

What do you need to know if you have a baby/child diagnosed with PKD?



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Pyruvate kinase deficiency, also known as PKD, is a rare genetic condition in which there is a reduced amount of the pyruvate kinase enzyme in red blood cells.

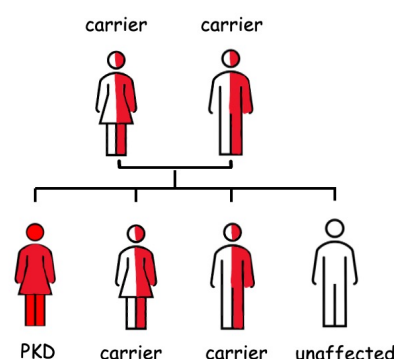
How does PKD affect red blood cells?

Normally, this enzyme helps to turn sugar into energy for red blood cells to use. If there is a deficiency in the pyruvate kinase enzyme it means there will be a reduced amount of energy, which causes the red blood cells to break down prematurely. Instead of the red blood cells surviving for 120 days like usual, in PKD they tend to be destroyed after a few days or weeks.

Why is there a reduced amount of pyruvate kinase enzyme?

There is a reduced amount of pyruvate kinase because there is a change in a gene which provides the instruction for this enzyme. This change is called a mutation. Many different gene mutations can cause PKD, however the most common one is a mutation in a gene called PKLR.

PKD is an autosomal recessive disorder. This means that you must inherit a mutated gene from both of your parents to develop this condition.



What are the symptoms of PKD?

PKD can cause a variety of symptoms which can range from mild to severe.

PKD patients often present with a type of anaemia, called haemolytic anaemia. Anaemia is a general term meaning that there is a reduced amount of red blood cells circulating in your blood, and haemolysis (or haemolytic) means that red cells are dying faster than they usually do. Many symptoms of PKD are due to the anaemia which is caused, for example:

- Fast heart beat and breathing
- Weakness
- Dizziness
- Paleness
- Slower growth
- You may notice your child not being able keep up with their friends during games or exercise



Some more possible symptoms

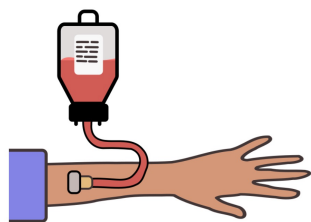
Other symptoms of PKD can include the spleen (an organ in the tummy which breaks down red blood cells) being enlarged and dark coloured urine. There may also be a build up of iron in the body; this is because the body absorbs more iron from food when someone is anaemic.

In severe cases of PKD there can also be more serious complications, for example gallstones or even kidney failure.

Jaundice is also a common presentation for children with PKD. This is when their skin and the whites of their eyes become a yellow colour. Jaundice is caused by a build up of a pigmented substance called bilirubin, which is a break down product from red blood cells. Jaundice can be mild to severe, and it can also vary from time to time. You may notice your child becomes more jaundiced (they become a more yellow colour) if they are ill.

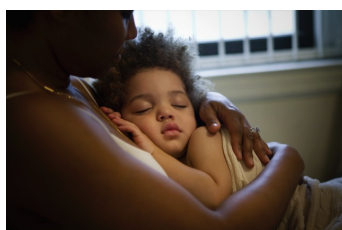
What are the treatments for PKD?

Children with mild cases of PKD often don't require treatment. However, treatment options for those with moderate to severe symptoms may include:



- Blood transfusions help to increase the number of red blood cells in your blood. It's possible that if you require regular blood transfusions as a child, you may no longer need them as an adult. However, some people with severe cases of PKD might require lifelong blood transfusions.

- Folic acid and vitamin supplements to help with your anaemia. Folic acid is needed to produce new red blood cells so important to ensure your body gets enough.
- UV light therapy can reduce jaundice in babies.
- Iron chelation is a type of therapy where medication used to reduce the amount of iron which is stored in the body.
- Sometimes a surgeon will partially or fully remove the spleen, this is called a splenectomy. This is done if your spleen has become too enlarged, due to PKD.
- There is a drug called mitapivat, which is showing positive effects on PKD patients in USA and the EU. This drug has not yet been authorised in the UK, but hopefully it will be ready for use in the near future.



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How will this condition impact my child's daily life?

This depends on the severity of your child's PKD and their symptoms. You may find that your child is more tired in comparison to other children, for example they may need to nap when they get home from school or activities. Sometimes children may also struggle with PE lessons. Exercise is fine for children with PKD however it's important to listen to how they feel. It might be good to make the PE teachers aware of your child's condition so that they can have breaks if they are tired/feel like it's too much.

Making sure that your child stays hydrated and has a balanced diet is also important.

Children with PKD can take a longer time to recover from illness than other children. Again, this is individual to each person but it's not uncommon for children with PKD to need longer breaks from school to recover.

Parvovirus is one illness in particular which may affect all people with haemolytic anaemias more seriously. This virus can stop the production of red blood cells and therefore send the patient into haemolytic crisis - patients will often require a blood transfusion. After you have had this virus once you will be immune to catching it again, but it's important to watch out for the symptoms. These include lethargy, loss of appetite, abdominal pain and bloating, fever, vomiting and diarrhea (which is often bloody).

Some questions you may want to ask a doctor if your child is diagnosed with PKD

Will my child need treatment?

How often will my child need treatment?

Which medical professionals will I work with to help manage my child's condition?

What symptoms should I watch out for?

How often will my child need testing to check their red blood cells?



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If you have any questions please contact CAN at info@togetherwecan.uk