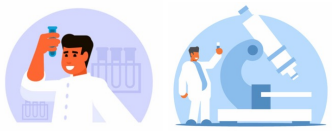
G6PD Deficiency – information for patients and parents

What is G6PD deficiency?

G6PD stands for glucose-6-phosphate dehydrogenase. This is an enzyme which is found in cells of the body and it helps to protect them from damage. G6PD deficiency is an inherited condition which is caused by an error in the gene that provides instructions for the G6PD enzyme. The scientific term for an error in a gene is a mutation. The mutation causes there to be a reduced amount of this enzyme produced. Therefor, red blood cells become damaged more easily and can't work properly. This means these red blood cells get destroyed too early, causing there to be a shortage of them in the blood.



How and when is G6PD deficiency diagnosed?

G6PD deficiency can be diagnosed with a simple blood test. The blood sample is then looked at under a microscope. Doctors can then assess the shape of the red blood cells and also do some chemical tests which will show if the patient has G6PD deficiency or not.

There is no specific age at which G6PD deficiency is diagnosed. Some people with this condition can be asymptomatic (have no symptoms) until adulthood, however others may be diagnosed just after birth.

Who is most likely to be affected by G6PD deficiency?

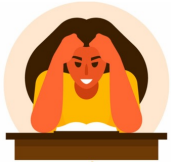
G6PD deficiency occurs most often in biological males. This is because it is an X-linked condition. This means the gene responsible for the condition is found on the X chromosome. Biological females have two X chromosomes whereas males only have one. This means that if there is a mutation in the X chromosome in females, there is another unaffected X chromosome which could be used. If the X chromosome is affected in males, there is no alternative X chromosome available, meaning that they will present with G6PD deficiency.

G6PD is more common in tropical or subtropical areas and also within people that have African, Mediterranean or Asian decent. This is thought to be because G6PD deficiency has been shown to have protective properties against malaria, however the reason for this is still being investigated.

What are the symptoms of G6PD deficiency?

The main symptom of G6PD deficiency is haemolytic anaemia. Haemolysis is the name for the process of breaking down red blood cells, and anaemia is when there is not enough red blood cells in the blood. Haemolytic anaemia is anaemia caused by red blood cells being destroyed too quickly.

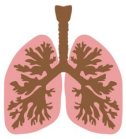
Haemolytic anaemia can cause patients to be fatigued, lacking energy and weak. Their skin might be jaundiced (a yellow colour) and they may experience confusion, dizziness and a decreased ability to concentrate.



Some more possible symptoms of G6PD deficiency include:

- Dark coloured urine
- Pale skin – For patients who have darker skin, paleness may be seen best in the mouth, especially on the lips or the tongue.

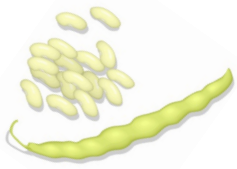
- Lower back pain
- Englarged spleen – The spleen is an organ in the abdomen which has a role of breaking down red blood cells. When someone is experiencing haemolytic anaemia, the spleen is working extra hard, making it become larger. This can sometimes cause abdominal pain.



- Breathlessness or a fast heart rate

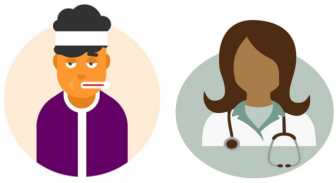
There are also some factors which can cause the symptoms of G6PD deficiency to become worse. These triggers include certain drugs, for example aspirin and some antibiotics, or certain infectious diseases like hepatitis and pneumonia. It's a good idea to ensure your doctor is aware of your/your child's condition before they prescribe any medications.

The most common trigger for G6PD deficiency is eating fava/broad beans.



In people with a reduced amount of the G6PD enzymes, fava/broad beans can cause damage to the walls of red blood cells. Therefor, these people will experience a reaction if they eat these beans. Their red blood cells will be destroyed more quickly, causing severe haemolytic anaemia. It's important to carefully check the ingredients on food packages, as it is not always clearly labelled if fava/broad beans are included. The worsened symptoms usually start to happen about 2-3 days after the exposure to a trigger, but it can sometimes be sooner.

A serious complication of G6PD deficienicy is haemolytic crisis. This is when a large amount of red blood cells are destroyed in a short amount of time. It can happen as a result of exposure to a trigger. It's important to seek professional help quickly if you think you are experiencing this, as it can be very dangerous. Some of the symptoms of haemolytic crisis include:



- A sudden rise in body temperature
- Heavy and fast breathing
- Weak and rapid pulse
- Feeling generally unwell
- Skin is more pale or jaundiced than normal

What is the treatment for G6PD deficiency?

G6PD deficiency is a genetic condition so there is no cure, however there are ways to improve the symptoms. The first way to do this is by removing the triggers which are worsening the symptoms, for example, stopping specific medications or treating an infection. It's important to discuss this with your doctor first, before making any changes.

Patients can also be treated with supportive care if they are experiencing severe anaemia. This includes providing the patient with IV fluids or oxygen whilst they are in hospital, to make sure they are maintaining their basic needs whilst they are recovering.

Some more treatment options

Blood transfusions can also be used to improve the symptoms of severe anaemia. Transfusions increase the number of healthy red blood cells which are circulating in the blood, helping to relieve symptoms like tiredness and a lack of energy.

Doctors may also prescribe a supplement called folic acid. This is a vitamin which is needed to produce red blood cells and therefor will help to keep up with the high demand for new blood cells to be made.

What can I do to help myself/my child with G6PD deficiency?

It's important to avoid the triggers of G6PD whenever possible. As we mentioned before, it's not always obvious when food products contain fava/broad beans so it's important to be vigilant when you are reading the ingredients.

Maintaining general health is a good way to reduce the chances of infections which can trigger G6PD deficiency, whilst also helping to sustain your overall well being. This can be done by staying hydrated and having a balanced diet, along with regular exercise and physical activities. Also, try to get enough rest and keep your stress levels low. These measures will all contribute to a healthy immune system, which is more protective against infections and illness.

Having G6PD deficiency does not mean giving up on living a full and active life.

If you smoke tobacco, it's a good idea to try quitting. Smoking can weaken your immune system which can make you more vulnerable to infections. It also increases the number of free radicals that can cause more damage to red blood cells. If you drink alcohol, it's important to avoid drinking too much as this can also damage red blood cells.

If your child has G6PD deficiency, you may notice they get more tired than other children their age. For example, they may need a nap after school, they might get more worn out in physical activities or struggle with their concentration in lessons. It could be a good idea to make their teachers aware of your child's condition, so they can have breaks or extra help if they need it.

Can we help?

CAN (congenital anaemia network) is a charity specifically for patients with inherited anaemias. It was started by a group of patients and doctors, working together to tackle some of the problems of having an inherited anaemia- the lack of information and the loneliness of not knowing anyone else with your condition. We run events for patients to meet each other, as well as provide some additional information, support and services for patients who need that extra bit of help.

Yes we can

CONGENITAL ANAEMIA NETWORK

If you want to know more visit www.togetherwecan.uk or email us on info@togetherwecan.uk