

What is Diamond Blackfan Anaemia?

Diamond Blackfan Anaemia (DBA) is a blood condition caused by a failure within the bone marrow. It is characterized by an inability to produce red blood cells (necessary to transport oxygen around the body). It is typically diagnosed before the patient's 2nd birthday with the majority of cases found before 4 months. The condition is extremely rare, affecting approximately 125 people in the UK and less than 1000 world-wide. It requires intensive therapy, and has no known cure. DBA has a range of symptoms, from those needing little or no treatment, through steroid responsive patients to those dependent on transfusions.

What causes DBA?

Little is known about the condition because it is so rare. Research has shown a genetic link in 25% of cases relating to the RPS19 gene and continues to try to find other links. In many cases however, it occurs sporadically.

What are the symptoms of DBA?

DBA has symptoms in common with all types of anaemia. Pallor (paleness), tachycardia (irregular heartbeat) and heart murmurs due to the increase in work the heart needs to do to keep oxygen moving around the body. This can lead to irritability, tiredness, and fainting. Since DBA is diagnosed at a very early age, it is difficult for children to tell their parents how they are feeling. Parents usually notice that 'something is wrong' well before diagnosis. In a third of cases there are related physical defects, including thumb, kidney and facial abnormalities, but in others there are no physical signs that the sufferer has DBA. Many affected children are very short for their age, and may have delayed puberty.

What Treatments are available?

Treatment currently consists of two main therapies-

- Steroid therapy

Around 70% of DBA-diagnosed patients respond initially to steroid therapy (usually prednisone) although response has been known to fail spontaneously. Almost all of those that do respond must continue to take steroids for the rest of their lives. Long term use of this therapy can result in side-effects which include stunting of growth, water retention, bone weakening, diabetes, reduced immunity, glaucoma, cataracts and high blood pressure. In order to minimise these effects the steroid dose is usually tapered until the lowest workable level can be found.

- Transfusion therapy

Those that do not respond to steroids, or need too high a dosage to sustain red cell count, require regular transfusions. The main side-effect of this treatment is iron overload. The body normally recycles iron from old red blood cells, storing it in the liver, heart and pancreas for reuse. DBA sufferers also store recycled iron, but as it is never reused it can lead to a build up, causing a poisoning of the system which will require regular chelation therapy to reduce the amount of iron. Iron retention can lead to heart and liver problems, as well as diabetes and growth problems.

What about Bone Marrow Transplants?

A bone marrow transplant can lead to normal red cell production, but this comes with its own challenges as matching bone marrow donors are hard to find. Stem cells taken from the umbilical cord of an unaffected sibling (with same parents) can also be used for this procedure.

Is there a cure?

Rarely, some individuals will have a spontaneous remission, in which the red cell production switches back on, but this is uncommon, and is impossible to predict.

Is there any research into finding a cure?

Research into the cause of DBA and other related bone marrow failures is being carried out at several centres internationally. In the UK this takes place at St. Mary's Hospital, London.

Pharmaceutical research is practically non-existent, due to the small number of DBA sufferers world-wide. However, pharmaceutical research into related conditions may lead to better treatment for DBA patients, especially related to transfusions and chelation therapy.

The following technical information may be of interest to Medical Professionals.

DBA usually presents within the first two years of life. Initial bloods are taken to test for low haemoglobin and high Adenosine Deaminase Activity (ADA) in the blood. This is not definitive, but points to DBA as a likely cause. Bone marrow biopsies can confirm that the erythroid (red cell) precursors are depleted, which can point to a viral infection (Parvovirus B19) or DBA. Doctors may also check the reticulocyte count. High reticulocyte counts point to conditions where blood is being created normally but is being destroyed. Low counts indicate a lack of red cell production. In DBA, reticulocyte counts are typically low, and can be zero. DBA sufferers usually have a very high MCV (Mean Cell Volume) so tend to have larger red blood cells, and a higher percentage of foetal haemoglobin (Hb F) than normal, all which can be tested through bloods and bone marrow biopsies.



DBA-UK

DBA-UK is the UK based patient support group for the condition Diamond Blackfan Anaemia (DBA).

With only approx. 125 known cases in the UK this rare condition leaves parents and patients feeling very isolated, as many of the Health Service staff they come into contact with will have never heard of the condition, let alone be aware of its treatment and prognosis.

The support group is made up of parents, family and friends of patients with DBA, along with the patients themselves. The charity aims to provide support for these families by bringing them together to provide mutual help. Sometimes just talking to someone who understands can be a relief in itself.

If your life has been personally affected by DBA, we have a UK support phone line which will take you through to Jane who kindly volunteers in this role. Jane has DBA and is also the mother of a DBA child.

**UK Patient and Family
support phone line:
0845 094 1548**

Support, Research and Hope: DBA UK

At DBA UK we aim to deliver support, research and hope to the DBA community by bringing families together to share their experiences, communicate the latest medical information and raise funds to support each other.

With your help, our website, information leaflets and telephone support line will provide invaluable support to patients and their families at a difficult and confusing time. We also hold an annual conference for all DBA families in the UK. The weekend provides respite and mutual support for patients and brings parents and carers together with medical professionals to improve their access to information and aid the consistency and quality of care across the country.

We hold the event at a PGL activity centre where children with DBA can meet and gain confidence and reassurance from sharing their experiences, away from the daily grind of their treatment routine. Thanks to your support, we will treat them to a weekend of adventure in a happy and inclusive camp atmosphere.

To find out how you can donate to DBA-UK please visit

www.diamondblackfan.org.uk/how-to-donate-donate-now/

If you're a medical professional, potential sponsor, journalist or other interested party and would like to get in touch with the charity please email the DBA UK secretary –

information@diamondblackfan.org.uk

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A guide for patients, families and
healthcare professionals.

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