents will not receive special notification. For abnormal results, parents will be notified by medical and nursing staff. Relevant health counselling will be given.

What are the symptoms associated with G6PD deficiency?

People with G6PD deficiency are generally asymptomatic unless they are exposed to oxidant stress such as infection, certain drugs or broad beans.

When there is massive damage of red blood cells causing excessive accumulation of bilirubin that overloads the liver, severe jaundice and even hemolytic crisis may occur. The symptoms of the patient depends on the severity of G6PD enzyme deficiency and the magnitude of the oxidant stress.
Parents should pay extra attention to the presence of jaundice in their babies. They have to contact the Maternal and Child Health Centres, family doctors or private doctors to follow up the health status of their babies.

**What are the symptoms of hemolytic crisis and what should we do?**

Hemolytic crisis occurs when there is massive destruction of red blood cells over a short period of time. As the loss of red blood cells is faster than the production of new red blood cells, acute or severe anaemia will be the result. Clinically, the patient will look pale. In addition, the skin and the white of the eyes turn yellow. They may also pass dark-colored urine. **Parents should observe the following symptoms and seek medical attention without delay:**

<table>
<thead>
<tr>
<th>Newborn baby</th>
<th>Children or adult</th>
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<tbody>
<tr>
<td>• Refuse feeding</td>
<td>• Pale looking</td>
</tr>
<tr>
<td>• Lacking energy</td>
<td>• Complain of tiredness</td>
</tr>
<tr>
<td>• Excessively sleepy</td>
<td>• Breathlessness</td>
</tr>
<tr>
<td></td>
<td>• Rapid heartbeats</td>
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What precautions should people with G6PD deficiency take?

G6PD deficiency is a genetic condition and is lifelong. There is no cure of the disease. It is most important to prevent the occurrence of acute hemolysis induced by oxidant stress. As such, people with G6PD deficiency should take the following precautions lifelong:

1.* Avoid certain Chinese herbal medicines:

<table>
<thead>
<tr>
<th>Rhizoma Coptidis (Huang Lian)</th>
<th>Flos Lonicerae (Jin Yin Hua)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Flos Chimonanthi Praecocis (La Mei Hua)</td>
<td>Calculus Bovis (Niu Huang)</td>
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<tr>
<td>Margaritas (Pearl powder) (e.g. Over-the-counter drugs such as Bo Ying Compound which contains Margaritas)</td>
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2. * Avoid certain western medicines:

- Certain **antipyretics**

- **Antibiotics** such as:
  - Nitrofurantoin
  - Nalidixic acid
  - Sulfamethoxazole

- **Antimalarials** such as:
  - Primaquine

- **Antispasmodics** such as:
  - Phenazopyridine

* This list is not exhaustive. Please consult your doctor for more information.*
3. Avoid eating fava or broad beans and their products:

(Example of assorted beans with broad beans)

(Example of vermicelli made of broad beans)

Pay attention to the food labelling
4. Avoid close contact with mothballs (Naphthalene) and naphthalene-containing products.
5. Inform your doctor or medical staff about your health condition during consultation, or show your G6PD deficiency document / alert card (Remarks: Alert card is available to babies born in hospitals under the Hospital Authority) to your doctor to ensure appropriate prescription.

6. Pregnant ladies should inform their medical and nursing staff about any family history of G6PD Deficiency.

7. Notes to lactating mothers having babies with G6PD deficiency:

- If you are sick and require medication, no matter Chinese or western medicine, please inform your doctor about your baby’s G6PD deficiency for appropriate prescription to you.

- Mothers should avoid eating broad beans and their products.

If the above precautions are taken, the affected people will enjoy normal growth and health.
Enquiry

For enquiries about G6PD and neonatal screening, please call the following enquiry hotline.

Genetic Screening Clinic
Clinical Genetics Service Unit
Hospital Authority

Hotline: (852) 5741 4280
Website: http://www31.ha.org.hk/hkch/Patients/Services/CGSU

Website of Clinical Genetics Service Unit, Hospital Authority