Diamond Blackfan Anaemia

What is Diamond Blackfan Anaemia?

It is an abnormality of red blood cells that is present at birth and often diagnosed before the child is one. Symptoms develop because the numbers of red blood cells made in the bone marrow are lower than usual. It is not understood why this happens, however research is being carried out into this condition in many countries around the world.

What are the signs and symptoms?

Symptoms are related to a child having a decreased amount of oxygen in their blood due to low numbers of red blood cells. Symptoms vary a lot but can include:

- Paleness
- Tiredness
- Weakness
- Irritability
- Poor appetite or failing to feed properly
- Poor growth
- Headaches

Some children have associated heart abnormalities and can develop an irregular heartbeat. A common trait found in children is an unusual facial appearance such as a snub nose, widely spaced eyes and a thick upper lip. Other physical differences may include the neck being shorter than usual, webbed and unable to move due to the bones in the neck being fused. The shoulder blades may stick out more than usual and hands may not be formed properly, i.e. abnormal thumbs.

How is Diamond Blackfan Anaemia diagnosed?

Diagnosis is based on symptoms i.e. a baby may be thought to have anaemia if pale and short of breath when feeding.
• A simple blood test is taken and usually reveals decreased red blood cells, low haemoglobin and high levels of adenosine deaminase (ADA), a particular red cell enzyme.

• Bone marrow biopsy – reveals not many new red blood cells are being made (please refer to section 4 “Procedures” for further information on this test).

How is Diamond Blackfan anaemia treated and what are the side effects?

Steroids - The usual form of treatment is with a steroid called Prednisone. It helps stimulate more red blood cells to be made. This treatment should start as early as possible. Dexamethasone is another steroid often used. There are pamphlets available which provide detailed information including side effects. Please ask medical or nursing staff.

Blood transfusions - are needed regularly to increase the number of red blood cells circulating in the body. More information including possible side effects can be found in section 3 “Information on Blood”.

Having multiple blood transfusions causes a build up of iron in the tissues of the body particularly the heart and liver. A drug called Desferal (Deferoxamine) is given to help reduce iron levels. This is known as chelation therapy. Desferal combines with iron in the bloodstream and is then removed from the body by the kidneys. By removing the excess iron, desferal lowers possible damage to the organs and tissues of the body.

Side effects of this drug can include: Bluish nails, lips, or skin; changes in vision, seizures, changes in breathing, rapid heartbeat; hearing problems and skin rash, hives, or itching.

The colour of your child’s urine may change to orange whilst taking Desferal.

Bone marrow transplant - This works by replacing the cells in the bone marrow with healthy cells so they can produce red blood cells. This is generally used for patients who cannot tolerate any other form of treatment but even BMT doesn’t always work.

(A Booklet on Stem cell transplant will be given if required).
Fortunately, a quarter (20%) of children recover spontaneously from this condition.

It is important to remember that you and your family will adjust to living with a chronic illness and can still lead a normal life.

References and More Information available at:

www.rarediseases.about.com

www.rarediseases.org/search/

Please refer to section 7 “Sources of More Information” or check the parent's library in McGuinness & McDermott children's clinic for further information.