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G6PD Deficiency

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

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.

Favism (G6PD deficiency) is one of the most common inherited enzyme deficiencies in Hong Kong, primarily affecting the stability of red blood cells. 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?

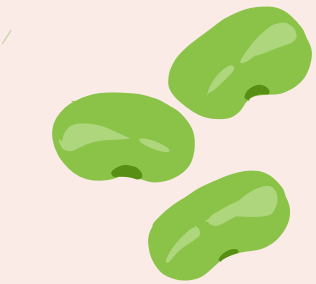
As males have only one X chromosome and thus one copy of G6PD gene, they would have G6PD deficiency if the G6PD gene carries a mutation. The G6PD activity in the blood will be reduced.

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.

Symptoms

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.

Most patients do not exhibit obvious symptoms when not exposed to triggering factors. However, after exposure to triggering factors, the following acute hemolytic symptoms may occur: yellowing of the skin and whites of the eyes (jaundice), and tea-colored urine, among others.



There is no cure for G6PD deficiency. Management focuses on prevention:

- Avoid **fava beans** and related products
- Avoid **medications** that can trigger hemolysis (e.g. certain antibiotics and antimalarials)
- Avoid exposure to naphthalene (e.g. **mothballs**)
- Inform healthcare providers by carrying a G6PD alert card

In severe cases, hospitalization or blood transfusions may be required.

Screening

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.

Useful Link

Hong Kong Children's hospital Clinical Genetics

References:
Hospital Authority
Family Health Service (FHS) of the Department of Health
St. Paul's Hospital

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